



Informed Consent and Trisomy Screening: Delineating Parent and Professional Interests

Emyr Owain Wile

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Abstract

The landmark decision of *Montgomery* established that patients' right to self-determination and autonomy underpins the doctrine of informed consent. However, a growing body of medical research routinely conclude that consent for trisomy screening is less than informed.

Consent for trisomy screening is not a 'one-off event': it is a multistage and multifaceted process, requiring the involvement and integration of interprofessional practices across the pathway. However, *Mordel* exposed systemic frailties and disconnects in terms of the processes for securing parent consent for trisomy screening: a dimension often missed by medico-legal studies in this field.

With the recent introduction of non-invasive prenatal testing (NIPT) and additional trisomies (Edwards' and Patau's Syndrome) to the traditional Down's Syndrome screening programme, this has exacerbated existing concerns around parent decision-making and consent for screening. Using empirical methods, this study seeks to delineate parent and professional interests for providing and securing consent for trisomy screening.

Introduction

Public Health England and Wales executed the recommendation of the UK National Screening Committee (UK NSC) to implement a reformed 'trisomy' screening pathway to existing antenatal screening programmes in 2016 and 2018, respectively.

Traditionally, only Down's Syndrome (DS) screening was offered to parents across England and Wales as an optional component to their antenatal care. The implementation of the 'trisomy' pathway saw the introduction of Edwards' (ES) and Patau's Syndromes (PS) to the traditional DS, providing parents with the choice to screen for the 'trisomies' under the same care pathway.

DS is the most common trisomy, with ES and PS being the second and third most common. The UK NSC's rationale for offering 'trisomy' screening was to provide parents with an opportunity to detect the common trisomies antenatally, enhancing reproductive choice and autonomy. In England and Wales, under the 'trisomy' pathway, parents have the opportunity to screen for DS only, ES and PS only, or all three conditions together.

The UK NSC's recommendation to implement the pathway was also accompanied by the introduction of a new method of testing, that had already existed under the private market. This new method of testing, or non-invasive prenatal testing (NIPT), was recommended as a 'safer' and more accurate method of testing for the common trisomies, circumventing the small but significant procedural risk of miscarriage associated with existing invasive methods of testing. The UK NSC's justification for recommending the addition of NIPT, as a method of screening, was to reduce the number of invasive tests being conducted on higher-risk parents.

While the UK NSC's objectives are primarily to enhance reproductive autonomy and choice, a growing body of research has underlined that consent for trisomy screening is less than informed.¹ Studies revealed that, under the traditional DS screening pathway, that parents were not providing valid consent, due to several key concerns; commonly, a lack of accurate

¹ Maria Tsouroufli, 'Routinisation and constraints on informed choice in a one-stop clinic offering first trimester chromosomal antenatal screening for Down's syndrome', (2011) 27 *Midwifery* 431, 436; and, Mollie A. Minear, Stephanie Alessi, Megan Allyse, Marsha Michie and Subhashini Chandrasekharan, 'Noninvasive Prenatal Genetic Testing: Current and Emerging Ethical, Legal, and Social Issues', (2015) 16 *Annu. Rev. Genomics Hum. Genet* 369, 373; and, Antina de Jong, Wybo J Dondorp and others, 'Non-invasive prenatal testing: ethical issues explored', (2010) 18 *European Journal of Human Genetics* 272, 277.

and balanced information and its perception as a 'routine' component of parents' antenatal care.²

With the implementation of the reformed trisomy pathway, and the introduction of NIPT to screening pathways, it was foreseeable that existing concerns for parent consent will only be further exacerbated in this regard. Indeed, while the 'trisomy' pathway was constructed to enhance reproductive autonomy and practical efficiency in screening, the conditions are very different in terms of their aetiology, pathogenesis and prognosis. In light of existing concerns for a lack of balanced and accurate information, it was likely that the presentation of trisomy screening, as a genetic model, could mislead parents and professionals into believing that decision-making and choice for the trisomies carry the same consequences; a concern forewarned by the UK NSC, in 2014.

Furthermore, early medical trials identified that NIPT was being fundamentally misunderstood among parent groups and sought to highlight this concern before its implementation to NHS screening programmes. Studies revealed that parents have an insufficient understanding of NIPT, in terms of its purpose and ability to fulfil patient expectation. Parents believed that NIPT replaced the need for invasive testing due to its reported '99%' accuracy, demonstrating a complete misunderstanding of the technology, hindering parent choice and decision-making. Academics have long questioned the compatibility of consent, informed decision-making and reproductive autonomy, in light of such developments.

Montgomery v Lanarkshire Health Board, in 2015, sought to promote and safeguard patient autonomy and self-determination under a shared decision-making model of care, and provided the doctrine of informed consent with a legal footing.³ The doctrine is said to be the antithesis of paternalism, a force which dominated the law of consent and information disclosure since the decision in *Bolam v Frierns Hospital Management Committee (1957)*.⁴

² García, E., Timmermans, D.R.M. & van Leeuwen, E, 'The impact of ethical beliefs on decisions about prenatal screening tests: Searching for justification', (2008) 66 *Social science & medicine* 753, 764; and, C. Lewis, C. Silcock and L.S. Chitty, 'Non-Invasive Prenatal Testing for DS: Pregnant Women's Views and Likely Uptake', (2013) 16 *Public Health Genomics*, 223, 230; and, Caroline Silcock and others, 'Will the Introduction of Non-invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?', (2015) 18 *International Journal of Public Participation in Health Care and Health Policy* 1658, 1660.

³ *Montgomery v Lanarkshire Health Board (2015) UKSC 11*.

⁴ *Bolam v Friern Hospital Management Committee (1957) 1 W.L.R.*

Under the historic ‘culture of paternalism’, the interests of patients and professionals were not considered in equal measure when assessing professional duty and commitment to consent and information disclosure. While patient autonomy was insufficiently protected at this time, a rights-based narrative began to emerge following the introduction of the Human Rights Act 1998. Despite the Act only being recognised as symbolic of patient rights’ by scholars at this time, it nevertheless subsequently proved to be highly disruptive and influential on the manner in which the judiciary approach the assessment of professional duty for information disclosure and consent.

Montgomery alluded to the significance of a continued dialogue between professional and patient, recognising that consent was dynamic in nature. Indeed, to treat consent as a ‘one-off’ event was becoming increasingly outdated and formed a simplistic view of the process. The importance of understanding consent as a continuing process between professional and patient was brought to light in the recent decision of *Mordel v Royal Berkshire NHS Trust*, in 2019.⁵

While Jay J, in *Mordel*, recognised the importance of *Montgomery*, in terms of providing sufficient information to patients before securing consent, and that patients were bearers of ‘rights’ in this regard, he also assessed the *Bolitho* ‘reasonableness’ of established systems for securing consent along the DS screening pathway. Due to a lack of interprofessional collaboration and communication within the maternity unit – ultimately between the sonographer and midwife – the case exposed existing frailties and disconnects with the process for securing parent consent for trisomy screening.

In light of the introduction of ES and PS to the DS pathway, compounded by the considerations raised in *Montgomery* and *Mordel*, an opportunity arises to conduct an empirical response as a means to delineate whether the plural interests of both the profession and parents are being equally valued, in the context of consenting to trisomy screening and testing. By employing a range of qualitative and quantitative techniques, the aim of this thesis is to initiate a dialogue between stakeholders, with the purpose of identifying and addressing the relationship between parent and professional interests and values for delivering and securing consent along the trisomy pathway.

⁵ *Mordel v Royal Berkshire NHS Trust (2019) EWHC 2591.*

Chapter 1 will map the developments of informed consent, in terms of its gradual integration and recognition by the British judiciary, beginning with the prominence of medical paternalism in *Bolam*, to the emergence of patients' rights in *Montgomery*. It will also map the systemic and procedural considerations for obtaining parent consent for trisomy screening and testing, outlined in *Mordel*. The chapter will conclude by foregrounding and framing an empirical response to the questions and key areas of consideration raised by *Montgomery* and *Mordel* to the issue of delivering and obtaining consent for trisomy screening and testing.

Chapter 2 will conduct a review of the existing literature and clinical guidelines on informed consent for trisomy screening. This will include providing an outline of the historic developments of antenatal and trisomy screening in England and Wales. It will also underline key themes that emerge from the literature and clinical guidelines on consent as a foundation for further empirical exploration.

Chapter 3 will outline the methodology and methods used for the purposes of collecting empirical data in this thesis. It will outline the researcher's ontological and epistemological assumptions for the purpose of constructing an appropriate research paradigm. The implications of COVID-19 on the intended research methods will also be discussed.

Chapter 4 will present the quantitative data collected from the parent and professional research populations to identify initial patterns and themes for further qualitative exploration.

Chapter 5 will present the qualitative data collected from the parent research populations, mapping the themes and subthemes for later discussion.

Chapter 6 will present the qualitative data collected from the professional research populations, mapping the themes and subthemes for later discussion.

Chapter 7 will discuss the research findings from both the parent and professional studies. It will evaluate the findings in light of existing literature and clinical guidelines on trisomy screening, subject to key case law, for the purpose of delineating professional and parent interests for securing consent along the pathway.

Chapter 1 – Informed Consent and Information Disclosure

This chapter will outline the historical development of the law of consent and information disclosure, from *Bolam* to the landmark cases of *Montgomery* and *Mordel*. Section 1.1 begins by outlining the development of case law in the development of informed consent, exploring the prominence of medical paternalism and the growing consciousness of the UK courts of the growing rights-based narrative, with the formation of the Human Rights Act, and trans-jurisdictional influences. Section 1.2 and 1.3 will explicitly outline and discuss the landmark cases of *Montgomery* and *Mordel*, framing an initial empirical response to the key considerations and areas of particular interest raised in these cases, specifically pertaining to the interests of parents and professionals when delivering and securing consent for trisomy screening.

1.1 The Historical Developments of Informed Consent

1.1.1 Establishing the Legal Framework

Historically, there was no legal duty on doctors to inform their patients with information, regarding the risks and benefits, or alternative options, associated with proposed treatments or their prognosis, as there is today.⁶ Typically, the patient's wishes were subservient to the interests of medical practitioners, in pre-and post-treatment care management. The historical battle between the interests of patients and professionals, has undoubtedly moulded the opaque conception of informed consent that exists today.⁷

The tautologous concept of 'informed' consent developed as a US common law doctrine, and has not always been germane to the socio-political landscape of Britain. Informed consent is synonymous with the autonomous patient and the right to self-determination for treatment and care. In contemporary Britain, obtaining valid consent is imperative for medical care, both legally and ethically.⁸

⁶ Emily Jackson, *Medical Law Texts, Cases and Materials*, (2nd edn, OUP 2010) 167.

⁷ Nils Hoppe and Jose Miola, *Medical Law and Medical Ethics*, (1st edn, Cambridge University Press 2014) 75-84.

⁸ Robert Timko, *Clinical ethics: Due Care and the Principle of Nonmaleficence*, (1st edn, University Press of America 2001), 1-194.

In broad terms, if a HCP were to proceed with a procedure without obtaining the patient's valid consent, this could amount to either battery or negligence, under a civil action. In very rare circumstances, this could amount to criminal liability. This is very rare in the context of healthcare, as the element of intention, or *mens rea*, must be satisfied to establish criminal liability. For this reason, civil action is deemed to be the more appropriate framework to use where a breach of duty to obtain valid consent has taken place, in the context of healthcare.

Battery

Under civil law, and in the context of medical treatment, if non-consensual direct contact with the Claimant's body occurs during the course of performing a medical procedure or intervention, without lawful excuse or justification, this could result to liability for battery.⁹ The tort of battery is a civil action, which requires a direct and intentional application of force to another person, without consent.¹⁰ Therefore, a justification for the unpermitted contact is to establish valid or informed consent.¹¹ The case of *Airedale* illustrated that consent is not an excuse to the unpermitted contact, but rather a justification:

'... why the consent of the patient is important is not that it furnishes a defence in itself, but because it is usually essential to the propriety of medical treatment'.¹²

The intentional torts are actionable *per se*, meaning that the Claimant can bring a civil action in battery, without having to prove that they suffered any damage, as a result.¹³ This means, under the framework of battery, it is not necessary for the Claimant to demonstrate damage has occurred by relying upon expert evidence, nor by reference to a professional medical opinion. Consequently, the threshold for establishing causation is significantly reduced – as the Claimant only needs to prove that the Defendant intentionally touched them in the absence of permission – as opposed to demonstrating they had not been adequately

⁹ Necessity can provide a defence in some emergency situations. If non-consensual force is used, the defence of necessity will apply where the force is the minimum necessary to preserve life. See *Leigh v Glandstone* (1909) 26 TLR 130 and *Re T* (1992).

¹⁰ *Collins v Wilcock* (1984) 3 All ER 374, Lord Goff defined battery as, "a battery is the actual infliction of unlawful force on another person", at 1177. See also, *Faulkner v Talbot* (1981) 3 All ER 468, Lord Lane at para 471.

¹¹ The defence of consent is not always a justification, see *R v Brown* (1993) 2 WLR 556.

¹² *Airedale NHS Trust v Bland* (1993) AC 789, 881.

¹³ *DPP v Little* (1992) 1 All ER 299.

informed to make an informed decision. The rationale of the courts, for this lenient requirement, is justified to preserve one's right to self-determination and bodily integrity.¹⁴

Consent operates to protect patient autonomy for medical treatment and healthcare. In practice, this involves expressly or impliedly providing consent, typically both verbally and in written form, evidenced by paper or electronic consent forms. In the case of surgical interventions (involving cutting), it is commonplace for consent to be rigorously documented, often requiring the patient to verbally express their consent, in addition to completing a written form as further evidence (signing a consent form). For non-surgical interventions (involving scans or non-invasive procedures), the patient is only required to deliver implicit or verbal consent, which is documented by the healthcare professional, in either electronic or paper form. However, it is important to note that consent forms will only provide evidence that the patient has consented to the course of treatment, as opposed to establishing that the patient has been adequately informed, or provided valid consent.¹⁵ To the contrary, where the patient has not signed a consent form, it would be incorrect to presume that consent has not been provided by the patient.¹⁶

McLean submits that there are unique advantages for the patient in pursuing a claim for battery, as it "establishes an uncompromising baseline for the protection for patients' self-determination".¹⁷ Furthermore, as there need not be any proof of harm caused by the act, the focus rests on the patient's autonomy or bodily integrity.¹⁸ One could also argue that an action in battery is favourable to the Claimant, in terms of its lenient requirements needed to establish causation.

However, pursuing a claim for battery becomes less advantageous in the context of non-surgical interventions, or in other words, where no touching has occurred (scans, distribution of therapeutic drugs, etc). An action in battery is incapable of safeguarding the patient's right

¹⁴ Harvey Teff, *Consent to Medical Procedures: Paternalism, Self-Determination or Therapeutic Alliance?* (1985) 101, *Law Quarterly Review*, 432, 436.

¹⁵ Lauren Sutherland QC, *A Guide to Consent in Clinical Negligence Post-Montgomery*, (1st edn, Law Brief Publishing 2018) 72.

¹⁶ *Chatterton v Gerson* (1981) 1 QB, "*getting the patient to sign a pro forma expressing consent to undergo the operation ... should be a valuable reminder to everyone of the need for explanation and consent. But it would be no defence to an action based on trespass to the person if no explanation had in fact been given. The consent would have been expressed in form only, not in reality*", Bristow LJ, at para 432.

¹⁷ Sheila A.M. McLean, *Autonomy, Consent and the Law*, (1st edn, Taylor & Francis Group 2009) 71.

¹⁸ *Ibid.*

to make decisions or to receive no treatment, and also to compensate those aggrieved patients who were unable to consider alternative therapeutic options, as a result of the doctor's failure to disclose them.¹⁹ The inappropriateness of pursuing a claim for battery, against a healthcare professional, also extends to policy considerations. The aim of doctors is to benefit the patient by offering treatment, or in other words, to adhere to the principle of beneficence; therefore, it is not the intentions of the court, nor is it desirable, to frame doctors in this way.

Negligence is considered the appropriate action, as opposed to battery, where an interference had taken place with patients' right-to-know. This submission has been reinforced by the case of *Chatterton v Gerson*.²⁰ As Mclean states, the standard of information disclosure, required under the framework of negligence, is more lenient than that required under battery, making it more difficult for Claimants to succeed in pursuing their action by proving their allegations to be true.²¹

Negligence

Under the conventional framework of negligence, the Claimant must establish a number of tests: the Claimant must establish that the Defendant (healthcare professional) owed the Claimant (the patient) a duty of care; that the Defendant breached that duty of care; the breach of the duty by the Defendant caused the damaged complained of by the Claimant; and that damage is not too remote. Actions in negligence far outweigh those of battery, in the context of informed consent cases. This is possibly due to the nature and historical definition of battery (element of hostility), and the repercussions a claim of battery could have on the sacrosanct doctor-patient relationship.

In informed consent cases, the focus under the tort of negligence, differs to that of battery. Under this framework, the courts focus less on the expression of informed consent, and more on whether the healthcare professional has effectively discharged their duty of care; this is

¹⁹ Ibid.

²⁰ *Chatterton v Gerson* (1981) QB 1, "In my judgment once the patient is informed in broad terms of the nature of the procedure, which is intended, and gives her consent, that consent is real, and the cause of action on which to base the claim for failure to go into risk and implications is negligence, not trespass", 443.

²¹ Sheila A.M. McLean (n17) 71.

demonstrated by establishing that they have taken reasonable steps to support patient choice and understanding, following the disclosure of material information.

Hippocratic Doctors

The approach of draconian doctors to medical practice was not synonymous with the notion that patients were autonomous entities, who obtained the ability to make their own decisions, regarding medical treatment.²² Patients were subservient beings, with the interests of the profession typically outweighing that of the patient. Indeed, it was commonplace for patients to consent to treatment, without obtaining the relevant material information to make an informed choice.²³

Hippocratic doctors aspired to preserve the bioethical principles of non-maleficence and beneficence, to develop a 'healthy' doctor-patient relationship.²⁴ Attributed to ancient Greece and the father of medicine, Hippocrates of Kos, the Hippocratic oath is an oath of ethics, which has been historically taken to guide HCPs in their approach to medical care.²⁵ While the oath was written almost 2500 years ago, it still forms a crucial component of the famous text in Western medicine.²⁶ To swear by the oath is a HCP's pledge to uphold specific medical standards of care.²⁷

While the oath was designed to act as a moral compass to HCPs, historically, Hippocratic doctors would consider a "good patient" to be a submissive patient, one who would not challenge the authority and treatment decisions of the HCP.²⁸ Doctors were viewed as all-knowing entities, with patients commonly placing their entire trust and confidence in the decision-making abilities of the doctor. The nature of this unilateral approach to care resembles the relationship between father and son, hence the prevalence and use of the term paternalism to describe the historic relationship, between professional and patient.²⁹ This

²² Emily Jackson (n6), 167.

²³ Jean McHale and Marie Fox, *Health Care Law*, (2nd edn, London Sweet & Maxwell 1997) 349-395.

²⁴ Riyaz Kaba & Prasanna Sooriakumaran, *The evolution of the doctor-patient relationship*, (2007) 5 *International Journal of Surgery*, 57.

²⁵ Kathy Oxtoby, *Is the Hippocratic Oath Still Relevant to Practising Doctors Today?*, (2016) 335 *British Medical Journal*, available at: <https://www.bmj.com/content/355/bmj.i6629> (accessed 08/07/2019).

²⁶ *Ibid.*

²⁷ *Ibid.*

²⁸ Riyaz Kaba & Prasanna Sooriakumaran, (n24) 57.

²⁹ JJ Chin, *Doctor-patient Relationship: from Medical Paternalism to Enhanced Autonomy*, (2002) 43 *Singapore Medical Journal*, 152-155.

period of medical history was largely dominated by utilitarian/consequentialist and paternalistic practices, commonly being reported as violating the Beauchamp and Childress four pillars of bioethics: patient autonomy, beneficence, non-maleficence and justice.³⁰

Jackson addressed the historical status of patients, in the context of medical practice. She explained that, historically, patients did not acquire an “autonomy-based right to be provided with information”.³¹ Traditional paternalistic practices would require patients to be notified by the doctors what was going to happen to them, on the basis that the decision would reflect the best interests of that patient.³²

Historical Prominence of Medical Paternalism

Philosophically, paternalism is described as the interference with a person’s autonomy or liberty, to either promote good or to prevent harm to that person.³³ The right to act autonomously, is often referred to as the antithesis to philosophical perspectives of paternalism. To be autonomous, is homogeneous with principles of self-determination and self-governance, and the ability to act freely, in the absence of physical or psychological constraints or actions from another.³⁴ Paternalists assert that an interference with patient autonomy is justified, where the doctor’s primary incentive of interfering with autonomy, is in patients’ best interests. To restrict autonomy, from a paternalist perspective, is ultimately to prevent any undue harm to the patient.³⁵ A paternalistic model of care often described as being grounded in the bioethical principle of beneficence.³⁶

An appreciation of early beneficence-motivated models of care, is historically relevant in the development of the standard of medical disclosure. In essence, adhering to the principle of beneficence is the duty to help others; this is fulfilled through prioritising patient welfare, in conjunction with the professional’s duty to benefit the patient, and weighing the risk of harm

³⁰ Rebecca Roache, *Making consequentialism more appealing*, (2015) 41, *Journal of Medical Ethics*, 359.

³¹ Emily Jackson, (n6) 168.

³² *Ibid*; see, *Slater v. Baker & Stapleton* (1767) 95 Eng. 860, 2 Wils. KB 359. In this case, the doctor broke the patient’s leg without obtaining consent first. Court found in favour of the Defendant, as the doctors actions were said to be justified in light of the situation.

³³ Robert Timko, (n8), 116.

³⁴ Wendy Margolis, *‘The Doctor Knows Best: Patient Capacity for Health Care Decision-making’*, (1992) 71 *Oregon Law Review*, 911.

³⁵ Emily Jackson, (n6) 167.

³⁶ Barry Main & Adair, S. R. L, *‘The Changing Face of Informed Consent’*, (2015) 219 *British Dental Journal*, 325.

against the potential benefits of an action.³⁷ This is also described as the Hippocratic model of care, whereby HCPs would take full responsibility for their patients' welfare, under the role as authoritarian decisionmakers.³⁸ However, this model of care was notoriously associated with the HCP's narrow duty of care, rescinding any *obligation* to address the patient's values, expectations and decisions.³⁹ Commentators and ethicists note that this beneficence-motivated model of care, created a foundation for the evolution of paternalism.⁴⁰

While scholars such as Margolis describe historical approaches to care as authoritarian, this term should not be used interchangeably with the notion of paternalism. Indeed, both models of care potentially result to an imbalance between professional and patient interests; however, the primary objective of paternalistic doctors was to impede autonomy for the patient's own well-being. Conversely, authoritarianism is synonymous with the notion that autonomy is restricted, to enhance the all-powerful status of the HCP.⁴¹

Legal literature typically neglects discussion of utilitarianism, in favour of labelling historic Hippocratic medical practices as wholly paternalistic. The theory of utilitarianism, a cousin of paternalism, accepts that compelling a patient to undergo unwanted treatment, is justified in the greater good, despite violating the patient's rights and desires.⁴² An act of utilitarianism analyses and balances the benefits and harms to promote an overall better consequence, without examining past evidence or experience.⁴³ Utilitarians are society-centred.⁴⁴ A more accurate reflection of the Hippocratic model of care encompasses elements of beneficence with paternalism, and utilitarianism.

Scholars commonly refer to the term 'medical paternalism', to describe the historical imbalance in patient and professional interests. An early commitment to beneficent models of care meant that Hippocratic doctors would decide on a course of treatment in the patient's best interests, and the outcome would retrospectively justify that decision.⁴⁵ McKinstry

³⁷ Wendy Margolis, (n34) 911.

³⁸ Ibid.

³⁹ Ibid.

⁴⁰ Ibid.

⁴¹ Ibid.

⁴² Wendy E. Roop, '*Not in My Womb: Compelled Prenatal Genetic Testing*', (2000) 27 *Hastings Const. L.Q.* 397.

⁴³ Jharna Mandal, Dinoop Korol Ponnambath and Subhash Chandra Parija, '*Utilitarian and Deontological Ethics in Medicine*' (2016) 6 *Tropical Parasitology* 5-7.

⁴⁴ Ibid.

⁴⁵ JJ Chin, (n29) 155.

explains that the central debate on paternalism, originates from whether doctors are justified in making decisions about patients' treatment, to which they know the patients would not agree to embark on the proposed course of treatment, if properly informed.⁴⁶

Foster argues that the reported 'paternalistic culture' extended from the lack of medical resources and technology, that was at HCPs' disposal in the early stages of medical care, limiting their ability, in some circumstances, to treat every patients' condition.⁴⁷ Without a sufficient understanding or knowledge of a patient's health, which was historically commonplace due to basic medical science and technology, this would place HCPs in a vulnerable position, exposing them to litigation. Creating a culture of paternalism may then have provided the collective protection for HCPs, when faced with claims for medical malpractice.⁴⁸

1.1.2 Paternalism and the 'Bolamisation' of Consent

The decision in *Bolam*⁴⁹ is often cited in the context of promoting medical paternalism, and for exhibiting an undue preference for professional opinion on matters of clinical judgment.⁵⁰ Sutherland QC expressed that "the problem with ... *Bolam* in the area of information disclosure is that this test is more concerned with professional consensus and standards than with the rights of the patient".⁵¹ The decision also raised questions regarding the constitution and balance of professional and patient interests, in clinical practice.

In the case of *Bolam*, the Claimant was mentally-ill and underwent electro convulsive therapy, as a recommended course of treatment. Correct medical practice would have required the HCP to administer a muscle relaxant drug, to avoid any injury caused during the treatment, and to also warn of the inherent risks associated with the drug and the electro-convulsive therapy itself. The HCP failed to administer the drug, which consequently resulted in the Claimant suffering from a serious fracture. The HCP had also failed to warn the patient of the

⁴⁶ Brian McKinstry, 'Paternalism and the doctor-patient relationship in general practice', (1992) 42 Br J Gen Pract, 340.

⁴⁷ Charles Foster, 'The rebirth of medical paternalism: An NHS Trust v Y', (2019) 43 Journal of Medical Ethics 3-7.

⁴⁸ Jo Samanta and Ash Samanta, *Medical Law*, (1st edn, Palgrave Macmillan Law Masters, 2011) 139-155.

⁴⁹ *Bolam v Friern Hospital Management Committee* (1957) 2 All ER 188.

⁵⁰ Lauren Sutherland QC, (n15), 22.

⁵¹ *Ibid*, at 22.

risks associated with the administration of the drug and the electro-convulsive therapy, in pre-treatment conversation.

Professional opinion was divided, in this case, as to whether the administration of the drug, and disclosure of the inherent risks, were necessary or not. The Claimant expressed that the HCP had breached their legal duty of care, by not administering the drug before performing the procedure. The House of Lords held that the doctor had not breached their duty of care. The 'prudent doctor' standard, for establishing liability in negligence, was forged by the British courts in *Bolam*, commonly referred to as the '*Bolam* test':

'... such failure as no ordinary doctor of skill would be guilty of, if acting with ordinary care'.

Typically, academic literature and case law refers to the *Bolam* test, in the context of assessing whether a medical practitioner has acted negligently, in accordance with whether the practice was accepted as 'proper' by a responsible body of medical men skilled in that particular art.⁵² McNair LJ, states:

'... a doctor is not negligent, if he is acting in accordance with a practice accepted as proper by a responsible body of medical men skilled in that particular art, merely because there is a body of such opinion that takes a contrary view'.

In *Bolam*, the judgment did not only refer to this test, it also referred to another test⁵³ from the case of *Hunter v Hanley*⁵⁴ of reasonable skill and care. Lord Clyde stated:

'... The true test for establishing negligence in diagnosis or treatment on the part of the doctor is whether he has been proved to be guilty of such failure as no doctor of ordinary skill would be guilty of, if acting with ordinary care'.⁵⁵

However, hereinafter, the '*Bolam* test' will refer to the 'responsible body of medical opinion' test.

Bolam set the legal standard for establishing negligence. It was deemed advantageous, at the time, as its clarity and robust characteristics could be easily understood by the legal and

⁵² Ibid

⁵³ Ibid.

⁵⁴ *Hunter v Hanley* (1955) SLT 213.

⁵⁵ Ibid.

medical professions.⁵⁶ The *Bolam* test provided the necessary certainty, which the law desired, placing a desired degree of discretion into the hands of the medical profession who were, at the time, deemed to be best placed to decide on such matters.⁵⁷

A pivotal criticism of *Bolam*, was the courts inability to distinguish between ‘what is done’, and ‘what ought to be done’⁵⁸; in other words, whether the *Bolam* test established if the HCP exercised reasonable skill and care, or whether the HCP complied with proper standards of practice.⁵⁹ Stone explains that the doctor’s non-disclosure of the risks, regarding electro-convulsive therapy, was largely overshadowed by the reported breach of the doctor’s duty of care, in failing to administer the required drug (muscle relaxant), to negate any potential harmful side-effects.⁶⁰

The actions of the HCP in *Bolam* should have been measured against a standard of what should have been done (warn of the risks inherent attached to the drug and electro-convulsive therapy), as opposed to what was done (failure to administer the drug). Therefore, there were two separate issues which needed to be considered by the courts: (i) what is the choice of approach to care management; and (ii) did the patient consent to that treatment.⁶¹ The *Bolam* test merely extended the HCP’s duty of care to post-treatment conduct, rather than placing a duty on HCPs to consider their pre-treatment discourse, exposing the standard to self-regulation.⁶²

Bolam is often referred to in the context of removing judicial discretion from the courts, and placing it into the hands of medical professionals, creating the historic paternalistic culture⁶³; this is not wholly true. While the professional body of medical *opinion* proved influential on the decision, the courts *still* retained judicial autonomy. Indeed, a misinterpretation of

⁵⁶ Kenyon Mason, ‘*Bolam, Bolam – Wherefore Art Thou Bolam*’, (2005) 9 Edinburgh Law Review, 299.

⁵⁷ Ibid.

⁵⁸ Ash Samantha and Jo Samantha, ‘*Legal Standard of Care: A Shift From the Traditional Bolam Test*’, (2003) 3 Clinical Medicine, 443.

⁵⁹ John Keown, ‘*Doctor Knows Best: The Rise and Rise of the Bolam Test*’, (1995) 5 Singapore Journal of Legal Studies, 343.

⁶⁰ Christopher Stone, ‘*The Decision in Birch Marks Another Step Away From the Much Criticised Sidaway Approach to Consent*’, (2010) 5 1-2.

⁶¹ Ibid.

⁶² Ash Samantha and Jo Samantha, (n58), 444.

⁶³ Ibid.

Bolam's application resulted to the removal of judicial discretion, with the courts exhibiting an undue preference for medical profession opinion on matters of clinical practice.

1.1.3 The Judicial Politics in *Sidaway* and its Ramifications

The House of Lords in *Sidaway v Board of Governors of the Bethlem Royal Hospital*⁶⁴ sought to delineate professional and patient interests, in their assessment of professional duty and information disclosure. While 'pragmatic' and familiar *Bolam* test swayed the majority in *Sidaway*, a minority expressed the importance of patients' rights, and the need to reinstate judicial discretion.

The case of *Sidaway* concerned a Claimant who had been left severely disabled following a spinal operation. Ms Sidaway claimed that the doctor had been negligent following the failure, in pre-treatment discussions, to disclose the risk of paralysis following the proposed procedure. Ms Sidaway based her claim on the surgeon's failure to adequately inform her of all the possible risks attached to that operation. Counsel for the Claimant submitted that the key issue, in this case, was whether the surgeon had adequately satisfied his twofold obligation, to provide the relevant information to the patient.⁶⁵ The first obligation was to ensure that the patient had delivered valid consent, and the second obligation was to satisfy the duty placed upon HCPs to advise and inform. This required the courts to assess the scope of HCPs' duty, and whether the risk was 'material'.

While Lords Diplock, Bridge/Keith and Templeman represented the majority decision – applying *Bolam* to assess the HCP's duty for information disclosure – Lord Scarman notably stressed his dissatisfaction toward the inflexibility of existing legal mechanisms for assessing professional duty. His focus rested on the importance of patients' rights to assess the doctor's duty, and while he did not specifically seek to apply the common law doctrine, he sought to promote the rights-based values of the North American and Canadian doctrine, to delineate patient and professional interests.⁶⁶

With the judiciary exhibiting a respect for the growing rights-based narrative, exhibited by Lord Scarman in *Sidaway*, an incremental blending of legal principles could be witnessed,

⁶⁴ *Sidaway v. Board of Governors of the Bethlem Royal Hospital* (1985) A.C 871

⁶⁵ *Ibid*, 874.

⁶⁶ *Ibid*, 888, "My Lords, I think the *Canterbury* propositions reflect a legal truth which too much judicial reliance on medical judgment tends to obscure ... the doctor's duty arises from his patient's rights".

moulding a standard of care, which accurately reflected the changing socio-political climate. Scholarly literature engaged in discussions of ‘rights’ and ‘patient autonomy’ at this time, an exercise which was rarely observed hitherto.⁶⁷

1.1.4 The Position of the Australian Jurisprudence – Taking an Early Lead

For the purpose of coherence, it is worth noting the approach of the Australian judiciary, in the anticipation of the enactment of the HRA, and its influence on the British legal system. Despite the conservative view of the British judiciary at this time, the Australian jurisprudence recognised that, to vindicate the rights of patients, reflects a parochialistic legal system, preventing progression towards a cogent standard of professional duty to disclosure. Indeed, the approach of the Australian jurisprudence arguably stimulated a domestic rights-based environment, and judicial discussion turned to that of the doctrine.⁶⁸ While the persuasiveness of these cases only became apparent a decade after the final judgment, a case which proved to be highly influential, in subsequent British case law⁶⁹, was the decision in *Rogers v Whittaker*⁷⁰, in 1993.

In *Rogers*, the Claimant had problems with her right eye. The Defendant (surgeon) advised the Claimant that the operation would improve the appearance of the eye, and could restore significant sight to it. The Claimant agreed to undergo the surgery, after persistent questioning of the risks associated with the procedure. Following the operation, there was no improvement to the right eye, and she had also developed a condition, called sympathetic ophthalmia, in her left (previously healthy) eye. This resulted to the loss of sight in her left eye. This risk was not disclosed by the Defendant, in pre-treatment discussion. The Claimant sought damages in negligence. The court found in her favour, highlighting that the risk of sympathetic ophthalmia was material, and should therefore have been disclosed, in pre-treatment discussion.

⁶⁷ Michael Jones, “Informed Consent and Other Fairy Stories”, (1999) 7 Medical Law Review 1, 103.

⁶⁸ Ibid.

⁶⁹ Namely *Montgomery v Lanarkshire Health Board* (2015).

⁷⁰ *Rogers v Whittaker* (1993) 4 Med. L.R. 79.

This case highlighted the sincere judicial dissatisfaction to apply the *Bolam* standard, in these circumstances.⁷¹ A medical body of opinion would have supported the premise that the risk of sympathetic ophthalmia should not have been disclosed, as the patient did not *specifically* or *directly* ask about the risk.

The assessment of materiality of risk, in *Rogers*, expanded on the approach of Lord Scarman in *Sidaway*, assessing whether a risk was material, in accordance with if a HCP *should* have an appreciation of its relevance to the patient, even if the patient was unaware of the risk:

*'... a risk is material if, in the circumstances of the particular case, a reasonable person in the patient's position, if warned of the risk, would be likely to attach significance to it or if the medical practitioner is or should reasonably be aware that the patient, if warned of the risk, would be likely to attach significance to it.'*⁷²

This two-fold test of materiality comprised of both objective and subjective criteria. The first limb – whether a reasonable or ordinary person in the patient's position would attach significance to the risk – would be assessed objectively.⁷³ The second limb – whether a medical practitioner is or should be reasonably aware that the particular patient would attach significance to it – would be assessed subjectively.⁷⁴ This second limb addresses the concept that a patient may *not* be reasonable, providing judicial discretion to consider the particular patient, and their "requirements or fears (reasonable or unreasonable)".⁷⁵

Rogers sparked a cross-fertilisation of legal principles.⁷⁶ This notion of cross-fertilisation is created when an external stimulus, that being the decision of Lord Scarman, encourages the evolution of an ideology or doctrine, in the receiving legal system.⁷⁷ A triangulation effect and communication can be observed between the North American, Australian and British legal systems, due to the raising matters of common concern pertaining to patients' rights and informed choice. As a result, these multi-national jurisprudences were (and still are) the

⁷¹ Ibid, 18.

⁷² Ibid, 16.

⁷³ Lauren Sutherland QC, '*Montgomery: Myths, Misconceptions and Misunderstandings*', (2019) 3 Journal of Personal Injury Law 157-167.

⁷⁴ Ibid.

⁷⁵ Ibid, 161.

⁷⁶ Margit Cohn, '*Legal Transplant Chronicles: The Evaluation of Unreasonableness and Proportionality Review of the Administration in the United Kingdom*', (2010) 58 The American Journal of Comparative Law 583, 629.

⁷⁷ Ibid, 595.

subject of international cross-fertilisation.⁷⁸ *Rogers* would prove to be very influential in subsequent British case law, amplifying the importance restoring the significance of patient and professional interests, to the question of duty and disclosure.

1.1.5 Reinstating Judicial Interests

The first noticeable deviation in the British legal narrative surfaced in the decision in *Bolitho v Hackney*, in 1997, which sought to restore judicial interests for actions in negligence, pertaining to diagnosis and treatment.⁷⁹

In *Bolitho*, a HCP failed to intubate a two-year old child, resulting in the child's death. Evidence was presented by another HCP, revealing that they would also not have intubated. The trial judge held that there was no breach of duty, following the application of *Bolam*. However, on appeal, the courts restricted the largely unfettered application of the *Bolam* test, by introducing a precondition to its application. The House of Lords in *Bolitho* provided a 'gloss' to *Bolam*. *Bolitho* rearticulated what was implicit in *Bolam*, that the final decision was for the court, which had to be satisfied that the requisite standard was met, but had fallen by the wayside in subsequent cases, displaying undue deference to medical opinion. Lord Browne-Wilkinson stated that:

'The effect of Bolam test is that the defendant must live up to the standard of the ordinary skilled man exercising and professing to have special skill. The existence of the practice is not of itself determinative of the issues of breach of duty. The court has to subject the expert medical evidence to scrutiny and to decide whether the practice is reasonable. The issue of reasonableness is for the court and not for the medical profession.'

'Reasonableness', according to Lord Browne-Wilkinson, was decided on the basis of balancing risks and benefits, and where medical practice is accepted by a responsible body of professionals, it must be shown that the method used was logical and defensible.

Bolitho was symbolic of reminding the courts that the judiciary were ultimately responsible for deciding whether a HCP's conduct met the prevailing standard of medical care. *Bolitho*

⁷⁸ See also *Arndt v Smith* (1997) 3 LRC 198. The Canadian Supreme Court had also reassessed the reasonable patient test.

⁷⁹ *Bolitho (Deceased) v City and Hackney HA* (1997) 3 W.L.R. 1151.

was said to have “clipped the wings”⁸⁰ of *Bolam*, with the articulation of what the *intended* role of the courts *should* have been, in *Bolam*. *Bolitho*’s introduction of a ‘reasonableness’ caveat restored the necessary judicial discretion in negligence cases.

On one hand, although it seems that the inclusion of the condition in *Bolitho* – that the practice had to be logical and defensible, as well as being accepted by a responsible body of professionals – would be beneficial to the position of the Claimant in negligence cases; this is not necessarily true, in practice. Arguably, the test in *Bolitho* only strengthens the position of the Defendant, as the Claimant would need to persuade the courts that the defence expert evidence fails the *Bolitho* test, and is not reasonable, logical or responsible. Essentially, *Bolitho* requires the court to dismiss the defence expert evidence as illogical, not responsible and unreasonable, which is an incredibly high threshold for a Claimant, and one that would rarely be satisfied.⁸¹

Influence of the Human Rights Act – Change of Climate

The advent of the Human Rights Act 1998 provided an unexpected influence on the law of consent and medical decision-making.⁸² The HRA represented a revolution in the preservation and protection of individual rights, in the United Kingdom.⁸³ The HRA became immediately relevant to healthcare providers, as it regulates the relationship between individuals (patients) and public authorities (the NHS), and would be unlawful for public authorities to ‘act in a way which is incompatible with a Convention right.’⁸⁴

The British Medical Association Committee on Medical Ethics reported that medical practitioners were not familiar with thinking in the context of ‘rights’.⁸⁵ However, post-HRA, medical professionals exhibited an increasing amount of respect for patients’ rights.⁸⁶ Professional guidelines, protocols and standards began to incorporate reassessments and

⁸⁰ Christopher Stone, ‘*From Bolam to Bolitho: Unravelling Medical Protectionism*’, (2011), 6.

⁸¹ *Ibid.*

⁸² British Medical Association ‘Committee on Medical Ethics: The Impact of the Human Rights Act 1998 on Medical Decision-Making’, (2000).

⁸³ *Ibid.*

⁸⁴ *Ibid.*

⁸⁵ *Ibid.*

⁸⁶ Barbara Hewson, ‘*Why the Human Rights Act Matters to Doctors: Definitions of Informed Consent and Negligence May Be Challenged, and Lack of Resources Will Not Excuse Poor Care*’, (2000) 321 *The British Medical Journal* 780, 781.

evaluations of professional medical conduct, in conjunction with standards of care, reflecting the act's values and principles.⁸⁷

Human rights are inalienable fundamental rights, providing persons with the entitlement to enjoy these rights, merely because he/she is a human being.⁸⁸ Human rights are indivisible, and include the right to life, property, health, education and more.⁸⁹ The HRA provides domestic force to the European Convention on Human Rights (ECHR)⁹⁰, in the United Kingdom. The Convention is a living instrument, meaning that it is capable, once interpreted, to correspond with the developing social narrative.⁹¹ As such, it could also be interpreted to reflect patient expectation, and conventional ethical standards.⁹² Furthermore, the ECHR, under the HRA, is directly applicable and enforceable in the UK.⁹³ Rights which fall under it may be separated into three different categories: (i) absolute rights are those which cannot be justifiably restricted or made subject to conditions; (ii) limited rights are those which can be restricted only in exceptional circumstances, such as protecting the rights of other people; and (iii) qualified rights are rights which may be interfered with to protect the rights of another, or in the public interest. Any limitations or interferences with a person's human rights must be justified, such as the interference is prescribed by law, has a legitimate aim or is proportionate.

All public authorities have a statutory duty to comply with the Convention rights. Under the Act, it is strictly unlawful, for public authorities, to act in a way which is incompatible with Convention rights. As the NHS is a public body, its practices, trusts, health authorities and clinics, which are licensed by the state, will fall beneath the ambit of the act.⁹⁴ Patients may possess the right to commence legal proceedings against the NHS, if they believe their rights have been violated.⁹⁵

⁸⁷ Andrew Grubb and Judith Laing, *Principles of Medical Law*, (2nd Edn, OUP 2004) 132.

⁸⁸ Philippe Hanna and Frank Vanclay, '*Human Rights, Indigenous Peoples and the Concept of Free, Prior and Informed Consent*' (2011) 31 Taylor & Francis 146, 157.

⁸⁹ Ibid.

⁹⁰ Council of Europe, *European Convention for the Protection of Human Rights and Fundamental Freedoms, as amended by Protocols Nos. 11 and 14*, 4 November 1950, ETS 5.

⁹¹ British Medical Association, '*Committee on Medical Ethics: The Impact of the Human Rights Act 1998 on Medical Decision-Making*' (2000)

⁹² Ibid.

⁹³ Ibid.

⁹⁴ Barbara Hewson (n86), 781.

⁹⁵ Ibid.

Article 8 of the European Convention of Human Rights, which protects the right of individual privacy and physical integrity, has been interpreted and justified (under Article 8(2) of the ECHR), to include the decision-making autonomy of the individual.⁹⁶ Through the interpretation and statutory duty conferred by the act, NHS professional standards, guidance and protocols, were prompted to reflect the importance of patient autonomy and patient-centric care, incorporating these principles into existing legal and ethical frameworks.

Scholars and practitioners anticipated that consent would be a “hot topic”⁹⁷, at common law, in the early years following the enactment of the HRA, and concerns existed as to the adequacy of the *Bolam* to effectively delineate patient and professional interests, in cases of information disclosure.⁹⁸

1.1.6 *Pearce* – Paving a Pro-Patient Path

The decisions at common law, on the crest of the implementation of the HRA 1998, highlighted a deviation in the legal narrative of the British jurisprudence. Socio-cultural values had started to weave a rights-thread into its expanding tapestry. In the early years post-HRA, it became commonplace to detect discussion of ‘rights’ and ‘patient choice’, in both scholarly literature, and judgments at common law.

The first example of case law, which exhibited a pro-patient trajectory, was the decision in *Pearce v United Bristol Healthcare*, in 1998.⁹⁹ Mrs Pearce was advised by the HCP to delay the induction of childbirth. The child was unfortunately stillborn. Mrs Pearce claimed that the HCP should have disclosed the risk that her child could have potentially been stillborn. The court held that a risk of 0.1-0.2% was not categorised as a ‘significant risk’, which consequentially justified the HCP’s failure to disclose that information.

The courts, in *Pearce*, ostensibly took steps to apply a reasonable patient standard, to assess materiality of risk, placing a responsibility on HCPs to inform the reasonable patient of significant risks, which could affect the patient’s judgement to treatment:

⁹⁶ *Pretty v UK* Application 2346/02 (2002) 66 BMLR 147 (ECtHR), 63. See also; *YF v Turkey* (2004) 39 EHRR 34 and *Glass v UK* (2004) 77 BMLR 120.

⁹⁷ Austen Garwood-Gowers, John Tingle and Tom Lewis, *Healthcare Law: The Impact of the Human Rights Act 1998*, (Cavendish Publishing 2001) 322.

⁹⁸ *Ibid.*

⁹⁹ *Pearce v United Bristol Healthcare NHS Trust* (1998) 48 BMLR 118.

*'... it seems to me to be the law ... that if there is a significant risk which would affect the judgment of a reasonable patient, then in the normal course it is the responsibility of a doctor to inform the patient of that significant risk ...'.*¹⁰⁰

Arguably, the decision effectively articulated the importance of placing the interests of patients, in equal measure to that of the profession, when deciding what information is 'material':

*'... the doctor ... has to take into account all the relevant considerations, which include the ability of the patient to comprehend what he has to say to him or her and the state of the patients at the particular time, both from the physical point of view and ... emotional'.*¹⁰¹

Following his assessment of materiality, Lord Woolf alluded to a duty that places an obligation on HCPs, to holistically consider the interests of the patient, by understanding their needs and wishes, at pre-treatment consultations.

Lord Woolf also placed emphasis on the expert witness evidence, which conveyed a 'significant' or 'material' risk, in terms of a percentage:

*'... if the risk ... was of the order of 10%, for instance, then of course it would be my duty to warn against such level of risk'.*¹⁰²

This complex amalgamation of the 'reasonable patient' and 'reasonable HCP' standard, was hailed by scholars as being pro-patient, despite the clear reliance on *Bolam* to assess materiality of risk.¹⁰³ It is also commended for endorsing patient rights and choice.¹⁰⁴ Stone goes further to explain that the objective reasonable patient standard, in *Pearce*, had effectively distanced *Bolam* from the legal standard.¹⁰⁵

¹⁰⁰ Ibid, 124.

¹⁰¹ Ibid, 125.

¹⁰² Ibid, 124, "When one refers to a "significant risk" it is not possible to talk in precise percentages, but I note, and it may be purely coincidental, that one of the expert doctors who gave evidence before the judge gave the following answer in evidence."

¹⁰³ Shaun Pattinson, *Medical Law and Ethics*, (Sweet & Maxwell 2011) 128.

¹⁰⁴ Jean McHale and Marie Fox, (n23) 386.

¹⁰⁵ Christopher Stone (n60) 4.

It is questionable, however, whether this decision took the law any further than the reasonable doctor test, due to the inherently disproportionate reliance upon the judgement of the HCPs, as to whether a risk was significant.¹⁰⁶

1.1.7 Wyatt – Reassessing Professional Duty

Lord Justice Sedley, in *Wyatt v Curtis*¹⁰⁷, reassessed the legal standard of care, by evaluating the historical judicial approaches to the standard of disclosure. He attempted to carve a standard that would clarify the ambiguity, post-*Pearce*, surrounding professional duty for information disclosure.

In this case, the HCP (Dr Curtis) failed to warn Miss Wyatt of the risks and complications associated with chickenpox, to the health of her unborn child. Her child was subsequently born with complications, due to the chickenpox. Sedley LJ assessed Lord Bridge's substantial risk exception from *Sidaway*, in conjunction with the standard set by Lord Woolf in *Pearce*, to evaluate whether the standard of disclosure should be positioned subjectively from the patient's perspective, as to what they considered to be a significant risk, or from the perspective of the HCP:

*'To the doctor, a chance in a hundred that the patient's chickenpox may produce an abnormality in the foetus may well be an insubstantial chance, and an abnormality may in any case not be grave. To the patient, a new risk which ... doubles, or at least enhances, the background risk of a potentially catastrophic abnormality may well be both substantial and grave, or at least sufficiently real for her to want to make an informed decision about it.'*¹⁰⁸

Sedley LJ found in favour of the Claimant, in this case. The "this patient" test was formulated following the decision. During his assessment of duty and materiality, Sedley LJ stated:

*'Lord Woolf's formulation refines Lord Bridge's test by recognising that what is substantial and what is grave are questions on which the doctor's and the patient's perception may differ, and in relation to which the doctor must therefore have regard to what may be the patient's perception.'*¹⁰⁹

¹⁰⁶ Robert Heywood, 'Re-Thinking the Decision in Pearce', (2005) 7 CIL 264, 270.

¹⁰⁷ *Wyatt v Curtis* (2003) EWCA Civ 1779.

¹⁰⁸ *Ibid*, 16.

¹⁰⁹ *Ibid*, 16.

Sedley LJ also placed emphasis and significance on pre-treatment care; in particular, the quality of conversation and dialogue, between doctors and patients, to effectively assess the patient's best interests, and what they *expect* from the treatment.¹¹⁰

1.1.8 Enigmatic *Chester*

Academic commentary on *Chester v Afshar*¹¹¹ commonly reinforce the significance of the decision, in terms of reshaping the law of causation; that is, its abandonment conventional causation principles, to correspond with existing policy: the preservation of patient autonomy and rights. However, the impact of *Chester*, for promoting patients' rights and autonomy in the context of assessing professional duty for information disclosure, is often omitted from discussion, but has transpired to be highly significant in this regard.

In *Chester*, the Claimant had suffered from chronic back pain for years. She had an MRI scan, which revealed that she had a disc protrusion, requiring surgery. This operation carried a 1-2% risk that, even if carried out properly, could worsen her condition, rather than improve it. The doctor failed to warn the patient of the risk. Following surgery, her condition worsened. The judiciary assessed three key considerations, in this case: (i) if evidence suggests that the patient agreed that she would never have undergone the operation if warned of the risk, damages could be awarded; (ii) if evidence revealed that she would still have undergone the operation, at the same time and in the same manner, then damages could not be awarded; and (iii) medical evidence asserted that, had she been warned of this risk, she would not have decided to undertake the surgery *immediately*, and would have taken time to consider the *alternative* options.¹¹² Crucially, she did not claim that she would have opted out of the operation altogether.¹¹³

The failure to disclose the risk was held not to invalidate the Claimant's consent to surgery; therefore, there could be no action in battery. However, if the HCP's failure to disclose the risk was deemed to be *unreasonable*, then the patient could have a claim in negligence. A majority of the House of Lords agreed with the decision of the Court of Appeal, that the

¹¹⁰ Jean McHale and Marie Fox, (n23) 395.

¹¹¹ *Chester v Afshar* (2004) UKHL 41.

¹¹² Tamsyn Clark and Donal Nolan, 'A Critique of *Chester v Afshar*' (2014) 34 *Oxford Journal of Legal Studies*, 659, 692.

¹¹³ *Chester* (n111) 40.

surgeon had not performed the operation negligently; however, his failure to warn of the 1-2% risk breached his duty of care.¹¹⁴ The judiciary held that, if the patient was provided with the choice to undertake the operation at a later date, it may have been successful, and therefore found in favour of the Claimant:

'... the law which imposed the duty to warn on the doctor has at its heart the right of the patient to make an informed choice as to whether, and if so when and by whom, to be operated on'.¹¹⁵

While the decision divided the opinion of eminent tort and medical law specialists, commentators commended the decision, concluding that the courts “got it right”, in the context of moulding an appropriate standard of care.¹¹⁶ It was evident that the court, in *Chester*, was committed to the surfacing socio-legal narrative surrounding the protection of patient choice; it sought to develop the obligations placed upon HCPs to preserve autonomy, and to demonstrate an appreciation of patient interests and choice, in deciding on materiality of risk.¹¹⁷ The courts broadened the scope of the HCP’s duty, to oblige the disclosure of information and advice, in relation to *alternative* and *variant* methods of treatment.¹¹⁸

Lord Steyn opined that “*medical paternalism no longer rules*”,¹¹⁹ and his commitment to patient autonomy and choice, parroted that of the judiciary in *Wyatt* and subsequent case law, which had previously confirmed that wholly paternal approaches to care had no place in British law.

The court and supporters of the decision in *Chester*, beat the ‘patient autonomy’ drum, as a justification for the decision. Indeed, the majority, in the decision, established that an interference with patients’ rights and diminished autonomy, transpires as a configuration of damage in negligence.¹²⁰ Nevertheless, commentators also reference *Chester* as being highly

¹¹⁴ Ibid, 16, “... a patient has a *prima facie* right to be informed by a surgeon of a small, but well established, risk of serious injury as a result of surgery”.

¹¹⁵ Ibid, 86.

¹¹⁶ Andrew Grubb and Judith Laing (n87) 183.

¹¹⁷ *Chester* (n111), at 40 “It is not disputed that the failure to warn ... caused the injury if Miss Chester’s position had been that she would never have undertaken the operation at all if that warning had been given. But ... she never claimed that, if adequately advised of the risks, she would never at any time have consented to surgery”.

¹¹⁸ Ibid, 98; “In making a decision which may have a profound effect on her health and well-being a patient is entitled to information and advice about possible alternative or variant treatments”.

¹¹⁹ Ibid, 16.

¹²⁰ Craig Purshouse, ‘Judicial reasoning and the concept of damage: Rethinking medical negligence cases’ (2015) 15 *Medical Law International* 155, 181.

problematic, with its abandonment of conventional principles, to facilitate a policy driven preservation of patients' autonomy.¹²¹

1.1.9 *Birch* – Focus on 'Alternatives'

The decision in *Birch v UCL Hospital NHS Foundation Trust*¹²², capitalised on the trend to endorse patients' rights and autonomy, in the context of professional duty and information disclosure. Indeed, it became increasingly evident that the courts continued to deviate from conventional principles of law, to accommodate the developing rights-based narrative.

In *Birch*, the Claimant was suffering from vascular third nerve palsy. Birch consented, by signing a form, to undergo an angiogram by catheter (invasive method of treatment). This method of treatment was associated with a small (1%), but very serious risk of stroke. The risk materialised, and the patient suffered a stroke following the procedure. Mrs Birch was unaware that an MRI could have eliminated the risk of stroke, associated with the agreed course of treatment. Birch claimed that the HCP had been negligent in failing to disclose reasonable alternatives, including an MRI, which was a safer, non-invasive means of conducting the procedure. The Trust was held to be liable in negligence, as the HCP's failure to disclose the implications of the alternative imaging options, and non-disclosure of the comparative risks associated with the available alternative methods, impeded fully informed consent:

'... No authority was cited to this effect but in my judgment... the duty to inform a patient of the significant risks will not be discharged unless she is made aware that fewer, or no risks, are associated with another procedure ... In other words, unless the patient is informed of the comparative risks of different procedures she will not be in a position to give her fully informed consent to one procedure rather than another'.¹²³

The decision in *Birch* is remarkably undervalued in scholarly literature, with academics typically alluding to the case, rather than unpacking its significance.¹²⁴ Indeed, in deciding that, while the HCP informed the patient of the significant risks associated with the procedure, but did not disclose that an alternative treatment that held fewer risks, amounted

¹²¹ Tamsyn Clark and Donal Nolan, (n112), 659.

¹²² *Birch v UCL Hospital NHS Foundation Trust* (2008) EWHC 2237 (QB).

¹²³ *Ibid*, 74.

¹²⁴ Christopher Stone, (n60) 6.

to negligence, this undoubtedly clarified the legal standard of medical disclosure, post-*Chester*.¹²⁵ The basis for the decision, in *Chester*, placed the patient's autonomy interests at its core, deciding that a patient has a broad right to autonomy, diluting conventional legal principles on the standard of disclosure. However, *Birch* gave a legal footing to the duty placed on HCPs to disclose comparative risks, a concept which had only been broadly introduced by *Chester*.

1.2 *Montgomery* and its Ramifications

The Supreme Court in *Montgomery v Lanarkshire Health Board*¹²⁶ was said to have recalibrated the doctor-patient relationship, and subjected the medical profession to "increasing external scrutiny".¹²⁷ Comparative law was a strong theme in the judgment, with the judiciary referring to case law from Canadian and Australian jurisprudences, highlighting dissatisfaction with the ability of domestic case law to refine the current standard of information disclosure. The growing commitment to preserve patients' right to autonomy and self-determination, by addressing the flaws with a paternalistic model of care, was clear throughout the judgment. The ramifications of *Montgomery* were wide-reaching, and it forms our current standard, in the context of information disclosure.

The Claimant, Nadine Montgomery, was pregnant with her baby, Sam. Mrs Montgomery was diabetic, which increased the risks of complications when delivering the baby naturally (vaginal birth), as there was a 9-10% risk of the baby having shoulder dystocia. Furthermore, Nadine was small in stature, which increased the risk of complications during labour. She was told that she was having a baby that was larger than usual; however, the HCP failed to warn her of the risk of her experiencing mechanical problems during labour. Crucially, she was not told about the risks of shoulder dystocia. The HCP explained that, despite the 9-10% chance of the baby having shoulder dystocia, she did not spend any time discussing the potential risks, as grave problems for the baby resulting from shoulder dystocia were very small. The availability of an elective caesarean was also omitted from discussion. At her 36-week scan, Nadine demonstrated concern and anxiety about the size of her baby, and her ability to

¹²⁵ Shaun Pattinson, (n103) 130.

¹²⁶ *Montgomery v Lanarkshire Health Board* (2015) UKSC 11.

¹²⁷ Judy Laing, 'Delivering informed consent post-Montgomery: implications for medical practice and professionalism', (2017) 33 P.N, 150.

deliver vaginally. A 12-minute period had lapsed between the baby's head appearing, and the delivery. During this period, the umbilical cord was completely or partially occluded, resulting in the deprivation of oxygen. The baby was subsequently diagnosed with cerebral palsy. Ms Montgomery claimed that the doctor had acted negligently, in failing to disclose the risk associated with shoulder dystocia and delivering the baby vaginally. It surfaced that had the option of c-section been disclosed to Mrs Montgomery, she would have opted to do this, and the baby would ultimately had been delivered by c-section, to prevent injury to the child.

While the doctor presented evidence to suggest that policy justified the withholding of information on the risk of shoulder dystocia, reliance upon the therapeutic exception was held not to be justified; it was not designed to enable HCPs to actively withhold information from patients, which would consequentially erode autonomous informed decision-making and consent. The Supreme Court ruled that the HCP was under a duty to explain that the recommended treatment option was preferable, in comparison to the other available options, carefully *ensuring* the patient is aware of the associated material risks, and of any reasonable alternative methods of treatment:

'An adult of sound mind is entitled to decide which, if any, of the available forms of treatment to undergo ... the doctor is under a duty ... to ensure that the patient is aware of any material risks ... and of any reasonable alternative ... treatments'.¹²⁸

The court also held that the doctor's advisory role extended to maintaining a dialogue with the patient, outlining the anticipated benefits and risks of the proposed treatment, and any reasonable alternatives, to inform decision-making:

'... the doctor's advisory role involves dialogue, the aim of which is to ensure that the patient understands the seriousness of her condition, and the anticipated benefits and risks of the proposed treatment and any reasonable alternatives, so that she is then in a position to make an informed decision.'¹²⁹

This mirrored the approach to duty of care established by the Canadian¹³⁰ and Australian jurisprudence.¹³¹ It also expanded the scope of professional duty set by *Birch*; that patients

¹²⁸ Montgomery (n126), 87.

¹²⁹ Ibid, 90.

¹³⁰ Reibl v Hughes (1980) 2 S.C.R. 880.

¹³¹ Rogers (n70), 16.

should be aware of comparative risks, before obtaining informed consent to the proposed treatment.

The court's assessment of materiality was heavily influenced by the Australian case of *Rogers*,¹³² which had been decided 25-years previously; there are very close parallels between the two cases.¹³³ The Supreme Court held that the test for materiality evaluates whether: (i) a reasonable person in the patient's position, if warned of the risk, is likely to attach significance to the risk; or (ii) that the HCP is or should be reasonably aware that the particular patient, if warned of the risk, would likely attach significance to it.¹³⁴ The introduction of this modernised two-stage test, derived from *Rogers*¹³⁵, placed focus on the principle of respect for patient choice, providing a clear legal footing for patient interests:

*'The social and legal developments which we have mentioned point away from a model of the relationship between the doctor and the patient based on medical paternalism. They also point away from a model based upon a view of the patient being entirely dependent on information provided by the doctor.'*¹³⁶

Commentators describe *Montgomery* as the key decision on informed consent, over the past 30-years.¹³⁷ Academics and practitioners posit that *Montgomery* had finally embraced the transatlantic doctrine, as a means to quash of medical paternalism.¹³⁸ The decision is commended for recognising that the law is a social construct, and is a product of the socio-cultural environment, which has substantially transitioned post-HRA:

*'... patients are now widely regarded as persons holding rights, rather than as the passive recipients of the care of the medical profession.'*¹³⁹ Indeed, the decision introduced a qualified 'right' to know.

¹³² Ibid.

¹³³ Rachael Mulheron, *'Has Montgomery Administered the Last Rites to Therapeutic Privilege? A Diagnosis and a Prognosis'*, (Oxford University Press, 2017) 149.

¹³⁴ *Montgomery* (n126) 87.

¹³⁵ *Rogers* (n70), 16.

¹³⁶ *Montgomery* (n126), 81.

¹³⁷ Clare Dyer, *'Doctors Should not Cherry Pick What Information to Give to Patients, Court Rules'* (2015) 350 *BMJ*, 141.

¹³⁸ Charles Foster, *'The Last Word on Consent?'* (2015) 165 *NLJ*, 8. Foster described this case as the "death knell of medical paternalism".

¹³⁹ *Montgomery* (n126) 75.

The Supreme Court had attempted to remove any historical doubts which existed in the context of the correct legal standard of disclosure, exercising a “tidying up of the law”.¹⁴⁰ While there were substantial social and cultural differences between Britain and North America – principally that North America placed an earlier significance on a patient-centric model of care, and patients’ right to self-determination – *Montgomery* produced a qualified symmetry between the British jurisprudence, with that of Australia and North America, confirming the social demand for revised standard of disclosure into British law.¹⁴¹ Indeed, it was stated, in *Montgomery*, that:

*‘the correct position, in relation to the risks of injury involved in treatment, can now be seen to be substantially that adopted in Sidaway by Lord Scarman, and Lord Woolf MR in Pearce subject to the refinement made by the High Court of Australia in Rogers.’*¹⁴²

However, following the judiciary’s conclusion – that the patient must be advised of any ‘reasonable’ alternative or variant methods of treatment, with the accompanying risks and benefits, before deciding to undertake treatment – the court failed to clarify how the ‘reasonableness’ standard should be assessed, in this context.¹⁴³ While caselaw, pre-*Montgomery*, addressed the concept of alternative methods of treatment, such as *Birch* (and to a degree *Chester*), the lack of judicial deliberation, in this in the decision, created further subjectivity, as to when an alternative method of treatment would require disclosure by a HCP.¹⁴⁴ This lack of judicial guidance could encourage the reincarnation of *Bolam* to decide on such matters.¹⁴⁵

The judiciary, in *Montgomery*, stated that the HCP’s advisory role extended to ensuring that the patient understood the information, before consenting to a course of treatment.¹⁴⁶ The HCP’s obligation would not be fulfilled, however, by bombarding the patient with technical information, in an attempt to meet the required standard. Indeed, the focus of the judiciary

¹⁴⁰ Michael Lamb, ‘*Montgomery: A Symbolic or Substantive Change to the Law?*’, (2017) 32 North East Law Review, 26.

¹⁴¹ *Montgomery* (n126), 81.

¹⁴² *Ibid*, 87.

¹⁴³ Louis V. Austin, “*Hii Chii Kok v (1) Ooi Peng Jin London Lucien; (2) National Cancer Centre: Modifying Montgomery*” (2019) 27 Med Law Rev 339, at 339-340.

¹⁴⁴ *Ibid*, 340.

¹⁴⁵ *Ibid*.

¹⁴⁶ *Montgomery* (n126), 90.

was building upon a continued dialogue with the patient, to understand their interests going to decision-making and consent:

*'This role will only be performed effectively if the information provided is comprehensible. The doctor's duty is not therefore fulfilled by bombarding the patient with technical information which she cannot reasonably be expected to grasp, let alone by routinely demanding her signature on a consent form.'*¹⁴⁷

Following its decision, it was clear that assessing the modified standard of materiality could be troublesome for the judiciary in future decisions, based on its interpretation in *Montgomery*. While the Supreme Court's evaluation of materiality was "symbiotically valuable",¹⁴⁸ as previous high court decisions were substantially ambiguous, it would be practically onerous for the courts to assess whether a HCP has taken reasonable care to increase the patient's awareness of any material risks, and whether the HCP should have been reasonably aware that the patient would attach significance to it.¹⁴⁹

While court agreed that materiality of risk cannot be quantified by medical statistics – as doing so would mean that the scope of the duty is determined by medical evidence – *Montgomery* had not sufficiently succeeded in clarifying key elements of the two-fold test.¹⁵⁰ The first limb of the test – whether a reasonable person in the patient's position, if warned of the risk, would be likely to attach significance to it – was objective; this brought focus to the requirement of a reasonable or ordinary person, in the position of the patient.¹⁵¹ The second limb – if the HCP is or should reasonably be aware that the particular patient, if warned of the risk, would be likely to attach significance to it – was subjective; this limb recognised that a patient may not be reasonable, and that the court has discretion to assess the position of the particular patient, in conjunction with, "*their requirements and fears, whether reasonable or unreasonable*".¹⁵² The decision in *Rogers*, which provided guidance to the judiciary in

¹⁴⁷ Ibid.

¹⁴⁸ Michael Lamb, (n140), 25.

¹⁴⁹ Charles Foster, (n138), 9

¹⁵⁰ *Montgomery* (n126), 89.

¹⁵¹ Ibid.

¹⁵² Ibid, 163.

Montgomery to create this revised test of materiality, was inappropriately narrow in its assessment of ‘the patient’s position’.¹⁵³

The court carved out a limited therapeutic exception, to justify the possible non-disclosure of material risks.¹⁵⁴ A range of scenarios were highlighted, which justified non-disclosure of material information: emergency situations, where the patient did not want to be informed (raising issues around capacity); or where the HCP determined that disclosure of material information would cause the patient serious physical or mental harm; which we know as the therapeutic privilege exception. Lords Kerr and Reed reminded the courts that this exception should not be abused, and is only applicable in very limited circumstances.¹⁵⁵ While *Montgomery* acknowledged that these scenarios could justify non-disclosure of material information, the judiciary reminded the court that these were *exceptions* to the general duty of disclosure, as opposed to justifications for non-disclosure.¹⁵⁶

This decision confirmed core elements and legal principles, to effectively deliver informed consent for treatment and care: the provision of sufficient information; supporting patient understanding and choice, pertaining to the proposed care management plan; maintaining an open and honest dialogue of communication between patient and professional; supporting patient understanding of the ‘risks’ associated with the proposed treatment(s), and of available alternative methods; and ensuring that patients are aware of any ‘reasonable’ alternative options, associated with proposed treatment and care plan.

Indeed, *Montgomery* promoted patients’ rights and autonomy; however, its impact in medical practice is said to be overstated.¹⁵⁷ Chan *et al.* note that it is difficult to identify any significant change to medical practice on the non-disclosure of information, pre-and post-*Montgomery*.¹⁵⁸ In this regard, the decision is said to merely confirm the General Medical Council’s (GMC) *existing* standards and protocols on good practice and patient choice/autonomy, making little, if any, difference to the practices of HCPs.¹⁵⁹ This suggests

¹⁵³ Louis V. Austin (n143), 345.

¹⁵⁴ *Montgomery* (n126), “The doctor is however entitled to withhold from the patient information as to a risk if he reasonably considers that its disclosure would be seriously detrimental to the patient’s health”, at para 88.

¹⁵⁵ *Ibid*, “... it is not intended to subvert that principle by enabling the doctor to prevent the patient from making an informed choice ...”, 91.

¹⁵⁶ Louis V. Austin, (n143), 347.

¹⁵⁷ Michael Lamb, (n140), 30.

¹⁵⁸ Sarah Chan and others, ‘*Montgomery and Informed Consent: Where Are We Now?*’ (2017) 357 *BMJ* 2224.

¹⁵⁹ *Ibid*.

that ethical standards, in the medical profession, have “long been higher than those required by law”,¹⁶⁰ spotlighting the law’s delay in reflecting the now rooted patient-autonomy ethos of the profession.

Lamb highlights that, as medical standards are imposed through a process of self-regulation by the medical regulatory bodies themselves (GMC Guidance on Consent), it is questionable whether *Montgomery* is likely to have *any* impact on the practice of HCPs for information disclosure, or merely bring the law into line with existing clinical guidelines on consent.¹⁶¹ He argues that, in this light, a significant concern is that *Montgomery* confirms that the law is typically “too blunt an instrument”, and is “too far removed from the practical realities” to satisfactorily influence the behaviours of HCPs.¹⁶² However, this is rather misconstrued evaluation. Litigation is never irrelevant to self-regulation, as the latter needs to satisfy the requirements of law; it does not replace the law. In some circumstances, the law is blunt instrument, as it only comes into play where acceptable boundaries have been crossed, but it does not render it redundant.

Nevertheless, it would be naïve to assume that the courts have achieved an appropriate balance between patients’ and professionals’ plural interests, in the context of assessing duty for consent. The courts battled to set an appropriate standard, blending, mixing and transplanting, both domestic and trans-jurisprudential legal principles, in an attempt to cultivate a refined and balanced standard.

Scholars argue whether the pendulum has swung too far in favour of patients’ rights, creating a power imbalance in terms of patient and professional interests.¹⁶³ Indeed, recent examples of case law demonstrate the complexity of delineating patient and professional interests, in the context of protecting autonomy and shared decision-making.¹⁶⁴ Judicial focus is also beginning to turn to establishing whether ‘reasonable’ systems are in place to effectively secure patient consent, accounting for the plural interests of patients (to deliver consent), and professionals (discharging their duty to secure it).

¹⁶⁰ Michael Lamb, (n140), 27.

¹⁶¹ *Ibid.*

¹⁶² *Ibid.*, 28.

¹⁶³ *Ibid.*

¹⁶⁴ *ABC v St George’s Healthcare NHS Foundation Trust* (2017) EWCA Civ 336.

1.3 Obtaining Informed Consent for Trisomy Screening – *Mordel v Royal Berkshire NHS Foundation Trust*¹⁶⁵

Mordel was a highly publicised and divisive case; not least due to its ‘wrongful birth’ label, which often stirs emotion among the general public. While the media and Down’s Syndrome (DS) campaign groups appropriated the decision, as being symbolic of “discrimination” against this community, it ultimately exposed the vulnerability of HCPs for effectively securing consent, across the DS screening pathway. While *Montgomery* was cited, in this case, to reiterate the significance of patients’ rights, and the importance of providing ‘sufficient information’ before securing consent, *Mordel* should also be understood as unearthing systemic disconnects and frailties for securing consent. Indeed, the primary focus of the court, in this case, was on whether ‘sufficient information’ was being imparted by HCPs, *within* a ‘reasonable system’ for securing parent consent.

In 2015, a first-time Mum gave birth to a baby, Aleksander Mordel, who was born with DS. Mrs Mordel sought damages, from the Trust, for negligently depriving her of the opportunity to have screening for the condition which, had the opportunity not been missed, would have resulted in termination.

Mrs Mordel left her booking appointment (23rd of June 2014) under the impression that she had accepted “all six” methods of screening, including the combined screening test, commonly used in the first trimester. Records demonstrated that the Claimant was booked in, by her midwife, to have her nuchal translucency (NT) measurement taken, as part of the combined screening, one month following her initial appointment (22nd of July 2014). Upon her arrival to the ultrasound appointment, to perform the scan (otherwise known as the ‘dating scan’), the sonographer reported the Mrs Mordel had *declined* the DS screening, on the basis that she did not want her NT measurement taken; this was recorded by the sonographer on the computerised ‘dropdown’ option box. While an overall ‘health check’ of the baby was still performed, it was also noted that the Claimant had declined having her bloods taken, forming another part of the combined test. Thus, it is important to note that *both* elements of the combined test were reportedly declined: the taking of the blood, and the performance of the NT measurement.

¹⁶⁵ *Mordel v Royal Berkshire NHS Foundation Trust* (2019) EWHC 2591.

A month following the ultrasound appointment (11th of August 2014), an appointment was arranged to see the midwife. Proper practice mandated that, if the parent did not have the combined test – as was Claimant’s position, in this case – an offer of the ‘quadruple’ test should have been discussed with the midwife; a second trimester blood test, that targets DS only, at 16-weeks’ gestation. The Claimant underwent the 20-week fetal anomaly scan; however, this appeared unremarkable. It was also noted that the fetal anomaly scan is *not* recommended to screen for DS, due to its inaccuracy in this regard. Following a caesarean section, baby Aleksander was born safely.

Jay J had the task of assessing whether consent had been delivered for screening and, if so, what decision would the Claimant have made, if they discovered the presence of DS. This required the court to consider whether sufficient information had been delivered by the HCPs, in anticipation of the parent delivering consent, and whether reasonable systems were in place for HCPs to effectively secure it. Jay J underlined the significance of *Montgomery*, in the context of patients being the bearers of rights, and that ‘sufficient information’ must be provided to patients, to inform consent. The court also underlined case law pertaining to the significance of establishing reasonable systems for securing consent, namely *ARB v IVF Hammersmith*.¹⁶⁶

Jay J methodically constructed six key issues for consideration, in his application and assessment of the law: (i) (a) did the sonographer offer Down’s Syndrome screening on the 22nd of July 2014; and, if so, what exactly did she say? (b) did the Claimant appear to decline the offer; and, if she did, what exactly did she say? (ii) did the sonographer discharge her duty to the Claimant, in terms of obtaining the latter’s informed consent? (iii) if the answer to (i)(a) and (b) is ‘yes’ and (ii) is ‘no’, was it in fact the Claimant’s wish not to undergo Down’s Syndrome screening on this occasion? (iv) did the midwife discharge her duty to the Claimant, on the 11th of August 2014, in not exploring why the combined test had not been carried out? (v) if the answer to (iv) is ‘no’, would the Claimant have informed the midwife pursuant to the exploration, that *ex hypothesi*, the latter should have been conducted that she wanted Down’s syndrome screening (i.e the quad test)? (vi) In the event that the answer to (iii) is ‘no’ and/or to (v) is ‘yes’ (on the assumption that either or both of these questions arise), would

¹⁶⁶ *ARB v IVF Hammersmith* (2018) EWCA 2803.

the Claimant have consented to invasive testing, and a termination of the pregnancy? While a more in-depth exploration of these issues will be considered in the discussion chapter, it is necessary to map the foundation of the decision and the reasoning of the court, in this regard.

Understandably, as judges are ultimately laypersons in these contexts, Jay J relied heavily on clinical guidelines, throughout the assessment of these issues; predominantly NICE, the 2007 Antenatal Screening Working Standards for Down's Syndrome screening (national standards at the time, preceded FASP trisomy guidelines), and local policy standards (those established under the Royal Berkshire Trust) were considered.

On the first issue, the Claimant maintained that no conversation occurred on DS screening, between herself and the sonographer, at the ultrasound appointment; however, the sonographer contended that the Claimant declined screening, evidenced by the exercise of selecting the 'dropdown decline' option, on the computerised system. As a failure to introduce *any* discussion on DS screening at the appointment would have amounted to a gross breach of duty by the sonographer, Jay J found in favour of the Defendant on this issue, explaining that this was likely to be an issue pertaining to the recollection of the Claimant.

Evaluation of the second issue was far less straightforward. Ultimately, this issue rested on the assessment of established systems for securing consent, and locating where the duty to obtain consent truly lay. "Do you want the Down's Syndrome screening" was deemed an insufficient and ambiguous means of broaching the conversation of screening by the sonographer, according to the expert witness for the Claimant. Indeed, the expert explained that some women enter the ultrasound appointment under the misapprehension of having the 'needle test' (amniocentesis), and thus risking a misunderstanding between combined testing (screening), and invasive testing (diagnostic). However, the expert witness for the Defendant reminded the court that it was not the role of the sonographer to re-counsel the parent, nor provide further information.¹⁶⁷

Another area of conflict pertained to whom obtained consent: whether this be the duty of midwife or sonographer. The expert for the Claimant explained that the midwife, at the booking appointment, 'goes through the issues' in significant detail, and obtains consent at that moment in time: the sonographer's role is limited to confirming whether consent is

¹⁶⁷ Mordel (n165), 70.

forthcoming on the day of the ultrasound appointment.¹⁶⁸ However, the expert for the Defendant contended that the parent does not provide informed consent at the booking appointment, rather this amounts to an ‘informed offer’: it is the duty of the sonographer to obtain consent, at the ultrasound appointment itself.¹⁶⁹ A change of mind, according to this expert, was not rare between the booking appointment and ultrasound scan; this was the justification for his practice of obtaining consent, at the ultrasound appointment.¹⁷⁰

The task for Jay J, applying *Bolitho’s* assessment of reasonableness to the question of established systems, was deciphering whether the sonographer’s practice was irresponsible, unreasonable and unrespectable, if not illogical, in light of the duty to take reasonable steps to secure informed consent.¹⁷¹ Informed consent is a fundamental principle of the modern NHS, according to Jay J, and that NHS guidelines, such as NICE, should not be prescriptive of how to secure it.¹⁷² A ‘gentle exploration’ of the parent’s state of mind is required, according to Jay J, for the purpose of checking parent understanding and choice, fortifying the principles of self-determination and autonomy; this was consistent with sections 7 and 8 of the National Standards, at that time.¹⁷³

Jay J agreed that the system works on the basis that the midwife informs the parent at the booking appointment; however, he disagreed that the sonographer’s role was confined to checking that the parent’s decision, from the booking appointment, was forthcoming on the day of the ultrasound 12-week scan.¹⁷⁴ Informed by clinical guidelines and expert witness evidence, Jay J’s assessment of a ‘reasonable’ system required the sonographer, to satisfy herself, that the patient is consenting to the procedure, either with or without the NT, before it is undertaken, on the basis of proper information; this would mean that her consent is ‘informed’.¹⁷⁵ This system works on the basis of:

‘(i) checking that there has been a discussion between patient and midwife; (ii) checking that the patient has been supplied with the NHS booklet; and (iii) ascertaining by brief questioning

¹⁶⁸ Ibid, 62.

¹⁶⁹ Ibid, 74.

¹⁷⁰ Ibid, 76.

¹⁷¹ Ibid, 84.

¹⁷² Ibid, 85.

¹⁷³ Ibid.

¹⁷⁴ Ibid, 89.

¹⁷⁵ Ibid.

that the patient understands the essential elements and purposes of scanning for Down's Syndrome'.¹⁷⁶

Upon assessment and measurement of a reasonable system, Jay J concluded that the established system was inadequate, finding against the sonographer in this regard.

The third issue reads uncomfortably, turning focus to the credibility of the Claimant. Council for the Defendant submitted that the Claimant bitterly regretted her change of mind, and that she persuaded herself that events happened as she wished, distorting her reality of the experience.¹⁷⁷ This did not sit comfortably with Jay J, concluding that given the importance of the decision, it was unlikely that she would have persuaded herself of the events, misremembered or forgotten it.¹⁷⁸ The court found in favour of the Claimant, holding that, in the heat of the moment, the Claimant did not process the opening question properly, and the sonographer's actions were consistent with the NT measurement being taken.¹⁷⁹

Focussed turned to the conduct of the midwife at the 16-week appointment, to assess the fourth issue. The question, for Jay J, was whether reasonable practice mandated that the midwife check, at the appointment on the 11th of August, why the Claimant had not undertaken the combined test, after being booked in to have it. The expert witness for the Claimant relied on NICE guidelines, pertaining to the practice of midwives at the 16-week appointment. The court probed the expert's perception of the NICE guidelines; the expert agreed that sections 7 and 8 of the guidelines were in play, promoting respect for parent choice.¹⁸⁰ The expert also contended that asking a limited number of open questions, at this stage, would not impede or interfere with the Claimant's autonomy, and freedom to choose.¹⁸¹

On the same issue, the experts for the Defendant rebutted the above point, and affirmed that the midwife's duty is triggered by the parent undertaking the screening test; in this case, the Claimant declined it.¹⁸² These experts added that there was no duty, on the midwife, to ask

¹⁷⁶ Ibid.

¹⁷⁷ Ibid, 102.

¹⁷⁸ Ibid, 103.

¹⁷⁹ Ibid, 113.

¹⁸⁰ Ibid, 121.

¹⁸¹ Ibid.

¹⁸² Ibid, 126.

further questions; their experience of practice was that questioning resulted to complaints by parents, ending up with invasive testing they did not want.¹⁸³ They concluded that National Standards – referring to the 2007 guidelines – placed emphasis on respecting the parent’s right to choose, and that revisiting the decision to not have testing, runs the risk of making the parent feel they have made the wrong choice, and/or creating pressure to change their decision.¹⁸⁴

Applying *Bolitho* to the question of systemic reasonableness, Jay J concluded that, while it cannot be incumbent on the midwife to undertake lengthy inquiry – where the parent was booked to have screening and later declined it – the matter should not have been left there.¹⁸⁵ A reasonable system required the midwife to take reasonable steps to explore and check whether the decision made – to not have screening, in this case – was in accordance with the parent’s wishes, placing the Claimant at the core of the decision-making process.¹⁸⁶ It was also incumbent on the midwife to check that “*everything has gone and is continuing to proceed according to plan*”, underlining the significance of a continued dialogue between parent and professional, and the dynamic nature of decision-making and consent in this regard.¹⁸⁷

Jay J briefly concluded that, given the ruling for the third issue, the answer to the fifth issue was ‘yes’, as no evidence was presented to undermine the fact that the Claimant declined DS screening at the sonographers appointment, and subsequently had a change of mind.¹⁸⁸

The sixth and final issue concerned causation. In a situation where no breach(es) had occurred, Jay J concluded that, on the balance of probabilities, it is probable that the Claimant would have undertaken invasive testing had they been told of a higher-risk DS result, and would have opted to terminate, due to her young age.¹⁸⁹

The decision reinforced the core principles from *Montgomery* on securing parent consent: ensuring provision of ‘sufficient’ information; supporting patient understanding and choice;

¹⁸³ Ibid, 129.

¹⁸⁴ Ibid, 129.

¹⁸⁵ Ibid, 136.

¹⁸⁶ Ibid.

¹⁸⁷ Ibid.

¹⁸⁸ Ibid, 142.

¹⁸⁹ Ibid, 152.

ensuring effective and open communication between patient and professional; and supporting patient understanding of available alternative options, to treatment and care.

Mordel established, primarily, that consent is an ongoing, dynamic process: it is not a ‘one-off’ event that occurs at the booking appointment. In addition, the decision has underlined that an assessment of informed consent also extends to consideration of establish systems for securing it, and whether said systems are indeed *Bolitho* ‘reasonable’.

This case also conveyed the current disconnect in systems and processes for obtaining consent, and the ambiguity surrounding HCPs’ duty of sufficiently informing parents of screening’s requisite components. The decision also threw into question the significance of interprofessional and interdisciplinary practices – in this case, midwifery and ultrasonography – and whether it needs enhancing in this regard.

1.4 Framing an Empirical Response to Considerations Raised in *Montgomery* and *Mordel* on Consent for Trisomy Screening

Montgomery and *Mordel* raise significant questions pertaining to the interests of stakeholders, when delivering and securing consent, along the recently established trisomy pathway. These cases spotlight the effectiveness and workability of a shared decision-making model of care, and the sustainability of current systems of consent in clinical practice.

The foremost significance of *Montgomery* rests upon its promotion for protecting patients’ right to self-determination and autonomy, in the context of medical care and treatment. The decision underlined the dynamic nature of consent, and that a model of care based on shared decision-making, is principal to effectively secure it.

Montgomery also isolated the practicality and importance of delivering ‘sufficient’ information to patients in pre-treatment consultations, ensuring that they are not bombarded with complex and technical information.¹⁹⁰ Supporting the patient’s understanding of the information delivered, by promoting open and honest communication between patient and professional, is also required to discharge their duty to secure consent.¹⁹¹

¹⁹⁰ *Montgomery* (n126), 90.

¹⁹¹ *Ibid.*

Supporting patient choice, by entering into a continued dialogue with the professional, was also identified, in *Montgomery*, as key to informed decision-making. Choice was framed as providing patients with the option to consider alternative methods of treatment and care, subject to discussion of material risks associated with proposed treatment plans. Individualising treatment, according to the patient's needs and wishes, was also identified as significant going to the duty of HCPs to secure consent, in *Montgomery*.

While *Mordel* reiterated and generalised these key findings from *Montgomery*, as scaffolding to effectively obtain parent consent for trisomy screening, Jay J threw into question the significance of assessing whether reasonable systems are in place for professionals to secure it. Indeed, *Mordel* exposed the fragility and ambiguity of current systems of consent for trisomy screening, raising significant questions around the *Bolitho* reasonableness of established systems. *Mordel* also threw into question the disconnect between professional roles when operating current systems of consent, and a need to enhance interprofessional practices in this regard.

An opportunity arises to undertake an empirical investigation into the reasoning in *Montgomery* and *Mordel*, as a means to delineate whether the plural interests of both professionals and parents are valued, in the context of delivering and obtaining consent for trisomy screening. Indeed, the focus of the reasoning in *Montgomery* falls upon the protection and consideration of patient interests; that their right to self-determination and autonomy is respected in clinical decision-making. *Mordel*, however, sought to assess broader systemic and practical considerations for professionals to effectively obtain parent consent for screening, exposing possible systemic deficiencies, and the need to enhance interprofessional practice.

The key areas identified, from *Montgomery* and *Mordel*, to delineate parent and professional interests for delivering and obtaining consent, extend to six broad considerations: the provision of information; support for parent understanding; supporting parent decision-making and choice; effective communication and the HCP-patient relationship; supporting parent understanding of the 'risks' associated with screening and testing; supporting parent understanding of alternative methods of treatment and care.

Conducting an empirical investigation may also initiate a dialogue between stakeholders, with a purpose of understanding how parent interests and values, underlined in *Montgomery*, relate to those of the profession; this conversation should also extend to the practical and systemic considerations for securing consent, revealed in *Mordel*, and its impact on professional duty to obtain it.

Mordel also indicated that the interests of individual stakeholders themselves, may also differ. Parent interests are shaped by personal values and perspectives on the provision of screening. Professional interests and values are ultimately dependent on their specific role and involvement along the trisomy pathway. Thus, an empirical exploration into the interests of these stakeholders, may also serve to reveal unique considerations, going to the process of delivering and obtaining consent for trisomy screening.

Chapter 2 – Informed Consent and Trisomy Screening

This chapter will explore the key considerations and areas of interest, identified from *Montgomery and Mordel*, with reference to the significant body of socio-legal and medical research on informed consent for trisomy screening. The historical development of antenatal screening, in England and Wales, will also be outlined. It will also provide a structural review and explanation of the trisomy pathway in England and Wales, and of its each individual components. Clinical guidelines on obtaining informed consent in medical practice, and consent to trisomy screening and testing, will also be outlined in this chapter, to further inform the key considerations and areas of interest identified from chapter 1.

2.1 Antenatal Screening and Testing in England and Wales

The improvement of outcomes, and an increase in reproductive autonomy and choice, have been central to the aims of the healthcare systems, in Western countries, when offering antenatal screening and testing to pregnant women. Over the last forty years, with rapid developments in reproductive genetics and assisted reproductive technology, parents now have the ability to discover information about their babies before birth.¹

It is described as a ‘rite of passage’ for parents, in Western society, to undertake antenatal screening and testing; whereby parents are presented with a series of scans and blood tests, to evaluate the fetal² and maternal health.³ Seeking reassurance on maternal or fetal health, discovering the sex of the baby, taking advantage of the opportunity to meet the baby for the

¹ Jamie S. King, ‘And Genetic Testing for All – The Coming Revolution in Non-Invasive Prenatal Genetic Testing’, (2011) 42 *Rulgers Law Journal* 599, 658.

² Whilst ‘fetal’ is consistent with US usage in common parlance, this is also the preferred current usage in the medical profession. Therefore, I will use ‘fetal’, rather than foetal, in this PhD.

³ Megan Best, “The Dilemma of Prenatal Screening”, *Journal of Ethics & Medicine*, 2018, Vol. 34:2, pp.113-123, at pp.113.

first time, and social pressure to fit the mould of a ‘responsible parent’, are all routinely framed as key motivators for engaging, or indeed disengaging, with antenatal screening.⁴

The spiritual, ethical and moral values of parents are challenged throughout the antenatal screening and testing process.⁵ This could be exacerbated when confronted with a difference⁶ or a complication in pregnancy.⁷ The professional-parent relationship is often tested in this context, requiring HCPs to consider parents’ best interests. This mandates HCPs to demonstrate an appreciation of the burden placed upon the shoulders of parents to make lifechanging reproductive decisions, in a very limited period of time. Indeed, depending on whether the mother has opted to undertake conventional screening, one in twenty women will receive an unexpected result, that their baby could have a biological difference.⁸

According to the latest report released by the Office of National Statistics (ONS), there were 731,213 live births across the United Kingdom, in 2018.⁹ Around 700,000 of these occurred in England and Wales.¹⁰ Of these, the Congenital Anomaly Register and Information Service (CARIS) states that up to 40,000 pregnancies occur in Wales each year.¹¹ It is reported that 74% of pregnant women who have access to the NHS services, across England and Wales, chose to embark on the antenatal screening and testing pathways.¹² The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) reports that, in 2017, across England alone, 6,798 cases with one or more congenital or chromosomal anomalies were

⁴ Kater-Kuipers A., E. M. Bunnik, I. D. de Beaufort and R. J. H. Galjaard, ‘Limits to the scope of non-invasive prenatal testing (NIPT): an analysis of the international ethical framework for prenatal screening and an interview study with Dutch professionals’, (2018) 18 BMC Pregnancy Childbirth, 409.

⁵ C. Lewis, C. Silcock and L.S. Chitty, ‘Non-Invasive Prenatal Testing for DS: Pregnant Women’s Views and Likely Uptake’, (2013) 16 Public Health Genomics 223, 232.

⁶ The term “difference” is used in place of “abnormality” or “affected baby” as these terms are offensive to those living with Down’s Syndrome, Edwards’ Syndrome or Patau’s Syndrome.

⁷ Megan Best (n3), 114.

⁸ Ibid, 113.

⁹ Office for National Statistics, “Vital statistics in the UK: births, deaths and marriages”, released November 2018, available at:

<https://www.ons.gov.uk/peoplepopulationandcommunity/populationandmigration/populationestimates/datasets/vitalstatisticspopulationandhealthreferencetables> (accessed 20/04/2020).

¹⁰ National Institute for Health and Care Excellence (NICE) Guidelines: Antenatal Care Draft Scope for Consultation (2018).

¹¹ Public Health Wales. Congenital Anomaly Register & Information Service (CARIS), at: <http://www.caris.wales.nhs.uk/chromosomal> (accessed 06/06/2020).

¹² Tom Shakespeare and others, ‘Non-invasive prenatal testing: ethical issues’ (2017) Nuffield Council on Bioethics, 2-138, 29.

notified to the NCARDRS, out of 320,031 total births (live and stillborn).¹³ According to NCARDRS, the most commonly detected anomalies are congenital heart anomalies and chromosomal anomalies.¹⁴

2.1.1 Structure of Antenatal Screening and Testing Programmes in England and Wales

The NHS in Wales and England offer all parents the opportunity to undergo antenatal screening and testing. Antenatal screening is commonly used to identify any potential differences in the development and health of the fetus which may need further investigation. The two main pathways, under the national fetal anomaly screening and testing programme, are ‘fetal anomaly’ and ‘trisomy screening’ (see Appendix 1 for a detailed diagram).

A growing body of research and case law, such as *Mordel*, suggests that parent consent for *trisomy screening* is less than informed.¹⁵ Indeed, academics and practitioners are becoming increasingly concerned with the evidence emerging surrounding the current challenges parents face delivering consent for trisomy screening, and the systems in place for HCPs to effectively secure it.¹⁶ For this reason – while there is a degree of overlap between the fetal anomaly and trisomy pathways – the scope of this thesis is focused primarily on the trisomy screening pathway.¹⁷

Trisomy Screening

Trisomy screening and testing is a method of antenatally detecting whether there is a difference in the chromosomal composition of the baby. The most common chromosomal conditions are Down’s Syndrome (DS), Edwards’ Syndrome (ES) and Patau’s Syndrome (PS). This pathway, under the national screening programme, was originally designed to specifically detect DS only; however, a UK-wide recommendation was made by the UK National Screening

¹³ Public Health England. National Congenital Anomaly and Rare Disease Registration Service. Congenital Anomaly Statistics 2017, 2019.

¹⁴ Ibid.

¹⁵ Sophie John and others, ‘A new tool to assess understanding of Down syndrome screening information presented by midwives’, (2019) 27 British Journal of Midwifery 2, 9.

¹⁶ Ibid.

¹⁷ The preferred methods of screening used are different for DS, ES and PS at the later stages of gestation (which mean different policy standards apply). For example, the fetal anomaly scan (18/20-week scan) is used to screen for Edwards’ and Patau’s syndrome (only 5% of those go undetected). On the other hand, the quadruple test (14-18 weeks) is used to screen for Down’s syndrome only. When attempting to screen for Down’s syndrome on the FAS (18/20-week scan), 50% of those go undetected which is far less sensitive. NHS spec no.16 focuses on screening for DS, ES and PS. NHS spec no.17 (fetal anomaly scan) provides standards and guidelines to screen for ES and PS only. There is an overlap between these primary policy standards.

Committee (UK NSC), to extend the traditional DS screening programme to include other common chromosomal anomalies, such as ES and PS.

UK National Screening Committee (UK NSC)

The UK NSC is an independent advisory body that make recommendations, about all aspects of population screening, to government ministers, across all four UK countries. It also supports the implementation of screening programmes. It is important to note that the UK NSC do not create policy, and does not implement the screening programmes; this is the role of the individual countries, which provide screening programmes with the NHS across the UK. The UK NSC comprises of independent experts, which include clinicians, academics and charities.

A review conducted by the UK NSC, in 2014, identified and recommended a host of improvements surrounding the implementation of developing screening programmes.¹⁸ A fundamental recommendation, made by the UK NSC review group, was that a common Code of Practice should be drafted and published to provide information regarding the “status, role, responsibility and procedures of the UK NSC ... the roles and relationships of different organisations, including PHE, the Department of Health in England ... the Welsh Government, the Department of Health, Social Services and Patient Safety ... with the UK NSC and how the Committee develops its recommendations”.¹⁹

The UK NSC reported the response of the Royal College of Obstetricians and Gynaecologists (RCOG) and the British Maternal and Fetal Medicine Society Executive Committee (BMFMS), in 2014, to recommend introducing screening for ES and PS, into the existing DS screening programmes.²⁰ The rationale behind this recommendation was to ultimately enhance the efficiency of screening programmes: DS risk-scores algorithms were capable of also producing risk-scores for ES and PS.²¹ For example, technically, ultrasound and serum markers, to screen

¹⁸ UK National Screening Committee, “Review of the UK National Screening Committee (UK NSC)”, 2015, (accessed 30/05/2019) (available at https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/443953/20150602_-_Final_Recommendations.pdf).

¹⁹ Ibid, 7.

²⁰ UK National Screening Committee: First Trimester Combined Screening for Trisomy 18 and Trisomy 13. Consultation Comments. (2014) 1-24, 19.

²¹ UK National Screening Committee: First Trimester Combined Screening for Trisomy 13 and Trisomy 18 – External review against programme appraisal criteria for the UK National Screening Committee (UK NSC), 5 (2013) 1-57, 36.

for DS in the first trimester, had long been in use, and this method of screening was also suitable for ES and PS detection. Indeed, karyotyping and/or the PCR (see glossary), when analysing high-risk groups for DS, can also detect the presence of ES and PS; therefore, many cases of ES and PS were being detected incidentally prior to the recommendation of the UK NSC, to specifically screen for ES and PS in the first trimester.

In 2016 and 2018, Public Health England (under the Fetal Anomaly Screening Programme)²² and Public Health Wales (under Antenatal Screening Wales)²³ respectively implemented the UK NSC recommendation, to introduce ‘trisomy’ screening to the existing DS pathway (see Appendix for diagram on the screening programme’s implementation). Note, however, that while the UK NSC acknowledged that ES and PS were very different to DS, in terms of their aetiology and prognosis, the implementation of this recommendation – to include screening and testing for ES and PS into existing DS screening pathways – conflated all three conditions under a genetic ‘trisomy’ model, rather than differentiating between individual phenotypes.²⁴ Commentators explain that this was due to promoting efficiency and execution of screening for trisomies.

2.2 The Trisomies

Every human body is constructed by cells. Within the nucleus of the cells, structures are found, called chromosomes. These chromosomes are composed of deoxyribonucleic acid (DNA) and proteins, containing genetic information. Each unit of the chromosome are called genes, which are inherited from the mother or father, or both. Forty-six chromosomes are typically found within the cells of a human body, arranged into twenty-three pairs, originating from the mother and father. Non-disjunction refers to an error which can arise in meiosis, resulting from the chromosomal pairs failing to separate. This may cause the presence of a usual number of chromosomes in the cell, which is referred to as ‘aneuploidy’. Where the cell has an extra chromosome, this is referred to as trisomy, that is, three copies of a particular chromosome rather than the usual two. Three copies of chromosomes 21, 18 or 13 will result in the baby having either DS, ES or PS.

²² FASP is an England-focussed screening programme.

²³ ASW is a Welsh-focussed screening programme.

²⁴ UK National Screening Committee (n21), 36.

Down's Syndrome

Mr John Langdon Down, over 150 years ago, identified common characteristics and features between a particular group of patients he was caring for at the time; these patients were short in stature, had thicker necks than usual, and a flatter skull.²⁵ He tailored the term “mongolism” to describe the group; however, this is recognised to be highly offensive and politically incorrect.²⁶ Due to Down's personal beliefs, DS was believed to be a racial defect for over a century.²⁷ At this period, people with DS were subjected to prejudice and discrimination, not only by society, but also the scientific community.²⁸ This prejudice also extended to the parents of a child with DS, with the general public condemning them for causing the condition in their child, by being alcoholics or from carrying sexually transmitted diseases.²⁹

Historically, a narrative existed that HCPs believed babies with DS could not achieve the common physical and intellectual milestones.³⁰ Persons born with DS were institutionalised, and could not attend mainstream education.³¹ Today, a high number of people with DS attend mainstream primary education (around 80%), and are also raised by their parents, rather than being institutionalised.³²

DS occurs in around 1 in every 1000, or 0.1%, of pregnancies.³³ According to the NDSCR for England and Wales, in 2013, 1,872 diagnosis of DS were made antenatally, with 717 live births.³⁴ CARIS states that an average of 78 cases of DS are reported annually in Wales.³⁵ A baby with DS may have some level of physical and learning differences. They may have

²⁵ Harold Ellis, 'John Langdon Down: DS', (2013) 23 *Journal of Perioperative Practice* 296, 297.

²⁶ Ibid.

²⁷ Marianna Karamanou and others, 'Jerome Lejeune (1926-1994): Father of Modern Genetics', (2012) 10 *Acta medico-historica Adriatica*, 311.

²⁸ Ibid.

²⁹ Ibid, 312.

³⁰ Barbara Barter and Richard Hastings, 'Consultation with Individuals with Down syndrome about Non-Invasive Prenatal Testing', (2017) *Mencap*, 7.

³¹ David Patterson and Alberto C. S. Costa, 'Down syndrome and genetics – a case of linked histories', (2005) 6 *Nature Reviews. Genetics*; London, 137.

³² Tom Shakespeare (n 12), 3

³³ Morris JK and Springett A. 'The National Down Syndrome Cytogenetic Register for England and Wales 2013 Annual Report'. Queen Mary University of London, Barts and The London School of Medicine and Dentistry (2014), 2

³⁴ Ibid, 2.

³⁵ Public Health Wales. Congenital Anomaly Register & Information Service (CARIS), at: <http://www.caris.wales.nhs.uk/chromosomal> (accessed 06/06/2020).

communication challenges, and difficulty managing some everyday tasks. Some health problems are more common in people with DS. These include heart conditions, and problems with hearing and vision.³⁶ However, due to revolutionary developments in medical science and technology, babies born today, with DS, have an ever-increasing life expectancy and commonly live happy, healthy lives, some well into their 60-70s.³⁷

Edwards' Syndrome and Patau's Syndrome

ES is the second most common autosomal trisomy syndrome behind DS.³⁸ ES, due to its rarity, was first described in 1960, by Edwards *et al.*³⁹ ES occurs in around 0.067% of pregnancies.⁴⁰ According to the NDSCR for England and Wales, in 2013, 473 diagnosis of ES were made antenatally, with 33 live births.⁴¹ CARIS states that an average of 21 cases of ES are reported annually in Wales.⁴² All women have a chance of having a baby with ES.⁴³ Babies born with ES commonly have a range of physical and learning differences.⁴⁴ They may have problems with their heart, respiratory system, kidneys and/or digestive system. Despite the increase standard of care due to medical and technological advances, almost 33-66% of fetuses with ES will not survive the full gestation period, and those that do survive, 50% will not live past the first week.⁴⁵ The survival rates are low, and of those babies born alive, only around 9-11% survive to hospital discharge.⁴⁶ Studies reveal, however, some babies may live until adulthood.⁴⁷

³⁶ In England and Wales, around 60% of children born with DS will have a heart defect, requiring around 30% of them to have an operation. Sadly, leukaemia is common in children with DS, requiring around 1 in every 200 needing treatment for this. In England and Wales, around 90% of those children born with DS will live past their 5th birthday.

³⁷ David Patterson (n31), 137.

³⁸ UK NSC. 'First Trimester Combined Screening for Trisomy 13 and Trisomy 18. External review against programme appraisal criteria for the UK National Screening Committee (UK NSC)', (2013) 5, 1-57.

³⁹ Angela I. Taylor, 'Autosomal Trisomy Syndromes: A Detailed Study of 27 Cases of ES and 27 Cases of PS', (1968) 5 J. Med. Genet, 277.

⁴⁰ Tom Shakespeare (n 12), 4.

⁴¹ Morris JK and Springett A, (n33), 2.

⁴² Public Health Wales. Congenital Anomaly Register & Information Service (CARIS), at: <http://www.caris.wales.nhs.uk/chromosomal> (accessed 06/06/2020).

⁴³ Antenatal Screening Wales, 'Information for women offered further tests for suspected chromosomal conditions', (2018), 1-35.

⁴⁴ Ibid.

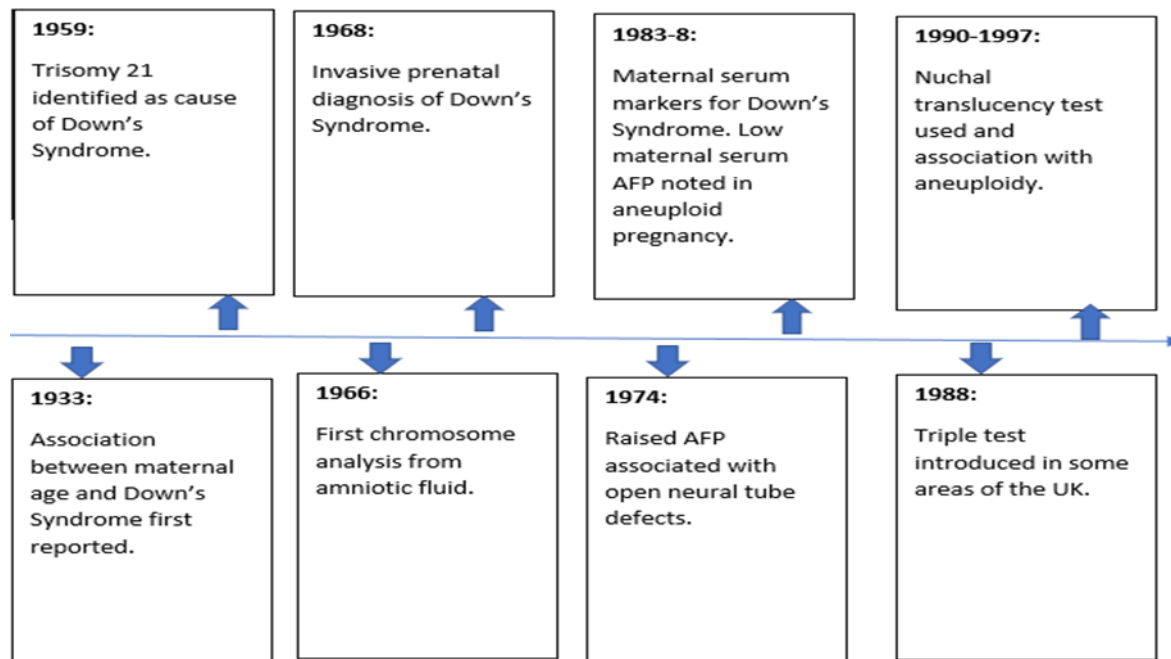
⁴⁵ Tom Shakespeare (n12), 6.

⁴⁶ Boghossian N.S and others, 'Mortality and morbidity of VLBW infants with trisomy 13 or trisomy 18', (2014) 133 Pediatrics 226, 235.

⁴⁷ Wu, J, Springett, A and Morris J.K, "Survival of trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome) in England and Wales", Am J Med Genet, 2013, 2512-2518.

PS is the third most common autosomal trisomy syndrome, behind ES and DS. It was first described by Patau *et al.*, in 1960.⁴⁸ PS occurs in around 0.025% of pregnancies.⁴⁹ According to the NDSCR for England and Wales, in 2013, 176 diagnosis of PS were made antenatally, with 18 live births.⁵⁰ CARIS states that an average of 8 cases of PS are reported annually in Wales.⁵¹ Again, all women have a chance of conceiving a baby with PS.⁵² Babies with PS may also have a range of physical and learning differences. They may have problems with their heart, respiratory system, kidneys and/or digestive system. Around half of babies with PS will have a cleft lip and palate. Babies with PS may have a low birthweight. The survival rates are low, and of those babies born alive, only around 9-11% survive to hospital discharge.⁵³ However, some babies may live until adulthood.⁵⁴

2.3 Methods of Screening and Testing for Detecting a Trisomy



Over the past century, an expansion in reproductive technology has revolutionised antenatal screening and testing, across the world. However, to appreciate the advancements in trisomy

⁴⁸ Angela I. Taylor, "Autosomal Trisomy Syndromes: A Detailed Study of 27 Cases of ES and 27 Cases of PS", *J. Med. Genet* (1968). 5, 277.

⁴⁹ Tom Shakespeare (n12).

⁵⁰ Morris JK and Springett A, (n33), 2.

⁵¹ Public Health Wales. Congenital Anomaly Register & Information Service (CARIS), at: <http://www.caris.wales.nhs.uk/chromosomal> (accessed 06/06/2020).

⁵² Antenatal Screening Wales, (n43)

⁵³ Boghossian N.S and others, (n46), 230.

⁵⁴ Wu, J, Springett and A, Morris J.K, (n47), 2516.

screening technology – and indeed the demand on HCPs to keep pace with the scientific developments, for the purpose of provision in counselling and consent – a brief history of the antenatal screening and testing landscape is required.

A century after John Langdon Down’s first characterisation of DS, a French paediatric doctor, Jerome Lejeune, revolutionised this field by discovering the scientific basis for what caused the condition.⁵⁵ Doctor Jerome Lejeune is commended for discovering that DS is caused by a chromosomal aberration.⁵⁶ In an attempt to “cure” those with DS, he discovered that those with the condition had the presence of a supernumerary chromosome, 47 as opposed to 46 chromosomes.⁵⁷ This discovery in chromosomal differences in patients paved an innovative pathway to modern clinical cytogenetics⁵⁸ and human pathology.⁵⁹

A decade following the discovery of the scientific basis of what causes the condition, medical advancements led to the routine use of the amniocentesis test, where a fine needle is inserted into the mother’s abdomen to extract fluid (amniotic fluid) from around the fetus.⁶⁰ This fluid contained the shed cells of the baby, and could subsequently be analysed in laboratories to see if they contained 47 chromosomes.⁶¹ This was the *first* method that existed of antenatally diagnosing DS.

In the 1980’s, women were having a blood test at 16-weeks’ gestation to measure a protein called alpha-fetoprotein (AFP).⁶² A correlation was discovered that when a woman’s alpha-fetoprotein levels, in the blood, were very low, this placed them in a higher-risk category of conceiving a child with DS.⁶³ This discovery led to the practice of extracting maternal serum

⁵⁵ Marianna Karamanou and others (n27), 312

⁵⁶ Ibid.

⁵⁷ Ibid. Note; a sad irony surrounded the career of doctor Lejeune. Whilst he dedicated his life to try to help those with the condition, he was instrumental in the eventual increase of terminations of those babies with DS following an antenatal diagnosis of the condition. He later opposed abortion, forming an antiabortion group named “Laissez-les vivre (Let Them Live)”, after discovering that clinicians had a tendency of recommending terminations to prevent the birth of a child with DS.

⁵⁸ Cytogenetics is a term used to refer to a discipline of medical practice regarding the study of chromosomes.

⁵⁹ Ibid, 314.

⁶⁰ Kypros Nicolaidis, ‘Screening for fetal aneuploidies at 11 to 13 weeks’. (2011) 31 Prenat Diagn, 2011, 7–15.

⁶¹ Ibid.

⁶² Ibid.

⁶³ Ibid.

to accurately assess whether that mother was of increased risk (along with measuring other markers), rather than relying on the less accurate 'age' test.⁶⁴

It was later discovered that the chemicals in the mother's blood, who carried a child *without* the condition, were different to those mothers who carried a child *with* the condition.⁶⁵ At this period of time, 'screening' was the practice of combining particular hormones, to produce a risk-score: alpha-fetoprotein, human chorionic gonadotropin (HCG), estriol and inhibin A, which are all produced by the placenta.⁶⁶ This gave rise to the double test, triple test and now the quadruple test.⁶⁷ These tests would identify the higher-risk population, and then these women would be offered amniocentesis for a definitive diagnosis. Note that the detection rate at this period was 60%, with a false positive rate of 5%.⁶⁸

In 1991, clinicians discovered – whilst conducting routine ultrasound scans at 12-weeks – a small black area of fluid at back of the baby's neck.⁶⁹ Clinicians discovered that there was a relationship between the pocket of fluid at the back of the baby's neck, and the likelihood that the baby had DS; known today as nuchal translucency (NT).⁷⁰ Women with an enlarged NT measurement were then correlated with the higher-risk category.⁷¹

In the years following this discovery, more blood was taken from the mother at 12-weeks' gestation, and clinicians realised there were two instrumental hormones that, if measured and assessed concurrently, could calculate the risk of DS in a pregnancy: HCG and PAPP A (pregnancy-associated plasma protein A).⁷² The age of the mother, the nuchal translucency, and the two hormones were combined (the combined test), to identify whether there

⁶⁴ Nicholas J Wald and others, 'Maternal serum screening for DS in early pregnancy'. (1998) 297 BMJ, 883-887.

⁶⁵ Ibid.

⁶⁶ Ibid., 885.

⁶⁷ H. Vandecruys, S. Faiola, M. Auer, N. Sebire and K. H Nicolaidis, "Screening for Trisomy 21 in Monochorionic Twins by Measurement of Fetal Nuchal Translucency Thickness", (2005) 25 Ultrasound Obstet Gynecol, 551, 553.

⁶⁸ Nicholas J Wald and others, (n61), 886.

⁶⁹ Kypros Nicolaidis, (n60), 13.

⁷⁰ Kypros Nicolaidis, "Nuchal translucency and other first-trimester sonographic markers of chromosomal abnormalities", Am J Obstet Gynecol, 2004, 191, 45-67.

⁷¹ Ibid.

⁷² Kypros Nicolaidis, Frank Chervenak, Laurence McCullough, Kyriaki Avgidou & Aris Papageorghiou, "Evidence-based obstetric ethics and informed decision-making by pregnant women about 122 invasive diagnosis after first-trimester assessment of risk for trisomy 21", 2005, American Journal of Obstetrics and Gynecology, 193, 322-326.

was a higher-risk. The ‘accuracy’ of this test was 90%. This also reduced the number of women who were subjected to unnecessary invasive tests, from 5% to 3%.

The purpose of mapping the historical expansion of trisomy screening technology was to underline the continuing demand on HCPs to adapt, modify and implement effective systems and provision to support parent decision-making and consent, in a rapidly developing area of medicine. It also demonstrates the increasing complexity of information parents are required to obtain to make an informed decision for trisomy screening.

Indeed, these concerns recently came to light in *Montgomery* and *Mordel*, which threw into question the dynamic, shared decision-making model of consent, and parent and professional interests for delivering and securing consent for trisomy screening. *Mordel*, in particular, provided a timely illustration of the urgent need to explore consent for trisomy screening, in particular, whether reasonable systems and sufficient provision of support were implemented, to secure parent consent along the screening pathway.

2.4 Devolution of Health Law-Making Powers and Divergences Between England and Wales

For the purposes of understanding the landscape of health law-making powers in the United Kingdom, it is necessary to outline the significance of devolution and divergence between England and Wales, and its potential impact on the implementation and execution of NHS antenatal screening policy.

Devolution is described as “the creation of autonomous, elected, governments for Northern Ireland, Scotland and Wales”.⁷³ Greer and Trench stipulate that “1999 marked a major constitutional and policy change for the UK, with the advent of political devolution and the creation of the National Assembly for Wales, the Scottish Parliament and the Northern Ireland Assembly.”⁷⁴ Indeed, new legislative bodies were established in Wales, Scotland and Northern Ireland, and the “exercise of executive power was transferred to administrations formed from

⁷³ Scott L. Greer, ‘Devolution and health in the UK: policy and its lesson since 1998’, (2016) 1 British Medical Bulletin 16.

⁷⁴ Scott L. Greer and Alan Trench, ‘Intergovernmental relations and health in Great Britain after devolution’, (2010) 38 Policy and Politics 509.

the political representatives elected to them”.⁷⁵ Each nation took over responsibility for a large part of spending and responsibilities in their part of the UK, and possessed the freedom to make distinctive policies in areas, including health.⁷⁶ At this period in time, a divergence in health law and policy emerged across the United Kingdom.⁷⁷

Since its conception, Peckham *et al.* argue that there have always been organisational distinctions and divergency, in the delivery of healthcare services across England, Northern Ireland, Scotland and Wales; however, there was “a strong family resemblance between the four sub-systems”.⁷⁸ Similarities were defined as possessing common systems for patient care, the supply and demand of healthcare, and convergent policy goals and objectives.⁷⁹

Devolution is also described as being a “complex phenomenon that brings into play both divergent and convergent influences.”⁸⁰ Devolution was largely welcomed, as it provided an opportunity to create “distinctive and innovative policies”, and health was deemed to be “one of the most significant areas to be devolved”.⁸¹ Smith and Hellowell remark that this makes health policy “of central interest to those interested in the impact of devolution.”⁸² As devolution is said to provide context for greater diversity, due to its scope for experimentation and differing emphasis on policy, Peckham *et al.* suggest that “the sharing of policy developments between one country and another” may also occur; otherwise known as ‘policy transfer’.⁸³

Conversely, Greer pointed to the public acceptance of divergent policies, which suggested “very little support” for divergent health policies and outcomes.⁸⁴ Early concerns emerged

⁷⁵ Kevin J. Woods, ‘Political Devolution and the Health Services in Great Britain’, (2004) 34 *International Journal of Health Services* 323; see also, David S Moon, Jennifer Thompson and Sophie Whiting, ‘Lost in the Process? The impact of devolution on abortion law in the United Kingdom’, (2019) 21 *The British Journal of Politics and International Relations* 728, at 730.

⁷⁶ Greer and Trench (n74), at 509.

⁷⁷ Woods (n75), at 323.

⁷⁸ Stephen Peckham, Nicholas Mays, David Hughes, Marie Sanderson, Pauline Allen, Lindsay Prior, Vikki Entwistle, Andrew Thompson and Huw Davies, ‘Devolution and Patient Choice: Policy Rhetoric versus Experience in Practice’, (2012) 46 *Social Policy and Administration* 199, at 200.

⁷⁹ *Ibid*, 213.

⁸⁰ *Ibid*, 201.

⁸¹ Katherine Smith and Mark Hellowell, ‘Beyond Rhetorical Differences: A Cohesive Account of Post-devolution Developments in the UK Health Policy’ (2012) 46 *Social Policy and Administration* 178, at 179.

⁸² *Ibid*, at 179.

⁸³ Peckham *et al.* (n78), 201.

⁸⁴ Scott L Greer, ‘Devolution and health policy in the UK’, (2008) 14 *Eurohealth* 22, at 24.

surrounding inequality of provision, political friction and the wellbeing of intergovernmental relations.⁸⁵

This period marked a shift in momentum, with the Welsh legislature moving toward obtaining (limited) autonomy and capacity in this respect.⁸⁶ Post-1998, an expectation existed that the newly autonomous systems would continue as variations to English policy; however, Welsh health policy was viewed as a “bilingual copy” of English policy, with distinctions between the English and Welsh systems being ultimately marginal.⁸⁷

For long periods, Wales was viewed as an English region, in the context of devolved services.⁸⁸ Indeed, with reference to the educational system in Wales, an entry was made in the Encyclopaedia Britannica stating: “for Wales, see England”.⁸⁹ Prior to devolution, the NHS in Wales was perceived as “forming an adjunct to the English health service”.⁹⁰

The allocation of powers to the four UK nations and jurisdictions sought to increase divergence in healthcare policy.⁹¹ Hale *et al.* note that “the devolved nations/jurisdictions can be seen as regulatory pioneers, with policy isomorphism where the regulatory ‘natural experiment’ is seen as successful”.⁹²

The Government of Wales Act (2006) provided the Welsh Assembly its own “primary law-making powers for the first time”.⁹³ Indeed, Greer, in 2009, affirmed that, “the four systems are heading in different directions, and in so far as policy affects the work of health systems it is turning them into four different working environments with ever more distinct cultures”.⁹⁴ More recently, the Public Health (Wales) Act (2017) provided scaffolding for public bodies to “carry out health impact assessments and impose a duty upon Welsh

⁸⁵ Greer and Trench (n74), 511.

⁸⁶ John Harrington, Barbara Hughes Moore and Erin Thomas, ‘Towards a Welsh health law: devolution, divergence and values’, (2021) 72 NILQ 65.

⁸⁷ Greer (n73) at 20.

⁸⁸ Sheelah Connolly, Gwyn Bevan and Nicholas Mays, “Funding and performance of healthcare systems in the four countries of the UK before and after devolution”, (2010) The Nuffield Trust for Research and Policy Studies in Health Services 8.

⁸⁹ *Ibid*, at 8, citing *Encyclopaedia Britannica* (1889).

⁹⁰ *Ibid*, at 8.

⁹¹ Jean McHale, Elizabeth M. Speakman, Tamara Hervey and Mark Flear, ‘Health law and policy, devolution and Brexit’, (2021) 9 Regional Studies 1565.

⁹² *Ibid*, at 1565.

⁹³ Harrington (n86), at 65.

⁹⁴ Scott Greer, ‘Devolution and divergence in UK health policies’, (2009) 338 British Medical Journal 80.

ministers to make regulations about the circumstances and ways in which they carry them out”.⁹⁵

As Harrington *et al.* state, while legislation is important in health law, case law remains central to the law of negligence and consent.⁹⁶ Consistent reference is made to *Montgomery* and *Mordel*, for example, in UK-wide and national clinical guidelines on consent (more generally), and trisomy screening. Harrington *et al.* also note that broader regulation standards of HCP are nestled in UK-wide professional bodies, such as the General Medical Council (GMC) and the Nursing and Midwifery Council (NMC). Indeed, these bodies provide training and recommendations to practice to all registered nurses, midwives and ultrasonographers, across the UK.⁹⁷

The regulatory and quality organisations of the UK have complex interactions and relationships with devolution.⁹⁸ This complexity extends from interplay between professional regulation and health service policy; the former being a reserved power, and the latter being largely devolved.⁹⁹ Greer and Trench purport that organisations, such as the National Institute for Health and Clinical Excellence (NICE) and the Healthcare Commission, have “ragged edges”.¹⁰⁰

Indeed, in 2003, Jervis and Plowden stated that the 1998 ‘Devolution and Health’ report revealed that the Royal Colleges, and other professional organisations, valued their unified all-UK networks, and conveyed concern toward a possible fragmentation under devolution.¹⁰¹ Jervis and Plowden also drew attention to the findings of the report, which suggested that professional bodies sought conformity, in the flow of information and ideas, standards of clinical practice, training and education, and conditions of service.¹⁰² Thus, these bodies were described as being a force for policy stability and commonality, constraining divergence between the four UK countries in this regard.¹⁰³ As Harrington *et al.* conclude, “... all four

⁹⁵ Harrington (n86), 70.

⁹⁶ *Ibid*, 75.

⁹⁷ *Ibid*.

⁹⁸ Greer and Trench (n74), 513

⁹⁹ *Ibid*, at 513

¹⁰⁰ *Ibid*.

¹⁰¹ Paul Jervis and William Plowden, ‘The impact of political devolution on the UK’s health services’, (2003) The Nuffield Trust Report of Project to Monitor Impact of Devolution on the UK’s Health Services, 1-91, at 59.

¹⁰² *Ibid*, 59-60.

¹⁰³ *Ibid*, 60.

retain a considerable family resemblance, due not least to the continuing UK-wide application of key statutes, but also to the shared past of a common NHS and even longer-standing public health practices”.¹⁰⁴

Greer and Trench dissect the significance of divergency, between England and Wales, on professional training and regulation.¹⁰⁵ They note that professional education and health policy is devolved; however, professional regulation is a reserved matter, with the legal framework of professionals, and the regulatory councils like the GMC, NMC, NICE for example, are reserved powers of Westminster.

While professional regulation remains a UK function, the implementation and execution of education and training, including continuing professional development, is devolved.¹⁰⁶ Professional bodies are required to ensure that “any changed arrangements are acceptable in the devolved administrations”. Jervis and Plowden conclude that, “there is no doubt that devolution has brought challenges for the UK's professional bodies ... the over-arching challenge to professional bodies in the UK is to find appropriate ways of addressing both regulatory functions and professional interests and issues”.¹⁰⁷

Greer and Rowland suggested that health policymakers failed to grasp that “devolution is about divergence in ends as well as means”.¹⁰⁸ They explained that this was a threat to the shared values that underpin British identity, and to the autonomy of the devolved governments, believing that this “may be storing up future problems for both health and devolution”.¹⁰⁹

An empirical exploration into the significance of devolution of health law-making powers, specifically the divergences as between England and Wales, may provide a novel cultural and contextual insight into the issues surrounding the implementation and execution of national and local NHS policy, such as antenatal screening pathways. As Katikireddi *et al.* emphasise, “If policy diverges ... there will be opportunities for researchers to contrast the ... differing

¹⁰⁴ Harrington (n86), 89.

¹⁰⁵ Greer and Trench (n74), 515.

¹⁰⁶ Jervis and Plowden (n101), 60.

¹⁰⁷ *Ibid*, at 63.

¹⁰⁸ Scott L. Greer and David Rowland, ‘Devolving Policy, Diverging Values? The Values of the United Kingdom’s National Health Services’, (2007) The Nuffield Trust 1-104, at 94.

¹⁰⁹ *Ibid*, 94-95.

approaches ... while these 'natural experiments' could be invaluable in research terms, policy variations could exacerbate geographic inequalities across the UK, with implications for front line health staff".¹¹⁰

2.5 Influence of Clinical Guidelines

While clinical guidelines, in isolation, are unlikely to be determinative of the standard of care, based on previous case law¹¹¹, an empirical study by Samanta *et al.* indicated that guidelines "are particularly influential on judicial decision-making in areas such as obstetrics and cancer referral".¹¹² The reasons given for this conclusion included: the provenance; the content of the guidance, in these areas, were perceived as either 'black or white'; and the presentation of clinical guidelines was in an easily understandable format.¹¹³ The study underlined that clinical guidelines, in these areas, were often seen by the judiciary "as a benchmark for reasonable clinical care", and a presumption exists that, to deviate from these evidence-based guidelines, would be considered substandard or negligent.¹¹⁴

This is consistent with Lord Brailsford's judgment in *KR v Lanarkshire*, underlining the significance and status of NICE and RCOG guidelines, to inform standards of clinical practice.¹¹⁵ This is also consistent with Jay J's judgment in *Mordel*, relying heavily upon clinical guidelines (namely NICE and National Standards), to inform his assessment of reasonable and responsible practice.¹¹⁶

However, as Samanta *et al.* observe, guidelines are constructed for the purpose of assisting HCPs in practice, as opposed to providing an inventory for judges to decide on appropriate management of care.¹¹⁷ Indeed, it has been highlighted that judicial involvement, in matters

¹¹⁰ Srinivasa Vittal Katikireddi, Katherine E. Smith, David Stuckler and Martin McKee, 'Devolution of power, revolution in public health?', (2016) 39 Journal of Public Health 243.

¹¹¹ *C v North Cumbria* (2014) EWHC 61 (QB), 84. "In conclusion my view is that *prima facie* a midwife who acts in accordance with the guidelines should be safe from a charge of negligence. However, in the present case since it is common ground that in some regards the guidelines are not satisfactory I do not decide this case upon the basis that adhering to guidelines is sufficient. I consider that the fact that Midwife Bragg acted in accordance with the guidelines is a factor militating against negligence but I also assess Midwife Bragg's conduct against the benchmark of the other surrounding facts and circumstances".

¹¹² Ash Samanta, Jo Samanta and Joanne Beswick, 'Responsible practice or restricted practice? An empirical study of the use of clinical guidelines in medical negligence litigation', (2021) 00 Medical Law Review 1, 18.

¹¹³ *Ibid.*

¹¹⁴ *Ibid.*

¹¹⁵ *KR v Lanarkshire Health Board* (2016) CSOH 133.

¹¹⁶ *Mordel v Royal Berkshire NHS Foundation Trust* (2019) EWHC 2591 (QB), 134 and 140.

¹¹⁷ Ash Samanta, Jo Samanta and Joanne Beswick, (n112), 18.

of clinical judgment, raises its own concerns, and opportunities for judicial intervention are confined to instances where clinical opinion is “evidentially illogical”, despite the ruling in *Bolitho*.¹¹⁸

The lack of a standardised implementation of guidelines may explain the court’s conservative approach to assessing clinical practice. Indeed, discrepancies are bound to occur between Trusts and national programmes, particularly in areas such as antenatal screening. While this is true of many other devolved areas and responsibilities allocated to local authorities, it signifies the importance of expression in national policy, and the role of policy-makers in meeting a test of efficiency in equivalent results for parents in this regard.¹¹⁹

2.6 Clinical Guidelines on Trisomy Screening in England and Wales

The researcher engaged into a comprehensive review of existing clinical guidelines on trisomy screening for NHS England and Wales, and research literature pertaining to the practice of trisomy screening. Using an inductive narrative synthesis technique – assisted by Nvivo12 software – broad themes emerged from the clinical guidelines and literature, highlighting key areas of interest. As will be discussed in more depth in chapter 3, Thematic Analysis techniques were used to provide a framework for the broader themes identified from the literature. While many clinical guidelines were reviewed (see Appendix 3), the researcher understood that NICE, FASP and ASW guidelines were directly applicable in this context.

2.6.1 HCPs’ Roles for Securing Consent for Trisomy Screening

While the importance of decision in *Montgomery* for effectively securing consent is pervasive throughout clinical guidelines in this area, ASW¹²⁰ and FASP¹²¹ guidelines only briefly outline HCP roles for securing consent for trisomy screening and testing. Indeed, the judgment in *Mordel* alluded to the current ambiguity surrounding the HCPs role, particularly sonographers and midwives, for securing consent under established systems.

¹¹⁸ Ibid.

¹¹⁹ T. T. Arvind and Aisling M. McMahon, ‘Responsiveness and the Role of Rights in Medical Law: Lessons from *Montgomery*’, (2020) 28 Medical Law Review 445, 467.

¹²⁰ Public Health Wales. Antenatal Screening Wales Policy, Standards and Protocols (2019) – 7.0 and 8.0 Antenatal Screening for DS, ES and PS, standard 7.3, standard 5.

¹²¹ Public Health England. NHS Public Health Functions Agreement (2018). Service Specification no.16. NHS Fetal Anomaly Screening Programme – Screening for DS, ES and PS (Trisomy 21, 18 & 13).

Under current trisomy screening pathways, both ASW and FASP mandate that the system operates on the basis of receiving parents' verbal consent, and the decisions to accept or refuse screening is be documented and recorded 'appropriately', at the booking appointment; this is either done in the parent's handheld or computerised records. The NMC and NICE highlight that clear and accurate records of practice must be kept, noting evidence of any identified risks or problems that have arisen.¹²² However, this is the extent of the guidelines, pertaining to the expected system for securing consent. Inevitably, there will be differences across Hospital Trusts as to how consent is obtained and documented, possibly explaining clinical guidelines' vagueness in this regard (see Appendix 2 for diagram on current system for securing consent).

Midwives

Clinical guidelines state that the midwife plays the leading role in securing consent for trisomy screening. It is common practice that once a woman discovers that they are pregnant, an appointment is arranged to see a midwife (either community or clinic). Fundamentally, midwives are supposed to counsel parents for all screening testing in pregnancy, including trisomy screening and testing. At first contact, or during the initial booking appointment, the parents will be provided with a comprehensive care package, where various information materials are delivered informing them of all aspects of their pregnancy journey. At this appointment, discussion will be had on an array of topics, including antenatal screening and testing. More specifically, parents will be informed of the trisomy screening and testing pathway. Guidelines indicated that it is the role of the *midwife* to obtain consent, from parents, before they decide to embark on the trisomy screening and testing pathway.

Sonographers

Sonographers are technicians, who are qualified to conduct ultrasound scans. Clinical guidelines state that their duty is to identify and report any anomalies on the scan which may require further investigation. Guidelines do not state that sonographers are duty-bound to explore parent consent, nor to counsel parents in this regard; however, sonographers will validate whether the parent's decision, from the booking appointment, still stands before

¹²² Nursing & Midwifery Council (2018) The Code: Professional Standards of Practice and Behaviour for Nurses, Midwives, and Nursing Associates, standard 10.

conducting the scan. Their duty is to record and report any atypical findings, that they see on the scan, to the obstetrician or fetal medicine consultant (in the fetal medicine unit). More specifically, sonographers usually conduct the NT measurement scan which forms one part of the combined screening. Consent to screening should have been obtained prior to the scan by the midwife, and therefore the sonographers proceed on the basis of the initial decision.¹²³

The duty of the sonographer is an overarching professional duty, not confined solely to screening. It is not up to the sonographer to overlook findings, which could be construed as abnormal; an appropriate referral to a fetal medicine consultant is required. However, in the context of trisomy screening, the decision to decline screening is respected, with shared decision-making values providing the foundation to the conversation.¹²⁴ Therefore, a parent, who has declined combined screening, will not have their risk-score calculated, as structural anomalies may arise from multiple underlying conditions, and it is not possible to reliably identify DS, ES and PS using ultrasound alone. It is important to note, however, that the desire of the parent to not be told about the chance of their baby having an anomaly, does not override the professional duty of the sonographer to report atypical findings on the scan.¹²⁵

Obstetricians and Fetal Medicine Consultants

An obstetrician is a qualified doctor, who has a specific knowledge in providing general antenatal and postnatal care. Their involvement in supporting parent decision-making is often limited to high-risk and/or complicated pregnancies. For example, they are consulted if the mother has any additional needs during the period of antenatal care or birth following screening and testing results. Obstetricians can perform a role in the delivery of the baby and can also perform surgery if required. While they work closely alongside midwives throughout antenatal care (including screening and testing) and post-delivery, their involvement in the consent process is often engaged following a higher-risk result and/or decisions surrounding continuing or ending pregnancies.

¹²³ NICE. Society of Radiographers (2018) – Obtaining Consent: A Clinical Guideline for the Diagnostic Imaging and Radiotherapy Workforce, point 3.

¹²⁴ The Society & College of Radiographers. BMUS: Guidelines for Professional Ultrasound Practice (2019), point 4.

¹²⁵ Ibid.

A fetal medicine consultant can also work closely alongside midwives, in the event that an observation needs to be conducted on parent who have more complex or atypical needs in pregnancy. They could also be described as specialist obstetricians. They are consulted to provide counselling and support to those parents who have a high-risk pregnancies, or if an anomaly has been identified during screening or testing.

2.6.2 Information on Trisomy Screening

Once a woman discovers they are pregnant, it is commonplace to book an appointment with a midwife. Antenatal Screening Wales (ASW) and the Fetal Anomaly Screening Programme (FASP) state that at first contact with the midwife, parents should be presented with information on antenatal screening for fetal anomaly (amongst other pregnancy information).¹²⁶ Midwives are required to convey the importance that parents have received and read the relevant information materials on trisomy screening and testing, before consenting to accept or refuse the offer.¹²⁷ According to NICE¹²⁸, ASW and FASP, HCPs must explain, at this stage, what trisomy screening and testing offers to parents, along with its purpose.¹²⁹

Jay J, in *Mordel*, drew upon the requirement for provision of information, in *Montgomery*, that parents should be provided with 'sufficient information' at first contact with the HCP before consenting to undertake screening and/or testing. *Mordel* also placed significance on the system of providing information, dividing this duty between the midwife and sonographer across the appointments to effectively support decision-making and consent. *Mordel* underlined that sonographers are required to check if the information had been received by the parents, from the midwife, before engaging with screening.

Research has recently conveyed concern towards the volume of information parents receive at the beginning of the trisomy screening and testing pathway, exacerbating the risk of "information overload".¹³⁰ Heuvel *et al.* reinforced the importance to not overwhelm parents

¹²⁶ ASW (n120), standard 7.1 (1), FASP (n121), point 5.

¹²⁷ *Ibid*, standard 7.2, *ibid*, point 5.2.

¹²⁸ The National Institute for Health and Care Excellence (2008 updated 2019) Antenatal Care for Uncomplicated Pregnancies. Clinical Guideline (CG62), point 1.1.1.8.

¹²⁹ ASW (n120), standard 7.2, FASP (n121), point 5.2.

¹³⁰ Mollie A. Minear and others, 'Noninvasive Prenatal Genetic Testing: Current and Emerging Ethical, Legal, and Social Issues', (2015) 16 *Annu. Rev. Genomics Hum. Genet* 369, 98.

with information, as they are still coming to terms with the pregnancy, and many parents are new to the screening experience.¹³¹ However, Polansky asserts the importance of acknowledging the increasing time restrictions midwives experience during the process of disclosing information at clinical appointments.¹³² Indeed, research has revealed that midwives typically spend thirty-seconds to two-minutes discussing trisomy screening at the booking appointments, due to the other informational demands.¹³³ Polansky emphasises that clinical guidelines are unsympathetic to the time-pressured and busy confines of the clinical setting.¹³⁴

The information that parents first receive is understandably generic, and is designed to cater to the entire population. However, guidelines mandate that midwives should seek to tailor and individualise the information, subject to the particular parent.¹³⁵ Indeed, midwives are required to engage into a continued dialogue with parents to create an individualised and flexible care plan.¹³⁶ However, academics question how midwives are expected to tailor the information within the time-limited confines of clinical appointments; clinical guidelines are not clear on this duty.

Trisomies

NICE, ASW and FASP guidelines state that parents should receive accurate and balanced information on the conditions being screened for; that is, DS, ES and PS.¹³⁷ Each tested condition has very distinct physical and developmental characteristics, and the counselling stage of trisomy screening should reflect this.¹³⁸ An accurate depiction of the aetiology and prognosis of a baby with DS, ES and PS should also be presented.¹³⁹

Midwives, at this stage, should also be aware that the phrase “trisomy screening” is reflective of a genetic model, and does not, in itself, distinguish between the individual characteristics

¹³¹ van den Heuvel A and others, ‘Will the introduction of noninvasive prenatal diagnostic testing erode informed choices? An experimental study of health care professionals’, (2009) 78 Patient Educ Couns 24, 28.

¹³² Samara Polansky, ‘Overcoming the Obstacles: A Collaborative Approach to Informed Consent in Prenatal Genetic Screening’, (2006) 14 Health Law Journal, 21, 31.

¹³³ Sophie John *et al.* (n15), 6.

¹³⁴ Samara Polansky (n132), 31.

¹³⁵ ASW (n120), standard 7.2, FASP (n121), point 5.3.

¹³⁶ NMC (n122), point 2.4.

¹³⁷ NICE (n123) point 1.7.2.5, ASW (n120), standard 7.2, FASP (n121), point 5.3.

¹³⁸ Houlihan, O.A, and O’Donoghue, K, “The natural history of pregnancies with a diagnosis of trisomy 18 or 13; a retrospective case series”, (2013), 13 BMC Preg Childbirth, 209.

¹³⁹ NICE (n123) point 1.7.2.4.

of each trisomy.¹⁴⁰ While the presentation of ‘trisomy’ screening as a genetic model is ultimately unavoidable following its construction, there must be reference to the distinct phenotype. Failing to do so could mislead women into thinking that all the trisomies are the same, in terms of aetiology and prognosis, which is not so. Indeed, upon construction of the ‘trisomy’ pathway, the British Maternal and Fetal Medicine Society executive committee (BMFMS) warned HCPs that PS and ES must not be presented as “just more severe cases of Down Syndrome” before its implementation.¹⁴¹

Guidelines outline that midwives are required to deliver clear, accurate and balanced information on the trisomies, at first contact.¹⁴² However, research is consistently outlining the concern parents have towards the information disclosed on the trisomies. Studies reveal that HCPs focus unduly on the negative implications of the trisomies, as opposed to delivering a balanced and accurate depiction of the conditions.¹⁴³

Indeed, Skotko *et al.* found that HCPs disclosed information on the negative implications of DS, before providing limited, if any, information on the condition’s positives.¹⁴⁴ Research also suggests that ES and PS are subject to similar treatment during pre-screening counselling, with many parents stating that they were “unhappy with how information on diagnosis and prognosis were communicated”.¹⁴⁵

Qualitative studies reveal that information on terminations often follows counselling on ES or PS, commonly labelled as ‘lethal’ conditions.¹⁴⁶ However, many cases of persons with ES and PS living into adulthood, suggesting an imbalance of information in this regard.¹⁴⁷ Scholars stress that up to date and accurate information should be delivered on the prognosis of each condition with developments in technology and treatment continuously improving life-

¹⁴⁰ Fenton L.J, “*Trisomy 13 and 18 and quality of life: treading ‘softly’*”, (2011) *Am J Med Genet*, 1527, 1528.

¹⁴¹ UK NSC First Trimester Combined Screening for Trisomy 18 and Trisomy 13. Consultation Comments. June 2014.

¹⁴² ASW (n120), standard 7.2, FASP (n121), point 5.6, NICE (n123), point 1.7.2.5.

¹⁴³ Sophie John and others, (n15), 3.

¹⁴⁴ Brian G Skotko, ‘*Postnatal diagnosis of Down syndrome: synthesis of the evidence on how best to deliver the news*’ (2009) *124 Paediatrics* 751, 758.

¹⁴⁵ *Ibid.*

¹⁴⁶ Dominic Wilkinson, Lachlan de Crespigny and Vicki Xafis, ‘*Ethical Language and Decision-Making for Prenatally Diagnosed Lethal Malformations*’, (2014), *19 Semin Fetal Neonatal Med*, 306, 311.

¹⁴⁷ Bhanumathi, B, Goyel N.A, Mishra, Z.A, ‘*Trisomy 18 in a 50-year old female*’, (2006) *12 Ind J Hum Genet*, 146, 147.

expectancy and care management.¹⁴⁸ ASW and FASP direct parents charities and organisations such as ARC¹⁴⁹, SOFT¹⁵⁰ and the DSA¹⁵¹ for balanced and accurate information on the diagnosis and prognosis of the trisomies.

Methods of Screening and Testing

Clinical guidelines state that, at first contact with the midwife, information must be delivered on the different methods of screening and testing.¹⁵² This requires the midwife to disclose information on combined screening, quadruple screening, and the methods of invasive testing: amniocentesis and CVS.¹⁵³

NICE, FASP and ASW stipulate that midwives should provide a clear and accurate description of what the combined test entails. In the first trimester of the woman's pregnancy, the combined test provides the *first* opportunity to detect chromosomal differences in the baby.¹⁵⁴ Between 11+2 weeks and 14+1 weeks of pregnancy, a sample of the mother's blood is taken to analyse biochemical markers, and an ultrasound scan is performed.¹⁵⁵

Midwives are required to explain that the 12-week (early pregnancy) ultrasound scan, sometimes called the "dating scan", will form part of the combined test for the purposes of trisomy screening. If the parent consents to have the combined test, a nuchal translucency (NT) measurement is taken from the back of the baby's neck (nuchal fold) during this ultrasound scan.¹⁵⁶ This scan detects any indicators which may need further investigation.¹⁵⁷

¹⁴⁸Irving C and others, 'Changes in fetal prevalence and outcome for trisomies 13 and 18: a population-based study over 23 years' (2011) 23 J Matern Fetal Neonatal Med 137, 141.

¹⁴⁹ Antenatal Results and Choices, available at: <https://www.arc-uk.org/> (accessed 26/05/2020).

¹⁵⁰ Support Organisation for Trisomy 13 and 18, available at: <https://www.soft.org.uk/> (accessed 26/05/2020).

¹⁵¹ Down's Syndrome Association, available at: https://www.downs-syndrome.org.uk/?gclid=EAlaIQobChMI0emyvuDR6QIV2u3tCh2JZgBTEAYASAAEgIEQPD_BwE (accessed 26/05/2020).

¹⁵² ASW (n120) standard 7.2, FASP (n121) point 5.3.

¹⁵³ Ibid.

¹⁵⁴ NICE (n123), standard 1.7.2.1, ASW (n120) standard 7.2, FASP (n121) point 5.2.

¹⁵⁵ A combination of the nuchal translucency (NT) scan measurement, conducted by a sonographer around 11+4 and 13+6 weeks gestation, with a blood sample from the mother which measures the concentration of pregnancy associated plasma protein-A (PAPP-A) and free beta human chorionic gonadotrophin (free beta hCG), is examined together with the mother's age, the gestation of the pregnancy, and the crown rump length (CRL) measurement (between 45.0mm and 84.0mm), to estimate the chances that the baby has DS, ES or PS. The chance-score is produced using the Fetal Medicine Foundation algorithm.

¹⁵⁶ A pocket of fluid is visible on the back of the baby's neck in the first trimester. Using an ultrasound scan, this pocket of fluid is measured to assess, in combination with other biological factors, the likelihood of the baby having a tested condition.

¹⁵⁷ Soft markers, or indicators, such as the size of the baby's nasal bone is also measured.

This scan usually takes between 10-15 minutes to complete.¹⁵⁸ Parents are still able to have the early pregnancy 12-week “dating” ultrasound scan, even if they *decline* the combined test for the purposes of trisomy screening. In some NHS maternity units, if the parents are unsure of whether they would like the 12-week dating scan to form part of the combined test, it is common practice to book the parents in to have the combined test (to take an NT measurement), giving them the option to withdraw from having the combined test on the day of the 12-week dating scan. If parents decide to withdraw, only the 12-week dating scan will be performed, and an NT measurement will not be taken.¹⁵⁹

Both FASP and ASW state that, if parents have not consented to have the combined test for the purposes of trisomy screening, an NT measurement will *not* be taken during the early pregnancy 12-week “dating” scan.¹⁶⁰ However, guidelines state that it should be clear that during the scan, if the nuchal fold (NT measurement) is *incidentally* identified as being enlarged ($\geq 3.5\text{mm}$), sonographers are under a *duty* to report the enlarged measurement to fetal medicine.¹⁶¹ An increased NT measurement may correlate to an increased chance of other differences in the baby.¹⁶² It should also be explained that a possible issue could arise in circumstances where the sonographer is unable to obtain an NT measurement, which occurs in 3% of all women; this may be due to an increased body mass index or the position of the baby in utero.¹⁶³

A clear and accurate description of what a quadruple test entails should be provided.¹⁶⁴ According to clinical guidelines, if the parent presents after 14-weeks’ gestation, and misses the opportunity to have the combined screening test, they should be offered ‘quadruple’ screening (often referred to as ‘the 16-week scan’, ‘serum screening’ or the ‘traditional blood test’). Furthermore, NICE guidelines state that, if it is not possible to obtain an NT measurement in the process of the combined test, possibly due to fetal position or an increased body mass index, parents should be offered the quadruple test as an alternative

¹⁵⁸ Discussion with practising sonographer.

¹⁵⁹ This is the course of practice at Liverpool Women’s maternity unit.

¹⁶⁰ ASW (n120), standard 8.2.3 (4), FASP (n121) point 5.5

¹⁶¹ Ibid.

¹⁶² Pilnick, A.M., Fraser, D.M. & James, D.K, ‘Presenting and discussing nuchal translucency screening for fetal abnormality in the UK’, (2004) 20 Midwifery 82-93.

¹⁶³ Ibid.

¹⁶⁴ ASW (n120) standard 8.2.3 (5), FASP (n121) point 5.4

between 14+2 and 20+0 weeks.¹⁶⁵ However, the optimum time for conducting this test is 16-weeks' gestation.¹⁶⁶ This method of testing has a reported detection rate of 70-75%, with a false positive rate of 3-4%.¹⁶⁷ Midwives should be clear that this screening test is only used to produce a risk-score for DS; however, the quadruple test is not as effective as the combined screening test at detecting DS. Quadruple screening is unable to produce a risk-score for ES or PS. Despite this, the quad test is the recommended screening strategy in the second trimester.

NICE, FASP and ASW guidelines also state that, at first contact with parents, midwives should provide an accurate description of the different methods of invasive diagnostic testing that can be performed, if a higher-risk result is returned following screening; that is, amniocentesis and chorionic villus sampling.¹⁶⁸ FASP and ASW also state that parents must have sufficient time to feel comfortable (around 24 hours) to decide whether or not to undertake invasive testing.¹⁶⁹

Amniocentesis is an invasive procedure undertaken from 15 completed weeks (15+0) and onwards to obtain a sample of amniotic fluid (liquor) surrounding the fetus. Using an aseptic technique whilst under continuous ultrasound guidance, a sterile thin needle is passed through the mother's abdomen, uterus and amniotic sac. A sample of amniotic fluid is aspirated with a syringe and sent for analysis to test for a range of chromosomal and inherited disorders. Up to 1:100 women who undertake amniocentesis will miscarry.

Chorionic villus sampling (CVS) is an abdominal or cervical procedure performed under continuous ultrasound guidance after 10 completed weeks in pregnancy to obtain a sample of placental tissue for chromosomal or genetic analysis (between 10-13 weeks gestation). The range of chromosomal and genetic conditions that can be detected is similar to those for

¹⁶⁵ NICE (n123), point 1.7.2.4.

¹⁶⁶ The test is most effective at this stage of gestation in terms of detection rate. This second trimester test measures the levels of four biochemical markers in the blood (serum); that is, the AFP, uE3, free beta hCG (or total hCG), and inhibin-A. Together with the maternal age, a risk-score is calculated. If the baby has DS, the AFP and uE3 levels are typically low and the inhibin levels are raised.

¹⁶⁷ Kerry Oxenford and others, *'Development and evaluation of training resources to prepare health professionals for counselling pregnant women about non-invasive prenatal testing for Down syndrome: a mixed methods study.'* (2017) 17 BMC Pregnancy and Childbirth, 132

¹⁶⁸ NICE (n123), point 1.7.2.5, ASW (n120) standard 7.4, FASP (n121), point 5.7.

¹⁶⁹ ASW (n120), standard 7.7.8 (5), FASP (n121), point 5.7.

amniocentesis. Up to 1-2 out of every 100 women who undertake CVS will miscarry.¹⁷⁰ Among the general obstetric population, 100 women will undergo invasive testing to discover 10 true positives.¹⁷¹

2.6.3 'Understanding' and Trisomy Screening

Once the information materials have been delivered and read, guidelines state that the midwife should confirm that parents understand the information pertaining to the methods of screening and testing, and the conditions being screened for, before recording consent.¹⁷² If not, guidelines state that an opportunity must be provided to ask questions; answers to these questions must be clear and accurate.¹⁷³

Clinical guidelines – namely the GMC and the Nursing and Midwifery Council (NMC) – refer to *Montgomery* in the context of ensuring that HCP(s) support patient understanding of the proposed treatment before providing informed consent.¹⁷⁴ *Mordel* also placed a duty upon the midwife and sonographer to take reasonable steps to explore parent understanding before securing consent for screening. Sonographers were assigned a duty by Jay J to check whether the parent has understood the information provided by the midwife at first contact. *Mordel* reminded HCPs that parent consent succeeds brief questioning, by the sonographer, to ascertain whether the parent has understood the essential elements of screening and/or testing.

The NMC, FASP and ASW also highlight that the midwife must 'check' parent understanding, to minimise the risk of misunderstanding and misinterpretation.¹⁷⁵ However, there is currently a lack of clear guidance for midwives as to how this objective can be effectively achieved. *Favre et al.* emphasised that inappropriate probing into the understanding of

¹⁷⁰ Ranjit Akolekar and others, 'Prediction of Miscarriage and Stillbirth at 11-13 Weeks and the Contribution of Chorionic Villus Sampling', (2011) 31 *Prenat Diagn*, 38, 45.

¹⁷¹ *Ibid.*

¹⁷² ASW (n120), standard 7.4, FASP (n121), point 5.6.

¹⁷³ *Ibid.*, standard 7.4.1, *Ibid.*, point 5.7.

¹⁷⁴ NMC (n123), point 9.

¹⁷⁵ NMC (n123), point 9, ASW (n120), standard 7.5, FASP (n121), point 5.7.

parents risks the “non-respect of ethical principles” during the screening process and could undermine reproductive autonomy.¹⁷⁶

Guidelines state that minimising the use of medical jargon and technical terms is imperative for supporting parent understanding.¹⁷⁷ Research has demonstrated that use of jargon is unavoidable when discussing information on trisomy screening and testing; however, a simple and accurate explanation of these terms should follow.¹⁷⁸ Favre *et al.* emphasise that overloading parents with medical or technical terminology is detrimental to understanding the information, and could subsequently impact on their ability to deliver truly informed consent.¹⁷⁹

ASW and FASP outline that midwives should support parent understanding on the potential consequences of having screening and testing, and the requisite steps under the care pathway following a screening or testing result.¹⁸⁰ NICE guidelines state that parents must understand the decisions that need to be made at each point along the pathway, and their consequences.¹⁸¹ For most parents, they will not receive an unexpected result following screening and testing and will receive standard antenatal care; however, this should not cloud the experience of the minority of parents, who could receive an unexpected result. Lewis *et al.* questioned whether one could ever be prepared for the consequences of a positive diagnosis, following the findings of their study of parents in this population.¹⁸²

Trisomies

Guidelines mandate that midwives support parents’ understanding of the *nature* of the tested conditions; this includes supporting understanding of the aetiology and pathogenesis of DS, ES and PS, in addition to the variability of each condition.¹⁸³ Midwives should draw attention

¹⁷⁶ Favre, R. and others, ‘*Is the non-respect of ethical principles by health professionals during first-trimester sonographic Down syndrome screening damaging to patient autonomy?*’, (2009) 34 *Ultrasound in Obstetrics & Gynecology*, 25-32.

¹⁷⁷ ASW (n120), standard 7.2, FASP (n121), point 5.3.

¹⁷⁸ *Ibid.*

¹⁷⁹ Favre R and others (n176), 26.

¹⁸⁰ ASW (n120), standard 7.1, FASP (n121), point 5.2.

¹⁸¹ NICE (n123), point 1.7.2.5.

¹⁸² C. Lewis *et al.* (n5), 232.

¹⁸³ ASW (n120), standard 7.3, FASP (n121), point 5.5. Full trisomy 21, 18 and 13 (meiotic nondisjunction), mosaic, translocation and partial are all biological variants of the tested conditions. Full trisomy 21, 18 and 13 (meiotic nondisjunction) is the most common form of DS, ES and PS. Where only some of the cells have an additional copy of either chromosome 21, 18 or 13 (half the cells are normal, and half are abnormal), this is

to the biological spectrum associated with DS, ES and PS, ranging from those babies who could have life-limiting differences, to those who only have moderate or mild differences for a balanced understanding.

Research suggests that HCPs insufficiently support parent understanding of the trisomies, resulting to an “imbalanced and biased” depiction of the conditions.¹⁸⁴ Heyman *et al.* suggests that the “positive aspects” of the trisomies – such as achievable life goals, advancements in health technology to extend prognosis, and an *accurate* prognosis of a baby with the tested conditions – are commonly omitted from pre-screening counselling.¹⁸⁵ This concern is said to be particularly pertinent to ES and PS, with researchers concluding that both ES and PS are still commonly described as being “incompatible with life”.¹⁸⁶

Qualitative studies with parents, who had babies with ES or PS, revealed that HCPs found it difficult to overlook the adverse statistics around the prognosis of these conditions.¹⁸⁷ Many parents felt angered, and often confused at the use of the term “incompatible with life”, as online searches for ES and PS generated images of older children with these conditions, conflicting with the HCP’s presentation of the condition.¹⁸⁸ The trust of parents towards the HCPs would often diminish as a result.¹⁸⁹ A study conducted by Guon *et al.* suggested that HCPs treat a baby with ES and PS as a diagnosis, rather than a baby, creating a significant amount of distress and anger among parents.¹⁹⁰ However, these academics maintain that there are several existing cases of adults living with either ES or PS, who are currently living happy, semi-independent and fulfilling lives, emphasising the value of obtaining experiential knowledge and lived experience to inform the provision of support along the screening pathway.¹⁹¹

referred to as mosaic DS, ES or PS which can sometimes be problematic in the context of detecting the abnormality in antenatal screening. Translocation DS, ES or PS is where the additional copy of chromosome 21, 18 or 13 attaches itself to another chromosome with partial DS, ES or PS occurring from the presence of extra genetic material originating from only a part of the chromosome.

¹⁸⁴ Heyman, B and others, ‘*On being at higher risk: A qualitative study of prenatal screening for chromosomal anomalies*’, (2006), 26 *Soc Sci Med* 2360, 2372.

¹⁸⁵ *Ibid.*, 2363.

¹⁸⁶ Dominic Wilkinson, Lachlan de Crespigny and Vicki Xafis, (n146), 306.

¹⁸⁷ *Ibid.*

¹⁸⁸ *Ibid.*

¹⁸⁹ *Ibid.*

¹⁹⁰ Jennifer Guon and others, ‘*Our children are not a diagnosis: The experience of parents who continue their pregnancy after a prenatal diagnosis of trisomy 13 or 18*’, (2014) 164 *Am J Genet* 308, 318.

¹⁹¹ *Ibid.*

Guidelines require midwives to support parents' understanding of the *causes* of the different tested conditions; that is, it is a chromosomal condition, it is seldom hereditary, and is associated with an increase in maternal age. A study by Heyman *et al.* found that parents believed their lifestyle choices and actions increased the likelihood of their baby having a trisomy; this is simply not true.¹⁹² According to ASW and FASP, counselling should also highlight that there is no "cure" for these conditions, however provision is in place to manage and care for babies with a trisomy.¹⁹³

ASW and FASP state that midwives should check that parents are aware that trusted charities, organisations and support groups are available to them if they desire; verified websites on trisomy screening and testing include ARC (Antenatal Results and Choices), SOFT (Support for Trisomy 13 and 18) and the DSA (Down's Syndrome Association).¹⁹⁴

Methods of Screening and Testing

Clinical guidelines state that midwives must support parent understanding of the purpose of screening and testing for trisomy, including their advantages, disadvantages and alternatives.¹⁹⁵ This is explained to be an important component to the discussion when balancing the parents' expectations of screening and testing with the reality.¹⁹⁶ A number of indicators has been highlighted by research as to why parents consider trisomy screening and testing as an option; these reasons include providing reassurance that the baby does not have a tested condition, providing sufficient time to prepare for a baby with a tested condition and consider any options which may follow, perceiving the offer of screening and testing as logical or sensible which stems from a trust towards the HCP, pressure from socio-cultural expectation as being a "responsible" parent, and the negative perception of the tested conditions.¹⁹⁷

With any method of screening – combined or quadruple screening for the purpose of detecting trisomy – the benefits are being able to identify a difference early on in the pregnancy. This will allow HCPs to prepare for any interventions that may need to be

¹⁹² Heyman, B and others, (n184), 2372.

¹⁹³ ASW (n120), standard 7.2.3, FASP (n121), point 5.8.

¹⁹⁴ ASW (n120) standard 7.8.7 (5), FASP (n121), point 6.

¹⁹⁵ ASW (n120), standard 7.2.3, FASP (n121), point 5.8.

¹⁹⁶ *Ibid*, *Ibid*.

¹⁹⁷ Tom Shakespeare and others (n12) 9.

performed, or to closely monitor the pregnancy if a difference is identified early on. The disadvantages to any method of screening are the possibility of false-positives and negatives; that is, the accuracy of the screening result is not definitive. The accuracy of each method of screening varies, however combined screening is deemed to be more accurate than quadruple screening which is used to screen for DS only.

The advantages to any method of testing (amniocentesis or CVS) is that it will provide a definitive result if the expectant mother opts to have it. It can be performed early in pregnancy which allows parents to consider all the possible implications. It may also allow HCPs to prepare for an alternative birth plan. The disadvantages to the traditional methods of testing are that it carries a 1-2%¹⁹⁸ risk of procedural miscarriage, however figures on the true risk of procedural miscarriage conflict between medical studies.¹⁹⁹

Understandably, many parents suffer a significant amount of stress and anxiety when presented with the option of invasive diagnostic testing. A study conducted by Lewis *et al.* revealed that parent experienced a substantial amount of distress at the thought of a needle being inserted into their womb whilst pregnant, with one participant to the study commenting that anxiety was induced by “having this large needle stuck in your stomach ... or poking the baby in the eye”.²⁰⁰ ASW and FASP outline that the midwife should support parent understanding of the disadvantages of invasive methods of testing clearly and accurately, providing them with an opportunity to ask any questions or raise concerns they may have.

2.6.4 ‘Choice’ and Trisomy Screening

In light of the developments from *Montgomery* – routinely cited by clinical guidelines in the context of preserving ‘choice’ – ASW and FASP endorse that patients’ rights and respect for

¹⁹⁸ The Royal College of Obstetricians and Gynaecology quotes a risk of procedural miscarriage of 1% for amniocentesis, and 1-2% for CVS. The American College of Obstetricians and Gynaecology quotes a risk of procedural miscarriage of 0.3-0.5% for amniocentesis, and 0.3-0.5% for CVS. Therefore, the figure varies between 0.3-2% depending on national programmes.

¹⁹⁹ Wulff, C and others, ‘Risk of Fetal Loss Associated with Invasive Testing Following Combined First-Trimester Screening for Down Syndrome: A National Cohort of 147, 987 Singleton Pregnancies’ (2016) 47 *Ultrasound Obstet Gynecol* 38, 44. This Danish study was the first to study the risk of invasive testing on the baby. The widely referred to 1% risk of miscarriage from invasive testing originated from this study. This study, however, could not definitively draw the distinction between procedural and non-procedural (natural) miscarriages.

²⁰⁰ C. Lewis *et al.* (n5), 228.

autonomy form a fundamental component of the ‘correct’ model of care.²⁰¹ Indeed, the NMC list the ‘rights’ that any persons accessing NHS services should possess in these contexts, placing emphasis on empowering and protection human rights and reproductive autonomy.²⁰²

Mordel placed a novel duty on sonographers to explore parent choice in the context of trisomy screening. Jay J required the sonographer to undertake reasonable steps to conduct a ‘gentle exploration’ of the parent’s state of mind, ensuring that their choice is forthcoming on the day of the scan, and to effectively secure the parent’s consent in this regard.

All parents, across England and Wales, should be offered the opportunity to undertake trisomy screening.²⁰³ This offer is described as intending to increase reproductive autonomy and choice.²⁰⁴ However, NICE, FASP and ASW guidelines warn that the midwife should clearly disclose that screening and testing is merely *optional*, and not a mandatory component of the mother’s antenatal care.²⁰⁵ The guidelines also emphasise that screening should not be presented as part and parcel, or a routine aspect, of their antenatal care.²⁰⁶

ASW and FASP guidelines outline that, while ‘trisomy’ screening is the collective term used to screen for DS, ES and PS under the pathway, parents have the option to decide whether they want to screen for all conditions together, DS only or ES and PS only.²⁰⁷

Routinisation of Screening

Studies suggest that parents do not always appreciate that trisomy screening is their choice.²⁰⁸ In a study conducted by Lewis *et al.*, parents revealed that they believed trisomy screening to be “one of those tests you do”, emphasising that they “did not think it was a choice”.²⁰⁹ Silcock *et al.*, outlined that parents are more likely to make an informed decision

²⁰¹ ASW (n120), standard 7.2.6, FASP (n121), point 5.5.

²⁰² NMC (n122) standards 1.1, 1.2, 1.3, 1.4 and 1.5

²⁰³ ASW (n120), standard 6, FASP (n121), point 5.

²⁰⁴ García, E., Timmermans, D.R.M. & van Leeuwen, E, ‘*The impact of ethical beliefs on decisions about prenatal screening tests: Searching for justification*’, (2008) 66 *Social science & medicine* 753, 764.

²⁰⁵ NICE (n123), point 1.7.2.1, ASW (n120), standard 7.3, FASP (n121), point 5.1.

²⁰⁶ NICE (n123), point 1.7.2.1, ASW (n120) standard 7.2-7.3, FASP (n121), point 5.1.

²⁰⁷ ASW (n120), standard 7.0, FASP (n121), point 5.1.

²⁰⁸ Yanikkerem, E., Ay, S., Çiftçi, A., Y, Ustgorul, S. and Goker, A, ‘*A survey of the awareness, use and attitudes of women towards Down syndrome screening*’, (2012) 22 *Journal of Clinical Nursing*, 1748, 1758.

²⁰⁹ C. Lewis *et al.* (n5), 230.

when trisomy screening and testing is presented as optional, as opposed to routine.²¹⁰ Gottfredsdottir *et al.* identified that disclosure of the option to refuse screening, or “informed refusal”, is often omitted during booking appointments.²¹¹ However, Garcia *et al.* identified factors influencing ‘choice’. They found that parents are at risk of feeling compelled to uptake screening, regardless of whether it is presented as optional or routine, as it is being *offered* by the HCPs.²¹² Indeed, academics question whether parents succumb to social or institutional pressure to have trisomy screening.²¹³

Guidelines also indicated that midwives should clearly explain that parents are able to opt out from the trisomy screening pathway, or decline further investigative testing at any time.²¹⁴ Some parents may not feel the need to embark on antenatal screening and/or testing, as they have previously decided that termination of the pregnancy is not an option regardless of any differences with the baby.²¹⁵ Others may decide that they would like to be informed whether or not their baby has a condition to plan the next steps, or to end the pregnancy.²¹⁶

ASW and FASP underline that midwives must make patient’s aware that they are able to change their minds at any time, and their decision should be respected.²¹⁷ In clinical practice, change of mind is reported to be very common, emphasising the significance of maintaining an open dialogue with parents across the pathway to support their choice.²¹⁸

A study by Aune and Moller revealed a conflict with the concept of ‘choice’.²¹⁹ The parents, in this study, felt that having a choice was important, but they did not always want to make

²¹⁰ Caroline Silcock and others, ‘Will the Introduction of Non-invasive Prenatal Testing for Down’s Syndrome Undermine Informed Choice?’, (2015) 18 International Journal of Public Participation in Health Care and Health Policy 1658, 1660.

²¹¹ Helga Gottfredsdóttir, Kristin Björnsdóttir and Jane Sandall, ‘How do prospective parents who decline prenatal screening account for their decision? A qualitative study’, (2009) 69 Social science & medicine 274, 277.

²¹² García, E., Timmermans et al. (n204), 843.

²¹³ Ahmed, S., Bryant, L.D. & Cole, P. ‘Midwives’ perceptions of their role as facilitators of informed choice in antenatal screening’, (2013) 29 Midwifery, 745, 749.

²¹⁴ ASW (n120), standard 7.2.2, FASP (n121), point 5.2.

²¹⁵ Favre, R. and others, ‘Is the non-respect of ethical principles by health professionals during first-trimester sonographic Down syndrome screening damaging to patient autonomy?’, (2009) 34 Ultrasound in Obstetrics & Gynecology, 25, 32.

²¹⁶ Ibid.

²¹⁷ ASW (n120), standard 7.6, FASP (n121), point 5.9.

²¹⁸ Favre and others (n215), 30.

²¹⁹ Ingvild Aune and Anders Möller, ‘I want a choice, but I don’t want to decide’—A qualitative study of pregnant women’s experiences regarding early ultrasound risk assessment for chromosomal anomalies’ (2012) 28 Midwifery 14, 23.

the final decision in relation to antenatal screening.²²⁰ The main reasons for wanting to place the decision-making power into the hands of the profession were due to social pressure, emotion (typically anxiety and stress) and control.²²¹

Supporting Choice Following Screening and Testing

A growing body of research is suggesting that parents, who receive a positive diagnosis for a trisomy following testing, are repeatedly presented with the option to terminate on several occasions throughout the pregnancy, despite declining this offer at the first instance.²²² ASW and FASP outline that parents should be offered bespoke counselling before deciding whether to continue with the pregnancy or not, and must not be coerced to make decisions in this regard.²²³

While termination will be presented as an option following a positive diagnosis, clinical guidelines also outline that palliative care and/or postnatal support should be available to the parents who decide to continue with the pregnancy.²²⁴ It was noted by the UK NSC that counselling parents on PS and ES will be very different to that of DS, requiring sensitive and informed disclosure of information on each condition.²²⁵ For example, parents, who have had a positive antenatal diagnosis of their baby having either ES or PS, will be asked whether they would want their baby to be resuscitated at birth; this often causes extreme emotional distress and confusion among parents in this population.²²⁶

2.6.5 Communication and the HCP-Parent Relationship

Referring to the decision in *Montgomery*, FASP and ASW guidelines both signify the importance of building and maintaining a dialogue of communication between parent and professional from the beginning of the screening experience.²²⁷ The guidelines specify that interactivity and shared decision-making, between parent and professional, is a key to

²²⁰ Ibid.

²²¹ Ibid.

²²² Brian Skotko, 'Mothers of children with Down syndrome reflect on their postnatal support', (2005) 115 Paediatrics 64.

²²³ ASW (n120), standard 8.2, FASP (n121), point 6.1.

²²⁴ Ibid, standard 8.3, Ibid, point 6.2.

²²⁵ UK NSC (n36).

²²⁶ Author unnamed, 'Never say never about our child', (2015) 350 British Medical Journal 1246.

²²⁷ ASW (n120), standard 7.8, FASP (n121), point 6.

successfully secure informed consent.²²⁸ Indeed, NICE outline that effective communication is essential to shared decision-making, and to protect reproductive autonomy.

Mordel sought to enhance communication not only between the parent and HCP, but also between the different professional groups operating under the established system of consent – namely sonographers and midwives – throughout the pathway. Jay J suggested that current systems of consent would benefit from improved interdisciplinary and interprofessional communication in clinical practice.

The GMC notes that a model of care based on shared decision-making requires HCPs to tailor the information to the needs, wishes and priorities of patients.²²⁹ However, the GMC emphasise that an individual may want more or less information or involvement in the decision-making process which must be appreciated by HCPs.²³⁰ For example, a study by Rowe *et al.* revealed that delivering too much information, before screening, could be detrimental to the bond the parents have with their unborn child, inducing anxiety and stress.²³¹

The GMC and NMC state that effective communication should also extend to clearly and accurately answering questions from parents.²³² The NICE guidelines acknowledge that parents should be given the “opportunity to discuss issues and ask questions”.²³³ ASW and FASP state that if parents ask any questions before or after receiving the information on trisomy screening and testing on its purpose, implications, limitations and benefits, these must be answered clearly and satisfactorily.²³⁴ The NMC highlights that, to achieve this, HCPs must work in partnership with patients, encouraging and empowering them to share in decisions about their preferred methods of treatment and care.²³⁵ The existence of anxiety or distress should be met with compassion and politeness.²³⁶ The GMC also state that “no

²²⁸ Ibid, Ibid.

²²⁹ General Medical Council – Consent: Patients and Doctors Making Decisions Together (2008), part 2 (7).

²³⁰ Ibid, part 1 (4).

²³¹ Heather Rowe, Jane Fisher and Julie Quinlivan, ‘Women who are well informed about prenatal genetic screening delay emotional attachment to their fetus’, (2009) 30 J Psychosom Obstet Gynaecol 34, 41.

²³² GMC (n229) part 2 (10), NMC (n122) standard 2.

²³³ NICE (n123), point 1.1.1.5.

²³⁴ ASW (n120) standard 7.1 (2), FASP (n121), point 5.5.

²³⁵ NMC (n122), standard 2.1, 2.2, 2.3, 2.4 and 2.5.

²³⁶ Ibid, standard 2.6.

single approach to discussions about treatment or care will suit every patient or apply in all circumstances".²³⁷

Midwives are required, as far as realistically possible, to create a comfortable environment for parents.²³⁸ While the clinical setting often conjures anxiety and fear, midwives should listen to parents to attempt to manage or alleviate these potentially harmful emotions, to facilitate effective communication and rapport with parents.²³⁹ The GMC states that a partnership should be built on trust and openness.²⁴⁰

Approach to HCP-Parent Communication

As NICE, ASW and FASP highlight, when delivering information on trisomy screening and testing, HCPs must remain unbiased, neutral and non-directive.²⁴¹ The term 'non-directiveness' relates to the exercise of removing personal views or opinions during the course of counselling, adopting an impartial or neutral role when delivering information and advice.²⁴² However, a body of research has underlined concern towards a "culture of bias" emerging from HCPs, and the institution in which they are situated, in favour of the provision of trisomy screening.²⁴³ Avoiding the use of loaded or emotive terms is also a key objective, according to clinical guidelines.²⁴⁴

Adopting a non-directive approach to care management is key to preserving reproductive autonomy, as it places discretion in the hands of the parents when executing choices, mitigating the presence of unregulated paternal practices.²⁴⁵ From a clinical perspective, the doctrine of non-directiveness is also a desirable approach to care management, as it removes or distances the emotion from the HCPs professional work.²⁴⁶ Clarke explains that distancing

²³⁷ GMC (n229) part 1 (4).

²³⁸ Reid, B., Sinclair, M., Barr, O., Dobbs, F. and Crealey, G., "A meta-synthesis of pregnant women's decision-making processes with regard to antenatal screening for Down syndrome", (2009), 69 Soc Sci Med, 1561, 1573.

²³⁹ Dahl, K., Hvidman, L., Jørgensen, F. & Kesmodel, 'Knowledge of prenatal screening and psychological management of test decisions', (2011) 38 Ultrasound in Obstetrics & Gynecology, 152, 157.

²⁴⁰ General Medical Council – Consent: Patients and Doctors Making Decisions Together (2008), part 1 (3).

²⁴¹ NICE (2008).

²⁴² Angus Clarke, 'The Process of Genetic Counselling – Beyond Non-Directiveness', (2011) 23 British Medical Journal, 179.

²⁴³ Clare Williams, Priscilla Alderson and Bobbie Farsides, 'Is nondirectiveness possible within the context of antenatal screening and testing?', (2002) 54 Soc Sci Med 339, 347.

²⁴⁴ Ibid.

²⁴⁵ Ibid, 340.

²⁴⁶ Angus Clarke (n242), 179.

personal emotion from the professional work also “assures that the clinician will far less likely to be subject to litigation for the decision made by the parents”.²⁴⁷

Williams *et al.* underline that adopting non-directive approach to care management may extend beyond the individual role of the HCP, to the institution itself.²⁴⁸ The presence of ‘structural’ or ‘institutional directiveness’ relates to how a programme or pathway is designed, which could inadvertently pressurise or encourage parents to reach a reproductive choice.²⁴⁹ While there are “misguided professionals” who may not adopt a non-directive approach, by failing to eradicate their own personal views from the communication of information and advice, structural directiveness risks placing an “unconscious pressure” on parents, creating a wider environment of coercion.²⁵⁰ Structural directiveness is said to be a significant issue, in the context of antenatal screening and testing, with studies finding that parents are carried away by the testing pathway, due to the programme’s design and presentation.²⁵¹

Time constraints are described as a prominent barrier to effective communication in the context of screening.²⁵² While growing time constraints pose significant challenges for HCPs to effectively build rapport, clinical guidelines state that HCPs must check that they dedicate sufficient time, in the pre-screening and testing consultations, to explain to parents what trisomy screening and testing involves, and the possible result they could receive.²⁵³ The Royal College of Surgeons’ guidance on consent and supported decision-making acknowledge that, while time pressures can often leave an insufficient amount of time to discuss, at length, the course of available treatment options, “... this does not change the fundamental legal requirement that ... doctors allocate sufficient time for discussion that will allow them to understand the individual patient and their needs”.²⁵⁴

²⁴⁷ Ibid, 182.

²⁴⁸ Clare Williams, Priscilla Alderson and Bobbie Farsides (n243), 346.

²⁴⁹ Angus Clarke, (n242), 180.

²⁵⁰ Ibid.

²⁵¹ Maria Tsouroufli, *‘Routinisation and constraints on informed choice in a one-stop clinic offering first trimester chromosomal antenatal screening for Down’s syndrome’*, (2011) 27 *Midwifery* 431, 436.

²⁵² Ibid, 433

²⁵³ Ibid, 435

²⁵⁴ Royal College of Surgeons. *Consent: Supported Decision-Making – A Guide to Good Practice* (2016), point 4.11.

In a study conducted by Lewis *et al.*, many parents highlighted the importance of building a continued dialogue with HCPs throughout the trisomy screening pathway.²⁵⁵ They also emphasised the need to take time to consider the option of testing, enabling them to gather information and materials from the midwife, and online, to make informed decisions.²⁵⁶ Newton underlines concern that NHS time constraints hinders an open and continued dialogue, raising questions around whether parents are given sufficient time to discuss screening options, and “refine one’s attitudes towards disability or impairment”.²⁵⁷

The NMC, FASP and ASW stress that HCPs must protect parents’ best interests by understanding their needs, wishes and expectations of screening.²⁵⁸ The GMC note that assumptions should not be made in relation to the information they may want or need, a patient’s level of knowledge or understanding, or the factors that patients may consider significant or material.²⁵⁹ To assume the patient’s best interests would be indicative of a paternalistic model of care.²⁶⁰

Williams *et al.* opines that a crucial balance must be struck between the competing interests of both HCPs and parents.²⁶¹ Placing the sole decision-making responsibility in the hands of the parents are not always in their best interest. Some may feel deeply uncomfortable with making the final decision, possibly due to not knowing what the best interests are for the baby, and for themselves. Other parents may prefer to merely receive the information from HCPs to form their own decision, with limited input from the HCP. This points towards a shared decision-making model of care management, with the support of clinicians available to make an informed final decision. It is reported that a model of care, that is strictly patient-autonomous, could result to parents making a decision that they feel is not truly informed, exacerbating anxiety, stress or fear.²⁶²

²⁵⁵ C. Lewis *et al.* (n5), 232.

²⁵⁶ *Ibid.*

²⁵⁷ Ainsley J. Newson, ‘*Ethical aspects arising from non-invasive fetal diagnosis*’, (2008) 13 *Seminars in Fetal and Neonatal Medicine*, 106.

²⁵⁸ NMC (n122), standard 9, ASW (n120), standard 8.3, FASP (n121), point 5.9.

²⁵⁹ GMC (n229), point 1.1.2.

²⁶⁰ Clare Williams and others (n243), 339.

²⁶¹ *Ibid.*, 341.

²⁶² *Ibid.*, 342.

2.6.6 'Risk' in the Context of Trisomy Screening

Clinical guidelines state that the midwife should support parents' understanding of 'risk' for trisomy screening; in this context, supporting parent understanding of the difference between screening and testing is required.²⁶³ *Montgomery* established that a patient should be aware of any 'material' risks associated with a proposed course of treatment, and of available alternative options: materiality is judged in accordance with the reasonable person in the patient's position. This case also underlined that the HCPs 'advisory' role involves a continued dialogue, with the aim of ensuring the patient understands the risks and benefits associated with the treatment.

NICE guidelines state that at first contact, midwives should explain to parents that screening does not provide a definitive result, and a full explanation of the risk-score they receive should follow.²⁶⁴ Guidelines also outline that it should be made clear that screening will produce a risk-score, or likelihood, of the baby having one of the tested conditions; this will not provide parents with a definitive answer.²⁶⁵ It is recommended that a follow-up investigative test should be conducted to obtain a definitive answer.²⁶⁶

First Trimester Screening

The accuracy of combined screening method of screening must be clearly explained. This method of screening has a reported "accuracy" rate of 84-90%, depending on the study.²⁶⁷ However, the term 'accuracy', in the context of combined screening, is misleading. To better understand the performance or true 'accuracy' of the combined screening test, consideration of the test's sensitivity, specificity, positive and negative predictive values is required.²⁶⁸

Sensitivity

Sensitivity is a measure of the tests performance; that is, how many of those with a tested condition will be identified as screen-positive. This is also referred to as the detection rate.

²⁶³ ASW (n120), standard 7, FASP (n121), point 5.

²⁶⁴ NICE (n123), point 1.7.2.5.

²⁶⁵ ASW (n120), standard 7, FASP (n121), point 5.

²⁶⁶ *Ibid*, *Ibid*.

²⁶⁷ Tom Shakespeare (n12), 6.

²⁶⁸ *Ibid*.

The sensitivity of the combined test for DS only, and DS, ES and PS together, is 84%.²⁶⁹ This figure is usually quoted as the test's 'accuracy'. Therefore, the false-negative rate of combined screening is 16-17%.²⁷⁰ This means that 1-2 in 10 babies, born with DS, ES or PS, are low-risk following combined screening.²⁷¹ This is why sensitivity is used as a method of screening, as it can highlight that there is a difference, but is unable to specifically determine what the difference is.

Specificity

Specificity is a measure of the test performance; that is, it is a measure of how many of those without the condition will be identified as screen-negative. The specificity of the combined test for DS only, and DS, ES and PS together, is 98%.²⁷² Therefore, the false-positive rate of combined screening is between 2-5%, depending on the study.²⁷³ This means that 2-5 in 100 babies born without DS, ES or PS will have a high-risk result, following combined screening. Tests that have a high specificity are used to make a definitive diagnosis on the tested condition.

Positive Predictive Value

A positive predictive value (PPV) relates to how many of those who had a screen-positive result truly had a tested condition. PPV should not be confused with sensitivity, as the focus is different for both. The focus for PPV is on whether those parents, who are placed in the high-risk category, truly had a baby with a tested condition. Furthermore, the prevalence of the tested conditions among populations are considered when calculating PPV, while the test's sensitivity is independent of this factor. The PPV will increase or decrease depending on the prevalence of the tested condition. In theory, as the prevalence of the tested condition

²⁶⁹ Ibid, 8. Other sources place the figure at 90% for Down's Syndrome, and 95% for Edwards' and Patau's Syndrome; see, Maria Angeles Sanchez-Duran, Andrea Bernabeu Garcia and Elena Carreras, 'Clinical Application of a Contingent Screening Strategy for Trisomies with cell-free DNA: A Pilot Study', (2019) 121 BMC Pregnancy and Childbirth 1, 18.

²⁷⁰ Francesco Padula and others, 'Retrospective study evaluating the performance of a first-trimester combined screening for trisomy 21 in an Italian unselected population', (2014) 8 Journal of Prenatal Medicine 50, 56.

²⁷¹ This was raised in a conversation I had with a sonographer and fetal medicine consultant. Begs the question why NIPT is only offered to screen-positive parents considering the false-negative statistics.

²⁷² Tom Shakespeare (n12), 8.

²⁷³ K. Schuchter, E and others, 'The First Trimester 'Combined Test' for the Detection of Down Syndrome Pregnancies in 4939 Unselected Pregnancies', (2002) 22 Prenat Diagn 211, 215.

increases, the PPV should also increase, as the number of false positives will be higher in the overall sum of true positives.

The PPV of the combined test, for DS only, is 10-11%.²⁷⁴ This means that of 10 parent who are placed in the higher-risk, or screen-positive, category following the test, only 1 will truly have a baby with DS. In other words, 90% of women in the higher-risk category will be carrying a typical karyotype baby; therefore, less than 10% of those placed in the higher-risk category will be carrying a baby with DS.²⁷⁵ The PPV of the combined test, for DS, ES and PS together, is slightly higher, at 13%.²⁷⁶ This means that of 10 parent who are placed in the higher-risk, or screen-positive, category following the test, only 1 will truly have a baby with either DS, ES or PS.

Negative Predictive Value

A negative predictive value (NPV) relates to how many of those, who had a screen-negative result, truly did not have a tested condition. NPV should not be confused with specificity, as the focus is different for both. The focus for NPV is on whether those parent who are placed in the low-risk category truly do not have a baby with a tested condition. Furthermore, similarly to PPV, the prevalence of the tested conditions among populations are considered when calculating NPV, while specificity is independent of this factor. The NPV will increase or decrease depending on the prevalence of the tested condition. As prevalence of the tested condition increases, the NPV should decrease as, in theory, the number of true negatives will be higher in the overall sum of false-negatives.

The NPV of the combined test for DS only, is 99.95%. This means that out of 10 parents, who are placed in the lower-risk or screen-negative category following the test, rarely (statistically) will the baby have DS. The NPV of the combined test for DS, ES and PS together, is >99.9%.²⁷⁷ This means that of 10 parents, who were placed in the lower-risk or screen-negative category following the test, rarely (statistically) will the baby have either DS, ES or PS.

Research has outlined that supporting parent understanding of 'risk' for screening is notoriously difficult, particularly within the confines of the booking appointment. John *et al.*

²⁷⁴ Tom Shakespeare (n12), 8.

²⁷⁵ Heyman (n184) 2360.

²⁷⁶ Ibid.

²⁷⁷ Ibid.

also identified that midwives experience difficulty communicating the concept of ‘risk’, with some midwives, in their study, incorrectly defining the lower-and higher-risk categories, during pre-screening counselling, or inaccurately describing the ‘accuracy’ of screening.²⁷⁸

ASW and FASP note that an explanation should be provided to parents as to the management of results.²⁷⁹ FASP reinforce that HCPs should notify parents of the period of time which they should be expected to wait for the results post-screening and testing, as the process of waiting can cause significant emotional stress, anxiety and harm.²⁸⁰ FASP state that results should be delivered within 2 weeks, or 2-3 days, depending on the risk-score.²⁸¹ ASW²⁸² FASP and NICE guidelines indicate that following a result, parents should have rapid face-to-face access with their midwife to discuss the result, or counselling by ‘trained staff’²⁸³; this would allow parents the opportunity to ask the HCP any questions about their result(s), and what it means for them, or highlight concerns or issues they may have; this requires HCPs to discuss the next steps, and available options, with the parents.

NICE, FASP and ASW outline that midwives are required to describe in simple, but accurate, terms what ‘low-and high-risk’ results mean, in the context of trisomy screening.²⁸⁴ Research underlines a risk that parents interpret a high-risk screening result to mean that the baby has a tested condition.²⁸⁵ Likewise, research has identified a risk that parents who receive a low-risk result, or screen-negative result, interpret the result to mean the baby does not have a tested condition. This misinterpretation of screening’s purpose is said to invoke emotions of anxiety and stress following discordant results, which are often difficult to effectively manage.²⁸⁶

Thus, a clear and accurate explanation of how parents are placed in the different categories of ‘risk’ should be conducted by the midwife at the booking appointment, according to

²⁷⁸ Sophie John (n15), 5.

²⁷⁹ ASW (n120), standard 7.6, FASP (n121), point 5.4

²⁸⁰ Ibid.

²⁸¹ Ibid.

²⁸² ASW (n120), standard 7.7.1, standard 25 – states that an appointment should be made within 24 hours of receiving high-risk result.

²⁸³ NICE (n123) point 1.7.2.6.

²⁸⁴ NICE (n123) point 1.7.5, ASW (n120) standard 7.7.2, FASP (n121), point 5.8.

²⁸⁵ Sophie John (n15), 5.

²⁸⁶ Ibid, 6.

guidelines.²⁸⁷ Policy dictates that a parent who scores 1:150 risk, or more, will be considered high-risk, or screen-positive, for having a baby with DS, ES or PS. Only 5:100 of parents in the general obstetric population will be placed into the higher-risk category following screening. Guidelines instruct midwives to explain that a higher-risk score does not necessarily mean the baby has a tested condition; the majority of women who are placed in the high-risk category, following screening, will *not* have a baby with a tested condition.²⁸⁸

A parent who scores 1:150, or less, will be considered low-risk, or screen-negative, of having a baby with one of the tested conditions.²⁸⁹ Over 95:100 of parents in the general obstetric population will be placed in the lower-risk category following screening. However, guidelines instruct midwives to explain that a lower-risk score does not necessarily mean the baby does not have a tested condition.²⁹⁰

NICE guidelines state that the screening pathway for both low-risk and high-risk populations should be explained by midwives to parents at the booking appointment.²⁹¹ These categories have been established to efficiently group those parents who should be offered further investigative testing, or not. Those parents who score high-risk will be promptly notified via a preferred method of communication of the result, and will be offered further investigative testing.²⁹² Those parents who score low-risk will not usually be offered any further investigative testing.

Invasive Testing

Clear and accurate disclosure of the risk of amniocentesis and CVS should be undertaken during counselling, following a higher-risk result, according to NICE, FASP and ASW.²⁹³ However, research has demonstrated concern towards the inconsistent use of statistics, in the context of risk for procedural miscarriage. As discussed previously, the risk of procedural miscarriage from invasive testing varies between research studies, ranging from 1-2%.

²⁸⁷ ASW (n120) standard 7.7, FASP (n121) point 6.7.

²⁸⁸ Ibid, Ibid.

²⁸⁹ Ibid, Ibid.

²⁹⁰ Ibid, Ibid.

²⁹¹ NICE (n123), point 1.7.2.5.

²⁹² ASW (n120) standard 8, FASP (n121) point 7.

²⁹³ NICE (n123), 1.7.2.3, ASW (n120) standard 7.6, FASP (n121), point 8.

John *et al.* stress that the ‘risk’ of procedural miscarriage is often trivialised during pre-testing consultations, with midwives undermining its significance, by using terminology such as ‘rare’ and ‘very unlikely’, before securing consent.²⁹⁴ However, commentators underline that, while the risk of invasive testing is objectively ‘small’, its consequences are significant to parents, if the risk materialises.²⁹⁵

2.6.7 ‘Alternative’ Methods of Screening and Testing

Non-invasive Prenatal Testing (NIPT)

The latest contribution to the screening method’s timeline, is the implementation of non-invasive prenatal testing (NIPT) to trisomy screening pathways. Alexander *et al.* note that NIPT technology has developed rapidly, describing it as a “paradigm shift” in the performance of antenatal screening around the world.²⁹⁶ However, with this rapid development, comes unprecedented legal and ethical implications, that frame the provision of supporting parent consent for NIPT along the NHS trisomy screening pathway.²⁹⁷ Minear *et al.* highlighted that the changing nature of antenatal screening and testing has “exacerbated the need for effective counselling and education of both antenatal care providers and patients”.²⁹⁸

NIPT has been available on the private market in the United Kingdom since 2011, however it has recently been introduced to NHS trisomy screening pathways.²⁹⁹ Non-invasive technology is said to have revolutionised the screening landscape, as the test only requires a sample of the maternal serum (blood) to detect the presence of a trisomy.

Literature also refers to NIPT as ‘cell-free fetal DNA’ screening. Leading medical scholars discovered that when a human cell dies, it fragments and releases DNA from its nucleus into the surrounding blood: these are better known as cell-free DNA (cfDNA) fragments.³⁰⁰ Professor Dennis Lo discovered that, in pregnancy, the life cycle of a cell shortens, releasing

²⁹⁴ Sophie John (n15), 6.

²⁹⁵ Ibid.

²⁹⁶ Elizabeth Alexander, Susan Kelly, Lauren Kerzin-Storarr, “Non-invasive prenatal testing: UK genetic counselors’ experiences and perspectives”, *Journal of Genetic Counseling*, 2015, 24(2), 300-311.

²⁹⁷ Ibid.

²⁹⁸ Mollie A. Minear *et al.* (n130), 370.

²⁹⁹ Tom Shakespeare (n12), 20.

³⁰⁰ Royal College of Obstetricians & Gynaecologists, ‘Non-invasive Prenatal Testing for Chromosomal Abnormality using Maternal Plasma DNA’, (2014) 15 Scientific Impact Paper, 1-14.

increased levels of cfDNA into the blood.³⁰¹ Lo also realised that a high portion of the cfDNA, released into the maternal blood, originated from the placenta.³⁰² Analysing this cell-free fetal DNA (cffDNA) in the blood – that originates from placenta – meant that this test was introduced as being a more ‘accurate’ method of screening for trisomy, than the conventional methods (combined and/or quadruple screening).

Implementation of NIPT to Screening Pathway

The recent UK NSC recommendation, to extend the DS screening pathway to screen for PS and ES, was accompanied by the recommendation to evaluatively roll-out NIPT, as a method of *screening* to the pathway, by NHS England and Wales. In 2016, the UK NSC stated that the intended implementation of NIPT was to be executed evaluatively by the NHS over an initial 3-year period – subject to their recommendations – to assess what impact it would have on the current antenatal screening and testing programmes, across England and Wales.³⁰³ More specifically, the recommendation provided by the UK NSC was made to evaluate the impact of offering NIPT, as a contingency option, to parents where their combined screening risk-score for DS, is greater, or equal to 1:150, and the risk-score for ES and PS is greater than, or equal to, 1:15 (i.e the predefined high-risk groups).³⁰⁴

The Welsh Government consulted ASW to lead the combined implementation of the UK NSC’s recommendation, to offer NIPT as a contingency test for higher-risk pregnancies, into the new trisomy pathway. In April 2018, NHS Wales, with ASW, had finalised a combined implementation of screening for PS and ES into the new pathway, along with a 3-year evaluative roll-out of NIPT.³⁰⁵ In 2016, NHS England, with FASP, had implemented the extension to the trisomy pathway to include screening and testing for PS and ES; however, to date, FASP have not finalised plans to standardise the roll-out NIPT across all Heath Trusts in England.³⁰⁶ It is important to note, however, that some Health Trusts, in England, provide

³⁰¹ Kater-Kuipers A., E. M. Bunnik, I. D. de Beaufort and R. J. H. Galjaard. ‘Limits to the scope of non-invasive prenatal testing (NIPT): an analysis of the international ethical framework for prenatal screening and an interview study with Dutch professionals’ (2018) 18 BMC Pregnancy Childbirth, 409.

³⁰² Ibid.

³⁰³ UK National Screening Committee, Public Health England Screening, “UK NSC Non-Invasive Prenatal Testing (NIPT) Recommendation”, 2016, available at:

https://legacyscreening.phe.org.uk/policydb_download.php?doc=602 (accessed 28/06/2019).

³⁰⁴ Ibid.

³⁰⁵ Public Health Wales. NHS Wales Antenatal Screening Wales Annual Report (2018-2019) 1-32.

³⁰⁶ A standardised implementation was finalised by June 2021.

NIPT on the NHS, such as Liverpool Women's, whilst others at a discounted price, or offered privately.

The Nuffield Council on Bioethics were commissioned, in 2017, to produce a report that considered the ethical, legal and regulatory implications of NIPT.³⁰⁷ This report provided a comprehensive evaluation of NIPT's use, in the NHS and private services, concluding that it should only be offered where couples are fully informed of its implications, to protect parent autonomy and choice. With the UK NSC providing complicated, combined recommendations at this time, this report attempted to achieve clarity, in relation to the implementation of NIPT, into first trimester screening.

The RAPID evaluation study also investigated the impact of implementing NIPT into NHS antenatal care pathways.³⁰⁸ This report was used to inform the UK NSC's recommendation to implement NIPT into existing antenatal screening pathways. This study considered the test's performance, including its benefits and disadvantages, uptake, outcomes and consequences, for both parents and HCPs.³⁰⁹ This study also concluded that the test would reduce the need for invasive procedures and could be easily integrated into existing screening arrangements.³¹⁰ According to conclusion of this research group, the study had successfully developed the requisite training programme, and information materials, for both HCPs and parents.³¹¹

The Royal College of Midwives (RCM), in 2018, stated that, in anticipation of the implementation of NIPT, training events must be provided, which were attended by over 400 NIPT 'champions', representing all NHS Trusts across England and Wales.³¹² These NIPT champions were responsible for ensuring that HCPs, in their relevant trusts, undertook cascade training, which included midwives, obstetricians, sonographers and fetal medicine

³⁰⁷ Tom Shakespeare (n12).

³⁰⁸ Lyn Chitty and others, '*RAPID non-invasive prenatal testing (NIPT) evaluation study – a report for the UK National Screening Committee*', (2015) Executive Summary, 2015, 1-8.

³⁰⁹ Ibid, 6

³¹⁰ Ibid.

³¹¹ Ibid.

³¹² Royal College of Midwives: 'Training for NIPT' (2018): <https://www.rcm.org.uk/news-views/rcm-opinion/rolling-out-nipt/> (accessed 10/12/2019).

practitioners.³¹³ This was completed in 2018, however the standardised implementation of NIPT was not finalised, raising questions on whether HCP would need re-training.

As stipulated by NHS policy standards, if the parent receives a higher-risk result following combined screening, they will be offered non-invasive prenatal testing (NIPT), as a contingency screening test, either privately or on the NHS, depending on national policy. This method of screening comprises of taking only a sample of the mother's blood, which carries no risk of harm to mother or baby. This method of screening is not diagnostic, and therefore it is correct practice to offer further investigative screening, to confirm whether or not the baby has a chromosomal anomaly. Investigative testing is usually conducted by amniocentesis or chorionic villus sampling (CVS), to provide a definitive diagnosis.

Scholars argue that the recent extension to first trimester screening, and the implementation of NIPT, will only pose additional challenges to "an already beleaguered informed consent regime".³¹⁴ Minear *et al.* emphasise that the unprecedented speed at which screening technology is developing, compounded by its expanding panel of tested conditions, could put informed decision-making and consent at serious risk.³¹⁵ According to these academics, with the new methods of screening and additional conditions being screened for, routine breaches to current standards of consent are highly probable.³¹⁶

Understanding NIPT

ASW state that, in pre-screening discussions with parents, midwives must describe the purpose, implications, limitations and benefits of NIPT, clearly and accurately.³¹⁷ The primary purpose of implementing NIPT, on the NHS, was to provide a contingency test to high-risk populations, offering the choice to screen for DS, ES and PS. NIPT has a higher detection and lower false-positive/negative rate than traditional trisomy screening methods (combined and quadruple).

Due to the risk of miscarriage associated with the invasive diagnostic methods of testing (around in 1 in every 100 babies being lost through the procedure), the implementation of

³¹³ Ibid.

³¹⁴ Mollie A. Minear *et al.* (n130), 373.

³¹⁵ Ibid.

³¹⁶ Ibid.

³¹⁷ ASW (n120), standard 7.1 (2).

NIPT was intended to reduce the number of invasive tests that were being performed, as NIPT is conducted by taking a sample of the maternal blood, for the purpose of analysing cffDNA, to discover whether the baby has either DS, ES or PS.

The UK NSC outlined research which suggested the number of invasive tests would decrease from 7,900 to 1,400 each year, resulting to a decrease in the number of procedural-related miscarriages, from 46 to 3 each year.³¹⁸ Indeed, early evidence indicates that the number of invasive procedures being performed in Wales, post-implementation of NIPT, has reduced.³¹⁹

However, research conducted by Cernat *et al.* into women's preferences to facilitate better informed decision-making on NIPT, revealed that parents were dissatisfied and disappointed with their consultation discussions with HCPs.³²⁰ Parents felt that the HCPs were not sufficiently informed about the technology to facilitate informed choice.³²¹ In particular, parents felt that the perceived lack of knowledge of HCPs was pronounced when discussing the conditions NIPT is able to detect (DS, ES and PS).³²² Those parents who had a baby with a tested condition emphasised that HCPs' lack of knowledge originated from an unfamiliarity with the conditions and lived experience, which hindered informed choice.³²³

Non-invasive prenatal screening, in the earlier stages of the pregnancy, is reported to provide "an array" of benefits for parents.³²⁴ As NIPT can be conducted earlier in pregnancy (around 9-10 weeks), with results being accessible quickly, it is reported to reduce stress and anxiety, and enhance the bond between parent and unborn child.³²⁵ Furthermore, Lewis *et al.* outline

³¹⁸ Anne Mackie, Public Health Screening England, 'Addition of Non-Invasive Test to Improve Screening for Pregnant Women', (2016) NHS Fetal Anomaly Screening Programme, UK National Screening Committee, 2016, available at: <https://phscreening.blog.gov.uk/2016/11/03/addition-of-non-invasive-test-to-improve-screening-for-pregnant-women/> (accessed 28/06/2019).

³¹⁹ NHS Wales Shared Service Partnership, Welsh Government, "Voluntary ex ante transparency notice", Section 2, Subsection 2(4), 2019, available at: https://www.sell2wales.gov.wales/Search/show/Search_View.aspx?ID=MAR287966 (accessed 05/06/2019).

³²⁰ Alexandra Cernat and others, 'Facilitating informed choice about non-invasive prenatal testing (NIPT): a systematic review and qualitative meta-synthesis of women's experiences', (2019) 19 BMC Pregnancy and Childbirth, 9.

³²¹ *Ibid.*, 10.

³²² *Ibid.*, 12

³²³ *Ibid.*

³²⁴ Antina de Jong, Wybo J Dondorp and others, 'Non-invasive prenatal testing: ethical issues explored', (2010) 18 European Journal of Human Genetics 272, 277.

³²⁵ Ruth M. Farrell and others, 'It's More Than a Blood Test: Patients' Perspectives on Noninvasive Prenatal Testing' (2014) 3 J Clin Med. 614, 631.

that NIPT reduces psychological harm among parents who have a positive diagnosis for a trisomy, as terminations could be performed earlier in the pregnancy.³²⁶

It is also reported that NIPT possesses practical benefits: it does not require specialist clinical skills or equipment to perform.³²⁷ The process involves obtaining a sample of the mother's blood, which is subsequently sent to specialist laboratories for analysis.³²⁸ In contrast, the other methods of screening and testing require expert input.³²⁹

A limitation of NIPT is that it is not a diagnostic test, requiring parents to undertake further invasive diagnostic testing, to confirm whether the baby has a tested condition. Ultimately, NIPT is another method of *screening*; therefore, NIPT is only capable of producing another risk-score for the parents, warranting further invasive investigation. A concerning misconception exists among HCPs and parents, that NIPT is similar, or equal to, invasive methods of diagnostic testing, which is fundamentally untrue.³³⁰

This misconception calls into question the requirement, in *Montgomery*, that HCPs should make patients aware of *reasonable* alternative methods of treatment and care, before obtaining their informed consent. Inaccuracies around NIPT's purpose, including its benefits and disadvantages, could ultimately mislead parents' understanding of the test, impacting on their ability to make an informed choice regarding their care management.

The misconception, in part, may originate from the instruction of clinical guidelines when offering NIPT to parents. Guidelines state that midwives should offer NIPT as a safer 'alternative' to invasive methods of testing.³³¹ Indeed, presenting NIPT as a safer 'alternatives', as the RCOG initially did, could be indicative of why NIPT's purpose is fundamentally misunderstood among stakeholders: it is not a replacement for amniocentesis and/or CVS.

While this is ultimately a matter of semantics – and possibly clumsiness when drafting policy guidelines – a growing body of research is beginning to demonstrate concern towards the current contextualisation of NIPT as an 'alternative' method of testing for trisomies. Indeed,

³²⁶ C. Lewis *et al.* (n5), 223-232.

³²⁷ Tom Shakespeare (n12), 4.

³²⁸ *Ibid.*

³²⁹ *Ibid.*

³³⁰ Ruth M. Farrell, Mary Beth Mercer, Patricia K. Agatista, Marissa B. Smith and Elliot Philipson, "It's More Than a Blood Test: Patients' Perspectives on Noninvasive Prenatal Testing" (2014) 3 J Clin Med 614, 620.

³³¹ ASW (n120) standard 7 and 8.

studies reveal that parents, and even HCPs, misunderstand NIPT's purpose as being a *replacement* to invasive testing, and serves the same purpose as invasive testing: to provide a definitive diagnosis. Cernat *et al.* found that NIPT is currently being misinterpreted by parents, and even professionals, as being or "more or less diagnostic".³³² Indeed, Dondorp *et al.* underline that it must be made clear to parents that NIPT is a method of screening, and does not serve the same purpose as invasive diagnostic testing.³³³ Studies have also revealed parents' dissatisfaction following NIPT screening, as they believed invasive testing was not required following NIPT.³³⁴

Dondorp *et al.* explain that invasive diagnostic testing must follow NIPT, as cffDNA is not always concordant with true fetal DNA.³³⁵ These academics conclude by reinforcing that it is fundamental parents, who opt to use NIPT, are fully informed about both the technical and biological limitations to the procedure, before making an informed choice.³³⁶

Indeed, it is important to remember NIPT is a method of screening, and still obtains a false positive rate, that reportedly ranges from 0.3-1%.³³⁷ In a study conducted by Farrell *et al.*, evidence suggested that parents expressed concern for the possibility, and consequences, of false positive results.³³⁸ It was deemed to be a principal consideration when contemplating whether to accept the offer of NIPT.³³⁹ Therefore, it is essential that this possibility is clearly and accurately explained to parents, before undertaking the screening test.

In some circumstances, parents may receive a no result, following NIPT screening. A 'no result' arises in situations where the laboratory has been unable to determine a definitive result, meaning that HCPs are unable to disclose whether the baby has a tested condition.³⁴⁰ Rates

³³² Cernat and others (n320), 20.

³³³ Wybo Dondorp, (n324), 277.

³³⁴ *Ibid.*

³³⁵ *Ibid.*

³³⁶ *Ibid.*

³³⁷ The NHS RAPID Study, available at: <http://www.rapid.nhs.uk/guides-to-nipd-nipt/nipt-for-down-syndrome/> (accessed 29/04/2019).

³³⁸ Ruth M. Farrell *et al.* (n330), 621.

³³⁹ *Ibid.*

³⁴⁰ This may result from a lack of cell-free fetal DNA being present in the maternal blood sample, due to early gestation, high BMI, low PAPP A or low hCG B, a fetus who has either ES or PS, or an issue related to the technology.

of 'no result' vary between 3-12%.³⁴¹ HCPs should offer a second NIPT test considering that the levels of cffDNA increases with gestation.³⁴² There is also a chance that the second attempt at NIPT will not provide a definitive result.³⁴³

The lengthy period of time, that parents are required to wait for NIPT results, is another significant disadvantage. Currently, it takes 1-2 weeks for NIPT results to return, as the bloods are usually sent to other countries who possess the necessary technology. This delay could increase levels of anxiety and stress for parents if termination of the pregnancy is an option, in the event of a positive diagnosis. While terminating a pregnancy is never an easy decision, research suggests that terminations, in the first trimester, are less traumatic than those performed in the later stages of gestation.³⁴⁴

'Accuracy' of NIPT

Correctly defining the 'accuracy' of NIPT has proven to be a complex issue.³⁴⁵ Studies routinely reveal that NIPT is being described as 98-99% "accurate" by HCPs in pre-screening consultations.³⁴⁶ However, its true "accuracy" is not 98-99%. The percentage will decrease or increase depending on the tested conditions, a spectrum of biological factors and the chance category or population that the expectant mother falls into. As with other methods of trisomy screening, better understanding the true accuracy of NIPT involves consideration of specificity, sensitivity, positive and negative predictive values.

Sensitivity

In the general obstetric population, NIPT's sensitivity for DS only, is 96%.³⁴⁷ This figure increases to 97% in the higher-risk population. In the general obstetric population, NIPT's sensitivity, for ES and PS, is 78-87%.³⁴⁸ This figure increases to 93-95% in the higher-risk population. The test's sensitivity varies between the conditions, with DS having a higher

³⁴¹ Public Health England, "NHS Fetal Anomaly Screening Programme (FASP) Screening Pathway for Down's, Edward's and Patau's syndrome: NIPT Cascade Training", 2018, available at: https://drive.google.com/drive/folders/1029Kb93jixr09_6GmLWV7iq9DC37daCZ (accessed 24/06/2019).

³⁴² Ibid.

³⁴³ Ibid.

³⁴⁴ Tom Shakespeare (n12), 42.

³⁴⁵ Ibid.

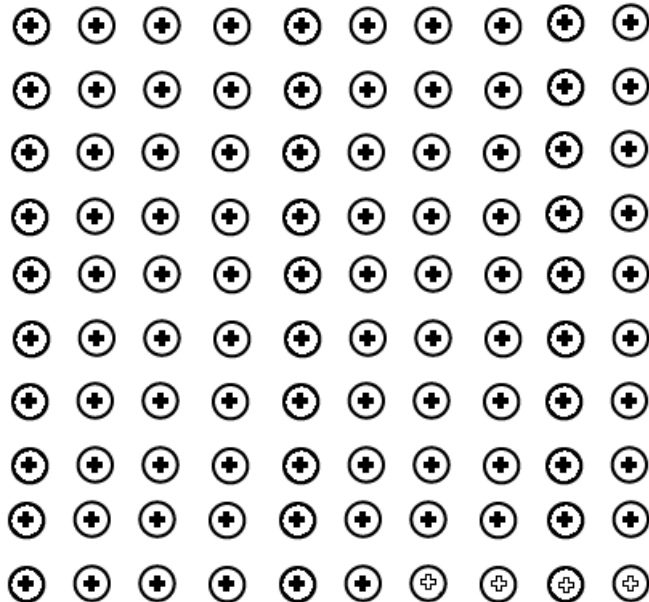
³⁴⁶ D. Petrova and R. Garcia-Retamero. "Can We Improve Risk Communication About Non-Invasive Prenatal Testing?" (2017) 125 BJOG: An International Journal of Obstetrics & Gynaecology, 265-396.

³⁴⁷ Tom Shakespeare (n12), 43.

³⁴⁸ Ibid.

sensitivity than ES and PS, due to their rarity.³⁴⁹ The false-negative rate of NIPT is widely disputed.³⁵⁰ While false-negatives are said to be very rare among medical literature, they can occur.³⁵¹

NIPT's Sensitivity Amongst the General Population for Down's Syndrome³⁵²



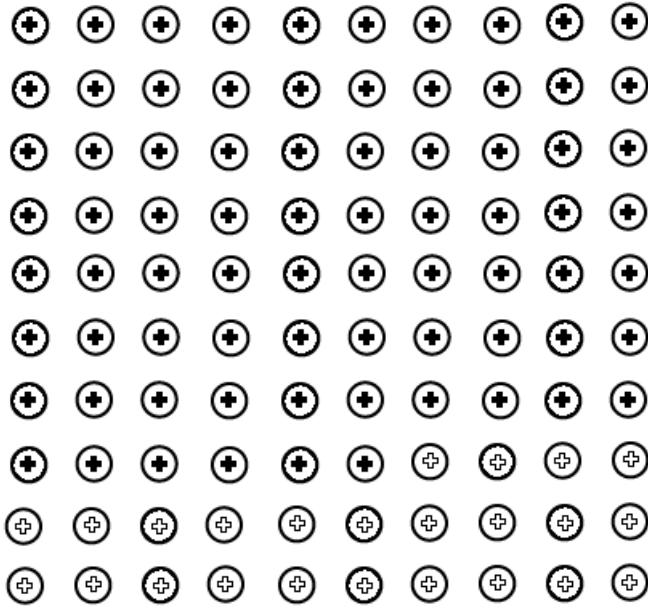
Edwards' Syndrome

³⁴⁹ Ibid.

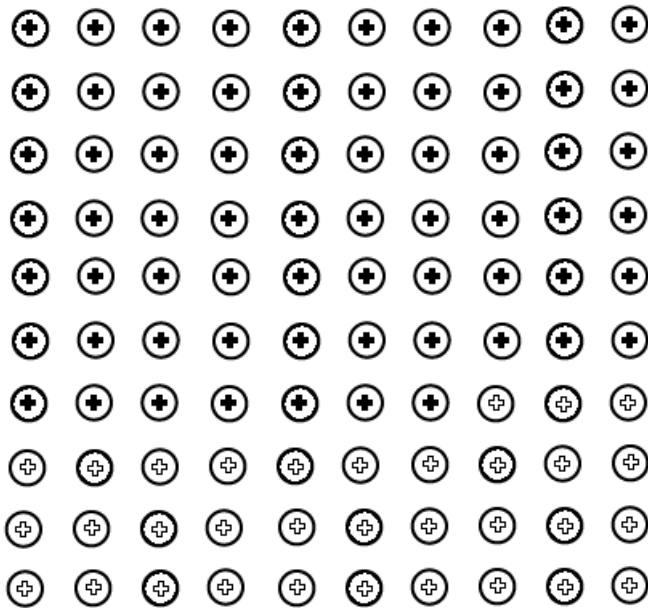
³⁵⁰ Ibid.

³⁵¹ Personally interviewed two women in the process of data collection who had a 1 in 1,000,000 chance of having a baby with DS after NIPT but had a baby with DS. Also interviewed a parent who had a 1 in 50,000 chance of having a baby with DS after NIPT but had a baby with DS.

³⁵² Black Positive represents those who have a baby with a tested condition and test positive, and a white positive represents those who have a baby with a tested condition but test negative (false-negative).



Patau's Syndrome



Specificity

In the general obstetric population, NIPT's specificity, for DS only, is 99.9%.³⁵³ This figure decreases to 99.7% in the higher-risk population.³⁵⁴ In the general obstetric population, NIPT's specificity, for ES and PS, is 99.8-99.9%.³⁵⁵ This figure decreases to 99.7-99.9 in the higher-risk

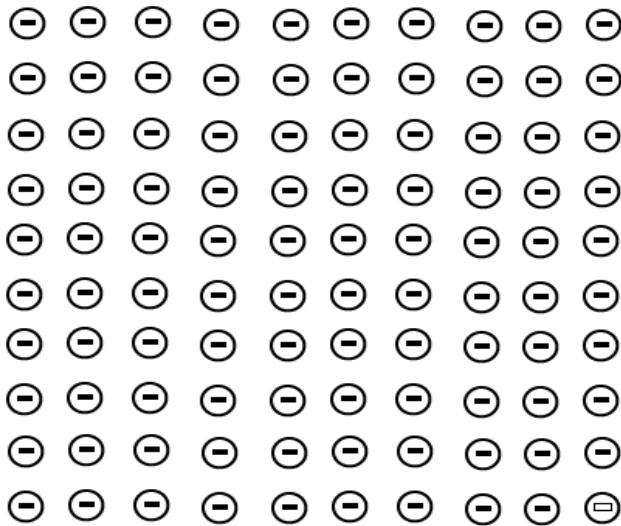
³⁵³ Tom Shakespeare (n12), 44.

³⁵⁴ Ibid.

³⁵⁵ Ibid.

population; therefore, the false-positive rate of NIPT is quoted to be in the region of 0.1-0.3%. Some studies quote the false-positive rate to be as low as 0.09%-0.13%.³⁵⁶

*NIPT's Specificity among the General Population for Down's Syndrome, Edwards' Syndrome and Patau's Syndrome Together*³⁵⁷



Positive Predictive Value

In the general obstetric population, the PPV of NIPT, when screening for DS only, is 82%.³⁵⁸ This means that of 10 parent from the general population, who have a high-risk or screen-positive NIPT result for DS, 8 will truly have a baby with DS. This figure increases to 91%, or 9 out of 10 parents, who are placed in the high-risk population.³⁵⁹ A recent study highlighted that, of those women who opted to screen for DS only, the PPV of NIPT ranges from 28-80% in a patient aged 35, with no other health factors.³⁶⁰

In the general obstetric population, the PPV of NIPT, when screening for ES and PS only, is 37-49%.³⁶¹ This means that of 10 parents from the general population, who have a high-risk or

³⁵⁶ Roberto Romero and Maurice J. Mahoney, 'Noninvasive Prenatal Testing and Detection of Maternal Cancer' (2015) 314 JAMA, 131-133.

³⁵⁷ Black negative represents those who do not have a baby with a tested condition and test negative, and a white negative represents those who do not have a baby with a tested condition but test positive (false-positive).

³⁵⁸ In this context, the general obstetric population refers to those parent who would not have had any prior screening before having the high-risk NIPT result.

³⁵⁹ In this context, the high-risk population refers to those parent who received a high-risk result from prior screening tests (usually combined) and went on to have a high-risk, or screen-positive, NIPT result.

³⁶⁰ Mollie A. Minear *et al.* (n130), 20.

³⁶¹ Tom Shakespeare (n12), 45.

screen-positive NIPT result for ES and PS, only 3-4 will truly have a baby with ES or PS. This figure increases to 84-87%, or 8 out of 10 parents, who are placed in the high-risk population.³⁶² However, as Farrell *et al.* stress, the PPV of NIPT is not well established among the low-risk population.³⁶³ This means 10 women undergo invasive testing, following a high-risk or screen-positive trisomy NIPT result, to discover 8-9 true positives.

Negative Predictive Value

In the general obstetric population, the NPV of NIPT, when screening for DS only, is 99.98%. This figure is 99.91% in the higher-risk population. In the general obstetric population, the NPV of NIPT, for ES and PS only, is 99.99%. This figure is 99.89-99.97% in the higher-risk population.

In a recent study, it was emphasised that parents, and even HCPs, experience significant difficulties when asked to interpret the sensitivity, specificity, positive and negative predictive value of NIPT.³⁶⁴ As a consequence, studies show that parents may misinterpret or fail to understand the likelihood of their baby having a tested condition, based on a positive NIPT result.³⁶⁵ The imperfect discussion of the term 'accuracy' may distract the attention of both HCPs and parents from the limitations of the test's performance.³⁶⁶ Cernat *et al.* highlighted that, in some cases, parents did not feel the need to confirm the NIPT result with invasive methods of testing, as the accuracy of NIPT was "sufficiently high".³⁶⁷ Evans *at al.* emphasise that the primary misunderstanding, in terms of NIPT's 'accuracy', is the failure of HCPs to effectively communicate that NIPT is incapable of definitively determining whether the baby has a tested condition, or not.³⁶⁸

³⁶² Ibid.

³⁶³ Ruth M. Farrell *et al.* (n330), 614.

³⁶⁴ D. Petrova and R. Garcia-Retamero. 'Can We Improve Risk Communication About Non-Invasive Prenatal Testing?' (2017) 125 BJOG: An International Journal of Obstetrics & Gynaecology, 265-396.

³⁶⁵ Ibid.

³⁶⁶ Wybo Dondorp and others, 'Non-invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening.' (2015) 23, 1438.

³⁶⁷ Alexandra Cernat, and others, 'Facilitating informed choice about non-invasive prenatal testing (NIPT): a systematic review and qualitative meta-synthesis of women's experiences" (2019), BMC Pregnancy and Childbirth, 9.

³⁶⁸ Mark I. Evans and others, 'Noninvasive prenatal screening or advanced diagnostic testing: caveat emptor', (2016) American Journal of Obstetrics & Gynecology, 298-303.

HCP are also expected to clearly and accurately describe common factors which affect the performance of NIPT.³⁶⁹ The reported false positive rate of non-invasive prenatal testing is between 0.3-1% for DS, ES and PS, according to the RAPID study.³⁷⁰ A category of biological factors could create a possible source of error.³⁷¹ The RCOG recommend HCPs to disclose that chromosomal mosaicism³⁷², gestational age³⁷³, maternal weight³⁷⁴, maternal conditions or malignancies³⁷⁵, placental mosaicism³⁷⁶ and multiple pregnancies (twins)³⁷⁷ have the potential to impede on the interpretation of NIPT results.³⁷⁸

Benn *et al.* suggest that HCPs, who offer NIPT screening, must be sufficiently trained to ensure clear and accurate information is being delivered to parents on the factors impeding NIPT's detection rate.³⁷⁹ Indisputably, the implementation of NIPT has been ambitious, with many

³⁶⁹ ASW (n120), standard 8.

³⁷⁰ <http://www.rapid.nhs.uk/guides-to-nipd-nipt/nipt-for-down-syndrome/> (accessed 29/04/2019).

³⁷¹ Kater-Kuipers A., E. M. Bunnik, I. D. de Beaufort and R. J. H. Galjaard, 'Limits to the scope of non-invasive prenatal testing (NIPT): an analysis of the international ethical framework for prenatal screening and an interview study with Dutch professionals', (2018) 18 BMC Pregnancy Childbirth, 409.

³⁷² There are two different types of mosaicism where the karyotype in the cytotrophoblast is normal and the fetus itself has a chromosomal anomaly. Crucially, both types of mosaicism will present with typical NIPT results. There is a significant chance that due to the nature of mosaicism, the chromosomal difference will remain undetected following the performance of NIPT.

³⁷³ There is a correlation with gestational age and the levels of cffDNA in maternal blood. The amount of fetal DNA in the blood increases with gestational age. If blood samples are taken too early in pregnancy for NIPT purposes, false-negative results become more likely.

³⁷⁴ Increased maternal weight or an increase in body mass index correlates directly to a lower fetal DNA percentage in the maternal blood, possibly due to a higher cell turnover or a dilution effect caused by the increased volume of blood. As obesity is becoming an increasing problem, predisposing parent to less accurate NIPT results, this maternal factor should be explained in pre-and post-screening consultation and counselling or provided in patient information materials.

³⁷⁵ Maternal chromosomal differences can be the cause of discordant results. Malignant disease such as cancer are very rare, but potential factors which could cause a false positive NIPT result.

³⁷⁶ Placental mosaicism can also lead to false-positive results, where there is a discrepancy between the chromosomal composition of the cells in the placenta and the fetus. In some instances, the chromosomal difference is confined to the placenta and not the baby itself.

³⁷⁷ Twins are commonly dizygotic (non-identical twins) or monozygotic (identical twins), with the vast majority being non-identical. If the twin pregnancy is monochorionic, as both babies either share everything (monochorionic monoamniotic) in the womb (placenta) or at least share everything apart from the amnion (monochorionic diamniotic), they are always concordant for fetal karyotype, thus NIPT will be more effective in detecting the tested conditions. In dichorionic twin pregnancies (each twin has their own chorionic and amniotic sacs) interpreting the results of NIPT would be significantly more difficult, as if one of the babies have either DS, ES or PS, it is highly likely that the other will not. Therefore, the effectiveness of NIPT is significantly impeded, as the 'atypical' placenta can mask the 'normal' placenta.

³⁷⁸ Royal College of Obstetricians & Gynaecologists, "Non-invasive Prenatal Testing for Chromosomal Abnormality using Maternal Plasma DNA", Scientific Impact Paper No.15, March 2014, pp.1-14.

³⁷⁹ Peter Benn, Sharon E. Plon and Diana W Bianchi, 'Current Controversies in Prenatal Diagnosis 2: NIPT Results Suggesting Maternal Cancer Should Always Be Disclosed', (2019) 39 Prenat Diagn, 339.

concerned that HCPs are required to perform a test without a comprehensive educational understanding and appreciation of its purpose and/or implications.³⁸⁰

'Routinisation' of NIPT

A growing body of research is beginning to underline the potential for NIPT's routinisation. Due to its procedural simplicity and ease, scholars are calling for a review of the counselling for NIPT, and the risks surrounding its routine acceptance by parents.

Concerns exist among academics and professionals toward the potential 'routinisation' of NIPT. It is reported that the simple and familiar technique used to perform NIPT has the potential to undermine informed choice.³⁸¹ Deans *et al.* conducted research into the views of stakeholders, finding that the implementation of NIPT had the potential to undermine informed consent, as it is being perceived to be a 'normal' and 'routine' blood test.³⁸² Furthermore, a study by Silcock *et al.* revealed that the simplistic nature of NIPT, and access to the test, could encourage parents to feel that they are acting irresponsibly for not undertaking the test.³⁸³ Trisomy charities express that the availability of NIPT, in conjunction with its procedural simplicity, could result to an accelerated increase in terminations of high-risk trisomy pregnancies.³⁸⁴

Indeed, in the first ever qualitative study conducted into pregnant women's attitudes towards NIPT, many women found that the procedure did not cause any significant stress or anxiety, as the process was very straightforward and easy.³⁸⁵ The participants perceived the procedure as just a blood test, something which parents had routinely undergone throughout the pregnancy, under the screening pathway. While this perception is justifiable, a risk existed that the relatively simple and familiar technique used to perform the test, masked its novel and far reaching implications.³⁸⁶

³⁸⁰ Cernat and others (n367), 12.

³⁸¹ Heather Skirton, Lesley Goldsmith and Lyn S Chitty, '*An Easy Test but a Hard Decision: Ethical Issues Concerning Non-invasive Prenatal Testing for Autosomal Recessive Disorders*', (2015) 23 *European Journal of Human Genetics*, 1004-1009.

³⁸² Zuzana Deans and others, '*Non-invasive prenatal testing for single gene disorders: exploring the ethics*', (2013) 21 *European Journal of Human Genetics*, 713.

³⁸³ Caroline Silcock *et al.* (n210), 1658.

³⁸⁴ Zuzana Deans (n382), 714.

³⁸⁵ C. Lewis *et al.* (n5), 223.

³⁸⁶ *Ibid.*

Kater-Kuipers *et al.* note that, “it is feared that women would step into what is called a ‘screening trap’ ... meaning that NIPT might put women on a pathway to invasive follow-up diagnostic testing and potentially termination of the pregnancy, while they not have fully assessed the consequences beforehand.”³⁸⁷ Lifechanging decisions are at risk of being undermined or trivialised, due to the simplicity of the procedure, potentially steering women down the unexpected path of termination, which they may not have considered previously.³⁸⁸

Concerning research findings by Minear *et al.* revealed that obstetricians and midwives, in the United Kingdom, admittedly anticipated dedicating significantly less time in pre-test counselling for NIPT, as opposed to the invasive methods of screening.³⁸⁹ While the study did not elaborate on the reasons for this, the findings suggested that this is due to the simple and familiar technique used to perform the test: a blood test that could be seamlessly integrated into the other blood tests parents have throughout pregnancy.³⁹⁰ Cernat *et al.* also found that HCPs believe the non-invasive, risk-free nature of NIPT, calls for less stringent requirements for informed consent.³⁹¹

As studies conducted in the UK revealed that HCPs viewed the consent process, for NIPT, to be less rigorous than it would for invasive testing,³⁹² suggestions have been made that a separate consent form should be provided before offering NIPT.³⁹³ Without a rigorous consent process, Davis worries that NIPT’s risk-free and familiar nature, will result to it becoming an opportunistic test.³⁹⁴ The beneficial perception of NIPT may correlate to the test being performed in the absence of informed consent, according to Davis.³⁹⁵

The attitudes and opinions of those parents, with lived experienced, is said to form a fundamental component to the evaluation of introducing NIPT into NHS trisomy screening

³⁸⁷ Adriana Kater-Kuipers and others, ‘*Rethinking counselling in prenatal screening: An ethical analysis of informed consent in the context of non-invasive prenatal testing (NIPT)*’, (2020) 34 *Bioethics*, 671.

³⁸⁸ *Ibid.*

³⁸⁹ Mollie A. Minear *et al.* (n130), 372.

³⁹⁰ *Ibid.*

³⁹¹ Cernat *et al.* (n367), 41.

³⁹² van den Heuvel A, Chitty L, Dormandy E, Newson A, Deans Z, Attwood S, Haynes S, Marteau TM, “*Will the introduction of noninvasive prenatal diagnostic testing erode informed choices? An experimental study of health care professionals*”, (2009) 78 *Patient Educ Couns*, 24.

³⁹³ Mollie A Minear *et al.* (n130), 391.

³⁹⁴ Dena S. Davis, ‘*Opportunistic Testing: The Death of Informed Consent*’, (2013) 23 *Health Matrix: Journal of Law-Medicine*, 50.

³⁹⁵ *Ibid.*

and testing pathways.³⁹⁶ As there is a significant lack of research exploring the views of those with lived experience, de Jong *et al.* stress that more empirical research is required to fully appreciate and understand the impact of NIPT on established consent regimes.³⁹⁷

Secondary or Unexpected Findings

Abnormal findings on the NIPT result have led to a *diagnosis* of maternal cancers, in some cases.³⁹⁸ As Lenaerts *et al.* explain, NIPT's design – to analyse cell-free fetal DNA (cffDNA) from the placenta – can also detect the presence of tumour-derived cfDNA.³⁹⁹ This is because the DNA from the tumour, or cancer cells, can be shed into the mother's bloodstream, which can be identified by NIPT technology.⁴⁰⁰ Maternal cancers were not initially considered due to its rarity, estimated to only occur in 1:1000 pregnant women.⁴⁰¹

Kater-Kuipers *et al.* question whether the 'risk' of incidental or unexpected findings, demand a restructure of the current process of obtaining consent for NIPT.⁴⁰² These scholars stress that "the next-generation sequencing technologies used for the test and its possible outcomes—trisomy 21, 13 and 18, and incidental findings—are increasingly complex ... there are concerns that women may lack understanding of relevant information about its aim, procedures, possible outcomes and consequences."⁴⁰³

Private Market

³⁹⁶ Dondorp, W.J., Page-Christiaens, G.C.M.L., de Wert, G.M.W.R. 'Genomic futures of prenatal screening: ethical reflection.' (2016) 89 *Clinical Genetics*, 538.

³⁹⁷ Antina de Jong and others, 'Non-invasive prenatal testing: ethical issues explored' (2010) 18 *European Journal of Human Genetics*, 277.

³⁹⁸ Marion Imbert-Bouteille and others, 'An incidental finding of maternal multiple myeloma by non-invasive prenatal testing', (2017) 37 *Prenat Diagn*, 1257-1260. These scholars explain that the reason behind abnormal NIPT results in the context of maternal cancers is that the tumour DNA contains duplications and deletions which create test failures or aneuploidies/monosomies.

³⁹⁹ Liesbeth Lenaerts and others, 'Noninvasive Prenatal Testing and Detection of Occult Maternal Malignancies', (2019) *Clinical Chemistry*, 1484.

⁴⁰⁰ Janel Case and Paige Hazelton, 'Genetic Counselors' Preparedness for Incidental Findings from Non-Invasive Prenatal Testing', (2018) 47 *Human Genetics Thesis Capstones*, 47.

⁴⁰¹ Marion Imbert-Bouteille and others, 'An incidental finding of maternal multiple myeloma by non-invasive prenatal testing', (2017) 37 *Prenat Diagn*, 1257.

⁴⁰² Adriana Kater-Kuipers and others, 'Rethinking counselling in prenatal screening: An ethical analysis of informed consent in the context of non-invasive prenatal testing (NIPT)', (2020) 34 *Bioethics*, 671. Meeting with Natera also revealed concern towards secondary findings and the impact it could have on the process of securing consent.

⁴⁰³ *Ibid.*

While research into the private market, particularly on NIPT, is scant across England and Wales, the Nuffield Council identified concerns surrounding the impact of its availability on NHS services for trisomy screening. While the use of NIPT on the NHS is to specifically target the common trisomies, private market providers of NIPT offer ‘whole panel screening’; this includes very rare conditions, such as microdeletions (i.e. DiGeorge Syndrome or 22q11 deletion) and sex chromosome aneuploidies (i.e. Turner’s Syndrome). Commentators question how parent consent, and reproductive autonomy, is preserved in these contexts, and the potential impact its availability may have on the provision of NHS services for supporting parent decision-making.

2.7 Constructing an Empirical Response to the Key Considerations Pertaining to Parent and Professional Interests on Consent for Trisomy Screening and Testing

This chapter has provided a comprehensive review of the key considerations and areas of interest identified from *Montgomery* and *Mordel*, with reference to existing clinical guidelines for England and Wales on informed consent and trisomy screening: provision of information on trisomy screening; understanding trisomy screening and testing; ‘choice’ and trisomy screening; communication and the HCP-parent relationship; ‘risk’ in the context of trisomy screening and testing; and ‘alternative’ methods of screening and testing.

This chapter has also referred to a significant body of existing research on consent for antenatal/trisomy screening and testing. The purpose of this exercise was to refine the themes identified from the case law – namely *Montgomery* and *Mordel* – but also locate any additional areas for consideration and further exploration from the current literature and research studies. A review of the clinical guidelines provided the researcher with an improved understanding of clinical expectation for obtaining consent for trisomy screening and/or testing, in conjunction with legal expectation.

This has also served as a means to distinguish, as far as possible, between the identified areas for consideration to construct a coherent socio-legal empirical response. Foregrounding an intended methodological approach, and research paradigm, would also be possible following a review of the terrain, accounting for the key area of law, clinical guidelines and medical literature in this regard.

Chapter 3 – Research Methodology and Methods

This chapter explains the research paradigm for this thesis. It explores the ontological, epistemological, methodological assumptions, and the methods that were evaluated and implemented, in the construction of this socio-legal empirical study. As the focus of this thesis rests on delineating parent and professional interests, to provide and secure informed consent for trisomy screening and testing, it is also necessary to contextualise methodological approach, by considering other socio-legal research into consent and reproduction. Consideration will also be given to methodological approach, with reference to other work on empirical bioethics and the law.

This chapter will outline the methodology and methods employed for each empirical study, exploring the interests of parents and professionals, in the context of decision-making and consent for trisomy screening, across the pathway. This chapter will also discuss the limitations of the research and the ethical considerations that were highlighted and subsequently managed.

3.1 Framing Socio-legal Empirical Research

It was decided that an exploration into parent and professional interests on consent for trisomy screening, would be better executed through a socio-legal empirical design, as opposed to the traditional doctrinal analysis of secondary sources. This was primarily due to the significant lack of contemporary research exploring the interplay between the practice of trisomy screening the law. It was also due to the researcher's desire to engage with the socio-legal matrix, in this context, keeping pace with the accelerating developments in trisomy screening. Engaging with an empirical exploration was also necessary to generate the desired impact, upholding the key objectives of the research study.

Among the medical and legal profession, there is a growing interest between the intersections of medicine and law, with the social sciences and humanities.¹ It raises intriguing questions between the applicability of the law to medical practice, but also where the law is located within the socio-cultural tapestry. The law is a product of social context and policy, and of

¹ Melanie L. Williams, *'Socio-Legal Studies and the Humanities – Law, Interdisciplinary and Integrity'*, (2014) 5 *International Journal of Law in Context*, 243.

public interest. It bonds a close relationship with normative development and cultural periods. However, this is not a view shared by all scholars, who stress that the law's attempt to appropriate the status of a science is unfounded, arguing that it is a "knowledge discipline ... isolated from social reality ... which holds nothing to contribute, epistemologically speaking, to our knowledge of the world as an empirical phenomenon".² This is a remarkable perception of the law, considering the abundance of theoretical evidence that exists, in case law and scholarly literature, demonstrating that the law is a social construct. As Williams insists, the "law is not merely a forensic exercise, but shares texts, languages and innate values drawn from the humanities ... it springs from the narratives and rhetorics of its 'subjects'".³

Empirical research in law is beginning to take centre-stage over other types of legal research.⁴ The traditional, purely doctrinal, analysis method of legal research has been criticised for its inward-looking and inflexible approach to understanding the law.⁵ It is highlighted by McConville and Chui that, as doctrinal research refers primarily to case law, the scope of the research, in terms of advancing our understanding the law and applying the law in practice, is narrower than non-doctrinal studies.⁶ However, this is a rather unnuanced and narrow perception. Undoubtedly, doctrinal research remains foundational in law. Without the requisite doctrinal basis, many empirical projects would lack a cogent theoretical underpinning and a nuanced understanding of the law. This would create a dissonance between the theoretical and practical implications of the law, undermining its impact and application to the socio-legal matrix.

A more accurate depiction of what empirical research in law offers, is that the discipline is awakening to the enrichment that well-crafted empirical research can bring. The emergence of empirical legal research – in addition to socio-legal research – has been recognised and encouraged by Universities, across Britain and the United States, for its potential to engage with broader socio-political contexts. It does so by procuring methods from other disciplines, such as the applied social sciences and humanities.⁷ This close relationship with empirical

² Geoffrey Samuel, *'Is Law Really a Social Science? A View from Comparative Law'*, (2013) 67 Cambridge Law Journal, 288.

³ Melanie Williams (n1), 259.

⁴ Mike McConville and Wing Hong Chui, *Research Methods for Law* (Edinburgh University Press 2007), 5.

⁵ Ibid.

⁶ Ibid, 6.

⁷ Ibid.

research in law and the social sciences, highlights that empirical studies are more effectively applied, and utilised, depending on the area of law. For example, empirical research could usefully contribute to human-focus areas, such as medical and tort law.

Empirical research in law facilitates the ability to combine a theoretical or conceptual framework – which doctrinal scholarship contributes to – with appropriate research methodology and methods, to produce the desired empirical evidence, to answer the research question or hypothesis.⁸ Empirical research in law is hailed as a means of advancing our understanding of law and the operation of legal systems.⁹ However, empirical research in law is not new, more newly in vogue. As early as the 1960s, socio-legal scholars recognised the importance of bridging the theoretical understanding of law with its application to practice.¹⁰ To achieve this, socio-legal research utilises methods which the applied social sciences are well accustomed, that is, qualitative and quantitative research.¹¹

3.1.1 Use of Socio-Legal Empirical Research into Matters of Public Health

Public health law research has been described as a “scientific study of the relation of law and legal practices to population health”.¹² To conclude whether a study is within the scope of public health law research, one must consider the purpose of the research.¹³ The purposes of public health law research is to “influence policy, improve the use of law as a public health tool, and better understand law as a social determinant of health”.¹⁴ It is different to other types of public health research, as it evaluates the effectiveness of the law as a means to implement or facilitate intervention.¹⁵ As Horton *et al* highlight, if a public health law study is to have any impact, it must assess public health practice in-line with requisite public health policy.¹⁶

⁸ Ibid.

⁹ Ibid, 47.

¹⁰ Ibid, 6.

¹¹ Ibid, 7.

¹² Scott Burris and Evan Anderson, ‘The Challenges of Quantitative Public Health Law Research’, (2010) 39 American Journal of Preventive Medicine, 99.

¹³ Heather Horton and others, ‘The Dimensions of Public Health Law Research’, (2002) 30 The Journal of Law, Medicine & Ethics, 197.

¹⁴ Ibid.

¹⁵ Scott Burris and others, ‘Making the Case for Laws That Improve Health: A Framework for Public Health Law Research’, (2010) 88 The Milbank Quarterly 169, 175.

¹⁶ Heather Horton, (n13), 198.

The law plays a vital role in the construction and development of public health regulations and policy. As highlighted by Wagenaar and Burris, “legal powers, duties and restraints structure the mission of public health agencies, and shape how it is carried out”.¹⁷ Wagenaar and Burris reinforce that the law, including legal research, is an effective tool to achieve necessary public health goals.¹⁸ While this is potentially true, it rather depends on the law in question. Empirical legal research, into public health, is necessary to produce the desired evidence to inform public health law and practice.¹⁹ This evidence is procured to justify amendments to policy, regulatory action and legal standards, consistent with fundamental human rights.²⁰ Thus, the experience of individual human beings must remain at the forefront of public health law research and the development of policy.

Horton *et al* correctly address the cultural gap between the health care practitioners, who produce and implement public health policy, and lawmakers, who produce and implement policies in law.²¹ A factual dissonance exists that research needs to identify and address. Historically, it is evident that the courts are continuously attempting to engage and keep pace with developments in medical culture and technology. The cultural gap between the courts and health care practitioners naturally generates differing perspectives, in regard to bettering public health policy and standards. A conflict between the interests of professional values, with law and policy, can produce this gap. Bridging the cultural gap between the law and healthcare profession, by establishing an open dialogue, is essential to the development of public health law.²² To homogenise this existing cultural dissonance must remain the key objective which drives empirical legal research into public health.

A research study of how the law impacts on population health through the “mediating structure of the health care system” fits within the definition of public health law research (PHLR).²³ However, an appreciation must be demonstrated towards the overlap between PHLR and health services research (HSR), “... the multidisciplinary field of scientific investigation that studies how social factors, financing systems, organisational structures and

¹⁷ Alexander C. Wagenaar and Scott Burris, *Public Health Law Research: Theory and Methods*, (1st edn Jossey-Bass 2013) 44.

¹⁸ *Ibid*, 45.

¹⁹ *Ibid*.

²⁰ *Ibid*.

²¹ Heather Horton (n13) 198.

²² *Ibid*.

²³ Alexander C. Wagenaar and Scott Burris (n17) 52.

processes, health technologies, and personal behaviours affect access to health care, the quality and cost of health care, and ultimately our health and well-being”.²⁴ Public health systems and services research (PHSSR) is another research paradigm which shares similar features to HSR, which focuses on the assessment of, “... organisations, financing, and delivery of public health services within communities and the impact of those services on public health”.²⁵ While there is a significant overlap between the two, the research should be designed to distinguish between PHSSR or HSR and PHLR, as PHLR focuses explicitly on the connection, and impact of law, on public or population health.²⁶ However, the overlap between PHSSR or HSR and PHLR is important for understanding how the law shapes the construction of health care systems, and delivery of public health services.²⁷ Therefore, a research study evaluating parent and professional interests, in the context of providing and securing consent for trisomy screening (under Public Health England and Wales), would fall into the framework of PHLR, and socio-legal empirical research. Consequently, the empirical research paradigm should correspond to this conceptual framework.

3.1.2 Typology: Socio-Legal Empirical Research in Health Care

Broadly speaking, legal research can be categorised into three groups: problem, policy and law reform research.²⁸ However, these tend to be interlinked aspects of applied legal research questions, rather than discrete categories. For a socio-legal empirical study into public health, these are all integral elements of the research inquiry, and are all interconnected. To assess the potential problem, evaluate the requisite policy, and ultimately decide whether a possible need for law reform is warranted, requires an empirical approach that would be quantitative, qualitative or a combination of the two.²⁹

When embarking on problem, policy and law reform-based research, the socio-legal researcher will be required to account for the social contextual factors, the impact of existing law and policy, and its application to practice.³⁰ In many ways, law and policy operates to steer and direct the application of legal provisions. To conduct a study of this nature, the

²⁴ Ibid.

²⁵ Ibid.

²⁶ Scott Burris and others, (n15), 176.

²⁷ Ibid.

²⁸ Mike McConville and Wing Hong Chui (n4) 19.

²⁹ Ibid, 20.

³⁰ Ibid.

researcher will also have to evaluate the appropriate available methods of research, typically surveys and interviews, with the desired demographic.³¹ Socio-legal researchers are encouraged to conduct qualitative research, as it is desirable to reach inferences or conclusions, based on the empirical evidence obtained.³²

To appropriate a humanist perspective is crucial to this research. Understanding the real, lived, human side of how the creation and implementation of law and policy correlates to the experience of trisomy screening and testing. We are not just studying the law, we are also studying human experience.

Socio-Legal Research into Public Health

More specifically, public health law studies are subdivided from the broad categorisations of empirical socio-legal research into different types. Policymaking studies are used to explore the question of how broader policy decisions are impactful in health contexts.³³ It uncovers whether the law possess the potential to promote health.³⁴ Quantitative and qualitative methods are deemed to be appropriate for policymaking studies.³⁵

The primary focus of public health law researchers is not on differentiating between the methodology used, but rather on the socio-cultural spheres of the project, and the clear communication of the research findings to healthcare professionals, law and policy-makers, and the general public.³⁶ Failure to effectively communicate the findings of a public health law study to healthcare professionals, law and policy-makers, and the general public, could hinder the likelihood of influencing change to public health policy, or to receive support from the public.³⁷

³¹ Ibid.

³² Ibid.

³³ Scott Burris and others (n15) 179.

³⁴ Ibid.

³⁵ Ibid, 181.

³⁶ Heather Horton and others (n13) 198.

³⁷ Ibid.

3.2 Contextualising Empirical and Methodological Approach: Socio-legal Research on Informed Consent and Bioethics

3.2.1 Reproduction and the Law: Socio-legal Research on Informed Consent

Socio-legal research, into bioethics, is cited as a relatively new field of study.³⁸ However, socio-legal research into issues like informed consent, decision-making and patient autonomy, have been explored for decades, in the context of reproductive torts³⁹ and negligence.

Walker explains “as technology has advanced, the level of control that can be exercised over the reproductive process has increased ... resulting in a number of claims in tort relating to pregnancy and birth”.⁴⁰ A significant body of socio-legal research exists exploring ‘reproduction and the law’, particularly on decision-making and consent for genetic and genomic testing, prenatal/antenatal screening, preimplantation genetic testing/diagnosis (PGT/D), and abortion. Indeed, this has been a growing field of study over the last twenty-years.⁴¹

‘Prenatal screening’ is taking centre stage in socio-legal studies, principally due to the rapid expansion of reproductive and screening technology, such as NIPT.⁴² A significant body of research exists exploring the socio-legal implications of patient autonomy and decision-making for ‘prenatal’ screening and diagnosis.⁴³ Existing socio-legal research, on prenatal

³⁸ Richard Huxtable ‘Friends, Foes and Flatmates: On the Relationship between Law and (Empirical) Bioethics’, in Bobbie Farsides and Clare Williams ‘Lessons from Experience: Establishing and Running Interdisciplinary Mixed-Method Bioethics Research’ (Cambridge University Press, 2017), 89; and Jonathan Montgomery, ‘Bioethics as a governance practice’, (2016) 24 Health Care Analysis 3, 5; and Richard Huxtable and Jonathan Ives, ‘Mapping, framing, shaping: a framework for empirical bioethics research projects’, (2019) 20 BMC Medical Ethics 1, 2.

³⁹ This is a broad term used to categorise claims for wrongful birth, wrongful life, and wrongful conception.

⁴⁰ Samuel Walker, ‘Applying the actual/potential person distinction to reproductive torts’, (2014) 14 Medical Law International 3-21.

⁴¹ Loane Skene, ‘Patients’ Rights or Family Responsibilities? Two Approaches to Genetic Testing’, (1998) 6 Medical Law Review 1, 7.

⁴² Jeffrey I. Wale, ‘Don’t forget the legal framework: The public provision of non-invasive prenatal testing in England and Wales’, (2016) 15 Medical Law International 203, 208.

⁴³ Rosamund Scott, ‘Prenatal Screening, Autonomy and Reasons: The Relationship Between the Law of Abortion and Wrongful Birth’, (2003) 11 Medical Law Review 265; and Roger Brownsword and Jeff Wale, ‘The Development of Non-Invasive Prenatal Testing: Some Legal and Ethical Questions’ (2016) 8 Modern Law Review 20; and Emily Jackson, ‘Abortion, Autonomy and Prenatal Diagnosis’, (2000) 9 Social and Legal Studies 467; and Jeffrey I. Wale, ‘Don’t forget the legal framework: The public provision of non-invasive prenatal testing in England and Wales’, (2016) 15 Medical Law International 203; and Michael Fay, ‘Genomic Negligence: An Interest in Autonomy as the Basis for Novel Negligence Claims Generated by Genetic Technology’, (2012) 20 Medical Law Review 642; and Louiza Kalokairinou, Heidi C. Howard and Pascal Borry,

screening, is often approached very broadly, extending its scope to a number of conditions and screening programmes; however, focus typically falls to Down's Syndrome, as it is the most common/commonly detected chromosomal condition.⁴⁴

'Genetic' and 'genomic' testing is often conflated with 'prenatal screening' in the body of socio-legal research, typically being referred to interchangeably in the academic literature for this field. 'Genetic' or 'genomic' testing is a broad term used to refer to a field of medical practice, which seeks to identify changes in DNA, that could cause present or future health implications. Scholars, such as Skene and Fay, have explored the socio-legal implications and complexity of consenting to genetic testing, in the context of 'carrier status screening'.⁴⁵ These scholars focussed primarily on conditions such as cystic fibrosis, Huntington's disease, and negligence surrounding the communication of neonatal genetic information more broadly.⁴⁶ Their research focussed on the uncertainty of screening results and diagnosis, and the implications of genetic information for the autonomy of individual patients, and their families.⁴⁷

Relatedly, Brownsword and Wale have undertaken socio-legal, and bioethical, work on the impact genetic testing – including non-invasive prenatal testing (NIPT) – has on patients' decision-making and consent.⁴⁸ They question the implications of advancements in genetic testing technology, not only on established legal frameworks for consent, but also on social institutions and systems.⁴⁹ Brownsword and Wale examine genetic testing in the context of assessing patients' "right not to know", due to the future implications of possessing this genetic information, not only on the individual patient, but also on the wider family system.⁵⁰

"Current developments in the regulation of direct-to-consumer genetic testing in Europe", (2015) 15 Medical Law Review 18.

⁴⁴ Timothy Mark Krahn, "Regulating Preimplantation Genetic Diagnosis: The Case of Down's Syndrome", (2011) 19 Medical Law Review 157, at 157; see also, Sonia M Suter, "The tyranny of choice: reproductive selection in the future", (2018) 5 Journal of Law and the Biosciences 262.

⁴⁵ Skene (n41), at 7; see also, Michael Fay, "Negligence and the Communication of Neonatal Genetic Information to Parents", (2012) 20 Medical Law Review 604, at 605.

⁴⁶ Skene (n41); and Fay (n45).

⁴⁷ Skene (n41), at 8; see also, Roy Gilbar and Charles Foster, "Doctor's Liability to the Patient's Relatives in Genetic Medicine: ABC v St George's Healthcare NHS Trust (2015) EWHC 1394 (QB)", (2016) 24 Medical Law Review 112.

⁴⁸ Roger Brownsword and Jeff Wale, "Testing Times Ahead: Non-Invasive Prenatal Testing and the Kind of Community We Want to Be", (2018) 81 Modern Law Review 646.

⁴⁹ Ibid.

⁵⁰ Roger Brownsword and Jeff Wale, 'The Development of Non-Invasive Prenatal Testing: Some Legal and Ethical Questions' (2016) 8 Modern Law Review 20.

Socio-legal researchers are also beginning to focus on the practical considerations, and implications, of 'direct-to-consumer' genetic testing on the ability of patients to deliver informed consent.⁵¹ Significant concerns are beginning to emerge surrounding the accessibility of the test, the lack of patient understanding and awareness for the test's potential implications, and the lack of follow-up support, by HCPs, after receiving the result.⁵² This research reveals that further empirical exploration is required to truly understand the lived experience of those patients undergoing this method of testing, and the ethical-legal implications it has on their ability to make an informed choice, in this regard.

Discussion of reproductive choice and consent, on 'prenatal screening', also falls to a body of socio-legal work around preimplantation genetic testing/diagnosis (PGT/D). PGT/D is the practice of identifying genetic or chromosomal conditions (through genetic profiling) in an embryo, used in conjunction with in vitro fertilisation (IVF). Krahn and Scott explore PGT/D regarding its impact on reproductive autonomy and choice, particularly in relation to the processes in place for HCPs to effectively secure parent consent.⁵³

Relatedly, socio-legal research into informed consent and reproductive choice, on prenatal screening/diagnosis and genetic testing, commonly sparks conversation around abortion. 'Abortion law' is gaining significant interest, particularly in the context of patient autonomy and informed consent.⁵⁴ Academics such as Scott and Prialux examine the 'social life' of abortion law, particularly the relationship between human rights and abortion practices. Attention is also beginning to turn to the socio-legal significance of the Abortion Act 1967 on reproductive choice and consent, and its (potentially discriminatory) impact on particular communities.⁵⁵ Research on reproductive choice, in this context, also assess parent and 'fetal'

⁵¹ Sivan Tamir, "Direct-To-Consumer Genetic Testing: Ethical-Legal Perspectives and Practical Considerations", (2010) 18 *Medical Law Review* 213; see also, Louiza Kalokairinou, Heidi Howard and Pascal Borry, "Current developments in the regulation of direct-to-consumer genetic testing in Europe", (2015) 15 *Medical Law International* 18.

⁵² *Ibid.*

⁵³ Krahn (n44); and Rosmund Scott, "The Uncertain Scope of Reproductive Autonomy in Preimplantation Genetic Diagnosis and Selective Abortion", (2005) 13 *Medical Law Review* 291.

⁵⁴ Rosamund Scott, "Prenatal Screening, Autonomy and Reasons: The Relationship Between the Law of Abortion and Wrongful Birth", (2003) 11 *Medical Law Review* 265; see also Nicolette Prialux, "Rethinking Progenitive Conflict: Why Reproductive Autonomy Matters", (2008) 16 *Medical Law Review* 169.

⁵⁵ Zoe L Tongue, "Crowter v Secretary of State for Health and Social Care (2021) EWHC 2536: Discrimination, Disability, and Access to Abortion", (2022) 30 *Medical Law Review* 177, 180.

interests.⁵⁶ Indeed, socio-legal academic, Michael Thomson, suggests that empirical research could build on existing socio-legal research in this area, to further explore the impact of consent systems on abortion law, and its relationship with the standards set by the medical profession.⁵⁷

The current study will contribute to the existing body of socio-legal research exploring informed consent and reproduction, focussing, for the first time, on the impact of parents' and professionals' plural interests on securing consent for trisomy screening. The empirical approach, taken in this study, will build upon the body of socio-legal research on reproductive torts and negligence, as a means to construct an appropriate methodological approach, to best respond to the considerations raised in *Montgomery*, and specifically *Mordel*, on securing consent for trisomy screening.

3.2.2 Methodological Considerations for Socio-Legal Research and Empirical Bioethics

Conducting a response to the issues raised in *Montgomery* and *Mordel*, regarding stakeholders' interests on consent for trisomy screening, requires careful consideration and contextualisation of methodological approach, appropriate for empirical bioethics research.

The term 'bioethics' is notoriously difficult to locate, and subsequently define.⁵⁸ 'Bioethics research' extends to a broad range of research, which possess a "multitude of different aims and objectives".⁵⁹ Richard Huxtable explains that 'bioethics' encompasses four sub-disciplines: normative ethics, which analyse and critique normative theories; applied ethics, which engage with theoretical perspectives from a specific discipline or field; meta-ethics, which engages into a reflexive process on identified concept(s); and descriptive ethics, which evaluate moral beliefs and practices.⁶⁰ All of the identified sub-disciplines overlap, and often difficult to address in strict isolation, as bioethics engages with the 'real' world.⁶¹

⁵⁶ Pamela Laufer-Ukeles, "Reproductive choices and informed consent: fetal interests, women's identity, and relational autonomy", (2011) 37 *American Journal of Law & Medicine* 567.

⁵⁷ Michael Thomson, "Abortion Law and Professional Boundaries", (2013) 22 *Social and Legal Studies* 191.

⁵⁸ Jonathan Ives and Heather Draper, 'Appropriate Methodologies for Empirical Bioethics: It's All Relative', (2009) 23 *Bioethics* 249.

⁵⁹ *Ibid*, at 249.

⁶⁰ Richard Huxtable 'Friends, Foes and Flatmates: On the Relationship between Law and (Empirical) Bioethics', in Bobbie Farsides and Clare Williams 'Lessons from Experience: Establishing and Running Interdisciplinary Mixed-Method Bioethics Research' (Cambridge University Press, 2017), 84.

⁶¹ *Ibid*.

Empirical bioethics refers to a wide range of varying methodologies, which have differing and conflicting views on how best to respond to “the challenge of connective normative bioethical analysis”, regarding realities of lived moral experience.⁶² Understanding the interaction between the requisite disciplinary approach, with ‘lived experience’, is said to be at the heart of any empirical bioethical methodologies.⁶³

The term ‘lived experience’ relates to the calculated positioning of the researcher to better understand how an ethical ‘problem’ may affect lives, what the provenance of the issue is, and how resolutions could be constructed, which affect the stakeholders.⁶⁴ Ives and Draper explain that “the advantage of this (obtaining lived experience) is that it contextualises a problem and sensitises one to the needs and experiences of those most affected by it.”⁶⁵ Achieving this understanding, according to Ives and Draper, requires researchers to become familiar with the literature, enter the field themselves to engage with the research participants, and collect the data, which is subsequently analysed and tailored to the research aims.⁶⁶ This process of locating, and subsequently analysing, the empirical data is also important to establish how concepts and meaning are utilised at ‘ground level’, ensuring that researcher and participants, particularly in interdisciplinary research, are “talking the same language and using terminology that are commensurate with the usage of the stakeholders.”⁶⁷

A considerable amount of uncertainty exists in terms of the “range and substance” of such methodologies, as researchers often experience difficulty communicating or articulating the aims and content of the research.⁶⁸ Therefore, a given methodological approach must be framed subject to the aims and objectives of the research, rather than attempting to locate and conform to existing empirical bioethics frameworks.⁶⁹

A four-part typology was outlined by Davies *et al.* to assist researchers in framing a strategic response, and appropriate methodology, to the aims and objectives of the empirical bioethics research: (i) use empirical data to describe attitudes toward an issue; (ii) use empirical data

⁶² Rachel Davies, Jonathan Ives and Michael Dunn, ‘A systematic review of empirical bioethics methodologies’, (2015) 16 BMC Medical Ethics 1, at 1.

⁶³ *Ibid*, at 2.

⁶⁴ Ives and Draper (n58), 251.

⁶⁵ *Ibid*, at 251.

⁶⁶ *Ibid*, 252.

⁶⁷ *Ibid*, 252.

⁶⁸ Davies *et al.* (n62), at 7.

⁶⁹ Ives and Draper (n58), at 249.

to explore the likely or actual consequences of bioethical policies and decisions; (iii) use empirical data to explore the ‘implicit normativity’ in scientific/clinical practice, and (iv) use empirical data to understand the institution of bioethics.⁷⁰ While this framework is useful to contextualise empirical methodological approach, this thesis also requires the researcher to locate socio-legal perspectives and influences on bioethical research.

Empirical research in law, and socio-legal studies more generally, are relatively new, particularly in the context of bioethics; therefore, it lacks an agreed meaning or scope.⁷¹ It may be, then, that empirical bioethics research could meaningfully contribute to the development of empirical legal research.⁷² Indeed, it is argued that the laws governing bio-practices are “indebted” to bioethics, as “bioethics helped to conceptualise problems, elucidate essential values, and influence the development of legal doctrines and processes”.⁷³ Huxtable explains that a meta-ethical bioethics paradigm can assist to clarify common legal concepts, such as the “reasonableness” and the “reasonable man”, and that normative/applied work would be best fit for exploring ethical principle, such as respect for autonomy and the value of human life.⁷⁴

A socio-legal researcher, investigating issues relating to empirical bioethics, should also be aware of the differing styles and conventions bioethics, and the law, ostensibly adopt. The law will seek to identify a winner: the focus of case law falls to the ‘winning’ argument, ‘winning’ the key points relating to a given legal issue, or simply being right or wrong.⁷⁵ Bioethical issues, on the other hand, are not as ‘black and white’, and often possess a multitude of ‘grey areas’, due to competing interests or rights.

The law also seeks to set standards, “drawing lines between the permissible and impermissible”.⁷⁶ The law may, therefore, draw the line in the wrong place, in terms of what *must* be done, whereas bioethics seeks to deduce what *should* be done. This issue becomes particularly prominent when we consider that the law often struggles to keep pace with

⁷⁰ Davies *et al.* (n62), at 7.

⁷¹ Richard Huxtable ‘Friends, Foes and Flatmates: On the Relationship between Law and (Empirical) Bioethics’, in Bobbie Farsides and Clare Williams ‘Lessons from Experience: Establishing and Running Interdisciplinary Mixed-Method Bioethics Research’ (Cambridge University Press, 2017), 85.

⁷² *Ibid*, at 85.

⁷³ *Ibid*, at 85.

⁷⁴ *Ibid*, at 85.

⁷⁵ *Ibid*, at 85.

⁷⁶ *Ibid*, at 85-86.

advances in medical technology and science, meaning a disconnect can appear between disciplines and disciplinary approach.

Law's purpose and contribution to bioethics is to guide human behaviour. Thus, bioethics and the law are said to share a conceptual relationship: both seek to engage with practice, and to guide people in the 'real world'.⁷⁷ The law and bioethics are also concerned with the 'process', and ultimately its application to systems of practice.⁷⁸ Indeed, the law is inherently empirical in nature, and must have a footing in the 'real world'; this could point to the law's potential to act as a testing ground for bioethical practices. However, the law typically adopts a top-down or doctrine-led approach, while bioethics typically work from the bottom-up, meaning that methodological approach can often be difficult to frame or locate.⁷⁹ Nevertheless, Huxtable concludes that "whichever extreme is preferred, the law will have important insights to offer", and that a 'middle-ground' can also be achieved in this regard.⁸⁰

Before conducting interdisciplinary empirical research in bioethics, Huxtable and Ives suggest a tri-phased approach in the construction of the research framework and paradigm: mapping, framing and shaping.⁸¹ The aim of the 'mapping phase' is to conduct a comprehensive survey to landscape the "general terrain"; this will allow the researcher to gain a sense of any initial themes within the researched area, and to formulate initial research questions.⁸² The aim of the 'framing phase' is to explore the landscaped terrain in greater depth; this will allow the researcher to develop their understanding of key research questions and issues therein, and to gain a comprehensive understanding of the experience of relevant stakeholders.⁸³ The aim of the final 'shaping phase' is to shape, or indeed reshape, the terrain, by analysing the findings from the mapping and framing exercise; this allows the researcher to obtain an informed insight into the researched area, and to formulate recommendations for future development or improvement.⁸⁴

⁷⁷ Ibid, at 86.

⁷⁸ Ibid, at 86.

⁷⁹ Ibid, at 86-87.

⁸⁰ Ibid, at 87.

⁸¹ Richard Huxtable and Jonathan Ives, "Mapping, framing, shaping: a framework for empirical bioethics research projects", (2019) 20 BMC Medical Ethics 1, at 3.

⁸² Ibid, at 3.

⁸³ Ibid, at 3-4.

⁸⁴ Ibid, at 4.

This contextualisation of socio-legal empirical research, in bioethics, alerts the researcher to a multitude of methodological factors when scoping the requisite paradigm to explore parent and professional (stakeholder) interests, in the context of providing and securing informed consent for trisomy screening.

3.3 Scoping Research Design

Assimilating the ontological and epistemological position of the researcher is necessary to inform the methodology, and methods, employed for an empirical study. It is fundamental to understand these concepts, and how they apply to the research, to decide whether the research paradigm is appropriate to achieve the study's primary objectives.

Firstly, it is necessary to identify where the study is situated within the wider tapestry of existing research and theory. Chapters 1 and 2 of this study form the requisite doctrinal and conceptual basis. A traditional doctrinal approach was employed in these chapters, whereby a chronological analysis of case law, and relevant medico-legal literature on informed consent for trisomy screening, was performed. Following the analysis, evaluation and critique of the literature, this process shaped the researchers understanding of the key areas, which require further empirical exploration. Themes and key areas of interest emerged from these chapters, providing theoretical and conceptual underpinning to the study; this ultimately informed the general socio-legal, and bioethical, empirical research design.

3.3.1 Philosophical Underpinning: Ontology and Epistemology

Empirical socio-legal research exploring the standard of informed consent under the NHS trisomy screening and testing pathway, to the researcher's knowledge and those of the healthcare professionals, had never been done before. Thus, the researcher was required to carefully evaluate the philosophical underpinning of the study, to carefully consider where it fits within the research paradigm.

A paradigm is essentially the entire basis of the research. Research paradigms have been described as "sets of beliefs and practices, shared by communities of researchers, which regulate inquiry within disciplines ... characterised by ontological, epistemological and

methodological differences”.⁸⁵ Lincoln and Guba expressed the importance of having a firm understanding of ontology, epistemology, methodology and axiology, as each component is closely related and are integrated, to produce the desired research paradigm.⁸⁶

3.3.2 Ontology

Firstly, it is essential that the researcher considers the various ontological perspectives. Ontology is the study of being.⁸⁷ Ontology is another word for a researcher’s beliefs about what constitutes reality.⁸⁸ It raises questions around what is true, what is real, and what exists.⁸⁹ Research originates from different belief systems and perceptions of truth, relating to what each individual researcher thinks truth is. Our own understanding of truth will shape our reality, and therefore it is crucial to understand ontology, in the context of our research paradigm.⁹⁰ Philosophies on these realities can be divided in various ways. Two categories of ontology are realism and relativism.

Realism

Realists believe that there is only one truth, and that truth does not change. Realists take a position of positivism, and believe that objects have an independent existence removed from that of the knower.⁹¹ Understanding of this single truth can be explored using objective measurements, according to realists. Once the truth is discovered, it can then be generalised to other situations. This view of reality influences the design of the research, and every single aspect of the study itself.

This research will need to consider a realist perspective to inform the theoretical foundation and underpinning. It will uncover any lacunae in legal coverage of parents’ experience of decision-making and consent, across the trisomy screening and testing pathway. It will be employed to confirm existing theories and to reveal patterns of behaviour. It will explore,

⁸⁵ Suzanne Bunniss & Diane R Kelly, *Research Paradigms in Medical Education Research*, (Blackwell Publishing Limited 2010) 360.

⁸⁶ Charles Kivunja & Ahmed Bawa Kuyini, ‘Understanding and Applying Research Paradigms in Educational Contexts’, (2017) 6 *International Journal of Higher Education*, 27.

⁸⁷ *Ibid.*

⁸⁸ *Ibid.*

⁸⁹ *Ibid.*

⁹⁰ Suzanne Bunniss & Diane R Kelly, (n85) 366.

⁹¹ James Scotland, ‘*Exploring the Philosophical Underpinnings of Research: Relating Ontology and Epistemology to the Methodology and Methods of the Scientific, Interpretive, and Critical Research Paradigms*’, (2012) 5 *Canadian Centre of Science and Education* 9, 10.

objectively, the various stages of the trisomy screening and testing pathway, to identify areas of particular interest. The findings will be objectively measured which can then be generalised into the corresponding demographics. While this lacks the required insight and individual view of the participants reality, it provides a theoretical framework and underpinning to further explore responses, through interpretative and subjective means.

Relativism

Relativists have entirely the opposite belief system to realists. Relativists adopt a position of interpretivism, taking a subjective view of reality which differs between individual beings.⁹² Therefore, relativists believe in multiple dimensions of reality; that is, the idea of truth is shaped and moulded by its context. Relativists believe that truth does not exist without meaning. Since reality is created by human perception, the notion of truth evolves and changes, depending on individual experience and context. Therefore, if reality is context bound, it is not capable of being generalised, like realists believe. Truth, in this sense, is transferrable to other similar contexts.

In the context of this research, there will be multiple realities between the position of the HCPs, and parents. Thus, parent and professional interests, in the context of securing and delivering consent, will inevitably vary: one being the recipient of care, and the other delivering the service. Status, the balance of powers between HCPs and parents, and idiosyncratic morals, values and expectations, will shape their reality. Furthermore, multiple realities will also exist within the parent and HCP populations.

Individual experience of providing consent for trisomy screening and testing, along the pathway, will inevitably differ between parents: some may have had a relatively mundane experience, while others might have received an unexpected result, requiring them to make decisions they may not have previously considered. Their reality of the experience may also have been shaped in retrospect, after having a baby with DS, ES or PS. Lived experience of having a baby with DS, ES or PS will also play a significant role in shaping their reality.

Securing consent, for trisomy screening, also requires the input of professionals, from a range of specialist fields: midwifery, ultrasonography, fetal medicine and perinatal practitioners.

⁹² Ibid, 11.

Thus, it is also vital that the study obtains the perspectives, and varying realities, of these professionals, to effectively delineate their interests going to broader systemic considerations for securing parent consent.

While generalisations are difficult to achieve without the objective framework, to measure data (associated with a realist ontology), it provides an insight into the responses of participants, to better understand their view of reality. Desirably, incorporating *both* a realist and relativist ontology could provide the most effective means of constructing the research design.

Realism and relativism can be combined coherently under the current research paradigm. A realist perspective allows the researcher to objectively identify patterns and behaviours between the areas of initial interest pertaining to the key research questions, and between the parent/professional populations: provision of information; understanding; choice; communication between HCP-parent; understanding of 'risk'; and alternative treatment options. This will also allow the researcher to harness an objective contextual understanding of the socio-legal, and medical considerations, required for this thesis.

A relativist perspective would permit the researcher to explore a subjective view of 'reality', pertaining to the key socio-legal areas of interest, and of the key stakeholders, gathered from the objective realist perspective. This allows the researcher to gain an enhanced, subjective interpretation of the conceptual relationship between the key areas of interest, and between the identified populations, with an aim of harnessing an understanding of the interaction between parent and professional interests, for delivering and securing consent for trisomy screening.

3.3.3 Epistemology

Epistemology refers to the form and nature of knowledge.⁹³ It is, in essence, a term which relates to the relationship the researcher has with the researched, in the context of how knowledge is created, obtained and communicated.⁹⁴ Therefore, it specifically focuses the attention of the researcher to the question of *how* we discover, and obtain, the required

⁹³ Charles Kivunja & Ahmed Bawa Kuyini, '*Understanding and Applying Research Paradigms in Educational Contexts*', (2017) 6 International Journal of Higher Education, 27.

⁹⁴ Ibid.

knowledge.⁹⁵ A researcher's ontological belief will dictate its epistemological belief; what the researcher believes about the nature of reality will govern what relationship the researcher has with the researched.⁹⁶

Positivist

There are two fundamental beliefs relating to how researchers should gather knowledge. Some researchers believe that this should be performed objectively, so that the researcher does not influence the data that is being collected. This school of thought believes that, in order to discover what truth is, the researcher needs to remove themselves, as much as possible, from the research. This is called an etic approach to research. This positivist epistemology is an objectivist position.⁹⁷ The ontology which fits with an etic approach to research is realism.

Interpretivist

However, the opposite approach to etic, is an emic approach. An emic approach is used by researchers who adopt a subjective position to reality: interacting with people about what truth means to them is required under this approach. The influence of the researcher on what is being researched is acknowledged; this is sometimes avoided but can also be embraced. Interaction with participants is required to gain an in-depth knowledge of their truth. The ontology which relates to this approach to research is relativism. This interpretivist epistemology adopts a position of subjectivism.⁹⁸ The truth is created by contextual meaning, requiring an in-depth discussion, with participants, to discover these 'truths'.

With the researcher's preference for a combined realist and relativist ontology for this study, it would be appropriate to also consider both a positivist and interpretivist epistemology. Thus, it is necessary for the researcher to consider an appropriate methodology to reflect both the ontological and epistemological positions, for the purpose of this thesis.

Positivism and interpretivism can also be combined coherently. A positivist perspective allows the researcher to objectively gather data to identify the 'truth', in terms of quantifiable

⁹⁵ Ibid.

⁹⁶ Ibid.

⁹⁷ James Scotland, (n91) 10.

⁹⁸ Ibid, 11.

patterns and behaviours within the dataset, as a means to contextualise the key areas of consideration, pertaining to parent and professional interests. An interpretivist would allow the researcher to subsequently gather data, by interacting with the key stakeholders, with an aim of obtaining an in-depth understanding of ‘truth’ means to them, and what their ‘truth’ is. A combination of both perspectives would allow the researcher to identify significant patterns of behaviour from the dataset, and an appreciation of what the ‘truth’ of these behaviour(s) mean to the research participants. This combination would allow the researcher to construct an appropriate empirical response to the research questions, and identified key areas of consideration, for delineating parent and professional interests on informed consent and trisomy screening.

3.4 Methodology

‘Methodology’ is a term that relates to how knowledge is discovered, and analysed, systematically. This term is typically confused by the term ‘methods’, which has a different meaning. Methodology specifically relates to the philosophy of how knowledge is discovered.⁹⁹ Methods relates to the tools and techniques of gathering the knowledge. Methodology is also governed by the researcher’s ontological and epistemological beliefs.

A realist or positivist methodology is focused on explaining relationships, attempting to identify the influence of causes on outcomes.¹⁰⁰ Correlation, and experimentation, are utilised to deconstruct interactions, by “empirical testing, controlled variables, and random samples”.¹⁰¹ These positivist methods typically produce quantitative data, involving questionnaires/surveys and standardised testing.¹⁰² The analysis of quantitative data requires descriptive and inferential statistics, allowing for population generalisations from the inferential statistics.¹⁰³ A paradigm of this nature suggests that an objective approach to research creates a desired robustness to empirical refutation.¹⁰⁴

There are distinct weaknesses to a positivist paradigm, in the context of socio-legal research. For example, it is typically difficult to isolate or discover variables. It is also an onerous task to

⁹⁹ Charles Kivunja & Ahmed Bawa Kuyini, (n93) 28.

¹⁰⁰ James Scotland, (n91) 10.

¹⁰¹ Ibid.

¹⁰² Ibid.

¹⁰³ Ibid.

¹⁰⁴ Ibid, 11.

identify, and utilise, the correct statistical test to analyse the data, which are commonly misused or misinterpreted in wider academic research. Furthermore, a lack of explanation as to *why* the results may differ, when constructing generalisations, is also a significant weaknesses.¹⁰⁵ These limitations would need to be carefully addressed, in the creation of the research paradigm.

A relativist or interpretivist methodology focuses on the “interaction between consciousness and phenomena”.¹⁰⁶ Obtaining the truth in this paradigm relies upon an awareness and appreciation of differing constructions of meaning, between individual beings. Truth is essentially co-constructed.¹⁰⁷ James Scotland, in the context of an interpretivist paradigm, remarks that, “... knowledge has the trait of being culturally deprived and historically situated ... the interpretive paradigm does not question ideologies; it accepts them”.¹⁰⁸

Interpretive theory is commonly grounded, which means that theory is grounded in physical data, with analysis and development of theory occurring post data-collection.¹⁰⁹ In-depth semi-structured or unstructured interviews, focus groups, observation, or open ended questionnaires, would be required to gather qualitative data under this research paradigm, yielding an extensive appreciation and understanding of human behaviour. Patterns and themes should emerge, and would subsequently be analysed, from the data, to reach a generalised theory. The data will always be presented in the context that it was studied. Qualitative data is satisfactory when measured in correspondence to its richness, credibility, reliability, dependability and transferability.

Scotland argues, however, that interpretive research is weakened by its rejection of an objective foundational base to knowledge, impacting on the data’s validity.¹¹⁰ Interpretivist paradigms pose difficulties: reaching a consensus due to its subjectivity that requires triangulation to improve validity; transferability due to the absence of unification of knowledge, and its highly contextual nature; the inability to satisfactorily apply the data to policy and issues; confidentiality and anonymity of participants, which requires researchers

¹⁰⁵ Ibid.

¹⁰⁶ Ibid.

¹⁰⁷ Ibid.

¹⁰⁸ Ibid, 12.

¹⁰⁹ Charles Kivunja & Ahmed Bawa Kuyini, (n93) 33.

¹¹⁰ James Scotland (n91) 12.

to modulate their contextualisation, to protect participant identity; the limited control of the researcher over the effect of their own presence on the interpretation; and collection and presentation of data, particularly with vulnerable populations.¹¹¹ Ultimately, under an interpretative paradigm, it is the researcher who steers the direction of the research, harnessing control over how the questions are delivered, desired approach to analysis and interpretation of the data set, with an aim of enhancing the study's credibility and reliability.¹¹²

3.4.1 Impact of COVID on the Research Paradigm

The outbreak of the COVID pandemic posed unprecedented challenges for researchers, particularly on those conducting empirical research. Lockdowns – both nationally and locally – meant that observational studies, within the NHS, were not possible. In addition, HCPs, across all areas of the NHS, were working tirelessly throughout the pandemic, which meant that some of the planned methods were not able to be executed effectively, in this regard.

Initially, it was planned that Grounded Theory (GT) would be implemented into the research paradigm. GT is a structured qualitative method that seeks to generate a theory, that is 'grounded' in the data collected, and subsequently analysed, by the researcher.¹¹³ The coding of data, under a GT methodology, is typically inductive, from which the researcher is then able to construct themes; thus, the theory is developed from the dataset itself.¹¹⁴ As such, GT was deemed not be an appropriate method for the aims and objectives of this thesis, and therefore could not be effectively integrated into the research paradigm.

A pragmatic paradigm took precedent, following interruptions to the proposed research methods. A pragmatic worldview allows the researcher to adapt and modify the study's design, subject to changes in circumstances and contexts. It also facilitates flexibility, in terms of utilising appropriate research methods, as a means to explore the research hypothesis.¹¹⁵ Importantly, the researcher, under the position of a pragmatist, is not bound by a particular reality or philosophy, but can reflect on the varying positions to construct an appropriate

¹¹¹ Ibid, 12-13.

¹¹² Ibid.

¹¹³ Diane Walker and Florence Myrick, "Grounded theory: An exploration of process and procedure", (2006) 16 *Qualitative Health Research* 547.

¹¹⁴ Ibid, at 549.

¹¹⁵ Mike McConville and Wing Hong Chui (n4) 256

research paradigm.¹¹⁶ Pragmatic theory was, objectively, the most appropriate framework for this study, in light of the aims and objectives of this thesis, and the evolving, unprecedented impact of the COVID pandemic on research projects.

3.4.2 Components to the Pragmatic Paradigm

Empirical research will typically be either quantitative or qualitative in nature, or a combination of both. Commonly, quantitative research uses statistics and a large sample size, while qualitative research uses a smaller sample size, and follows with thematic analysis.¹¹⁷ Historically, however, healthcare research has benefitted from the use of qualitative and mixed method research.¹¹⁸ Thus, it is important to focus on the distinct advantages, and shortcomings, of both qualitative and quantitative approaches to methods.

Qualitative Component

McConville explains that an empirical (or non-doctrinal) study, which explores problems, policy and legal reform in a qualitative context, are usually divided into two types: descriptive and evaluative.¹¹⁹ Non-doctrinal research can also be a combination of the two, with descriptive and evaluative elements. However, McConville should also emphasise that socio-legal research often requires a doctrinal foundation to identify the problem to be addressed; therefore, empirically focused law research designs typically involve a mixed methods approach, with a strong doctrinal foundation, as an integral component.

Qualitative methods are fundamental to discovering and understanding the insightful perspectives of research participants.¹²⁰ This approach to methods attempts to uncover questions relating to 'why' and 'how'.¹²¹ Qualitative methods are particularly suitable for "exploring new topics and obtaining insightful data on complex issues".¹²² Qualitative methods are utilised to achieve three main objectives: to understand context, and how different contexts shape understanding of knowledge; how our understanding of phenomena

¹¹⁶ Ibid.

¹¹⁷ Ibid, 222.

¹¹⁸ Shoshanna Sofaer, 'Qualitative Research Methods', (2002) 14 International Journal for Quality and Healthcare, 330.

¹¹⁹ Mike McConville and Wing Hong Chui, (n4) 32.

¹²⁰ Edwin van Teijlingen and Karen Forrest, 'The Range of Qualitative Research Methods in Family Planning and Reproductive Health Care', (2004) 30 Journal of Family Planning Reproductive Health Care, 171, 173.

¹²¹ Ibid, 171.

¹²² Ibid.

has different meaning, depending on context; and the purpose behind why people behave in different ways, providing an insight into the operation of behaviours, relationships and social systems.¹²³

Quantitative Component

Empirical (or non-doctrinal) socio-legal research can also incorporate quantitative methods. This methodology can be employed to explore “complexities of law, legal actors and legal activities”, and is used as a tool to ameliorate a researcher’s ability to collect and analyse data “in a scientific and systematic manner”.¹²⁴ Quantitative research is utilised to confirm existing theories, and to further explain patterns of behaviour.¹²⁵ It can also reveal lacunae in legal coverage, of a given area. The findings are measured objectively, removing the existence of bias, and the researcher’s personal values, which may impact the results, enhancing the reliability and credibility of the process of examining the relationship between variables.¹²⁶

Qualitative and quantitative research is typically very distinct, with each belonging to differing paradigms.¹²⁷ ‘Quantitative methods in research’ broadly refers to, “... the adoption of the natural science experiment as the model of scientific research, its key features being quantitative measurement of the phenomena studied and systematic control of the theoretical variables influencing those phenomena”.¹²⁸ Quantitative studies commonly adopt a positivist epistemology, under an objectivist position, focusing on the cause-and-effect relationship. McConville and Chui neatly highlight that quantitative research is adopted to evaluate and verify the appropriateness of existing theories, primarily as a means to explain behaviour or phenomenon, but is not used to produce new theories or insights into understanding the behaviour or phenomenon.¹²⁹ Quantitative methods are typically used to produce and analyse statistics, objectively measuring the data, and relationships, between the relevant variables.

¹²³ Ibid.

¹²⁴ Mike McConville and Wing Hong Chui (n4), 47.

¹²⁵ Ibid, 48.

¹²⁶ Ibid, 49.

¹²⁷ Julia Brannen, ‘Mixed Methods: Qualitative and Quantitative Research’, (Routledge Publishers 2017) 192.

¹²⁸ Mike McConville and Wing Hong Chui, (n4) 48.

¹²⁹ Ibid.

Quantitative research designs can be explanatory and descriptive in nature.¹³⁰ Explanatory studies are commonly used to obtain initial insights, or to inform ideas for the research study. They identify variables which are linked to the socio-legal phenomenological issues. It is explained that this quantitative design is used as a “first phase” to the broader research study.¹³¹ Descriptive designs aim to describe the phenomenon in question.¹³² They raise questions relating to the ‘what’ and ‘how’ of a particular issue. These studies can be subdivided into cross-sectional or longitudinal designs.¹³³

Socio-legal researchers must identify the research questions, hypotheses and variables of their study. Failing to construct a solid theoretical or conceptual framework could impact the study’s ability to produce credible concepts, variables or hypotheses to examine. A thorough and extensive review of the literature is fundamental to informing the theoretical and conceptual framework of the study.¹³⁴

3.4.3 Selecting Appropriate Methods Following the Impact of COVID

From a Triangulation to Explanatory Mixed Methods Research Design

Pre-COVID, it was planned that the parent and HCP study would adopt a triangulation mixed-methods design, comprising of surveys, interviews and observational methods. However, due to the outbreak of COVID, local and national lockdowns meant that observational methods could not be executed. A priority, during the selection of appropriate methods, was ensuring the well-being and safety of the researcher and participants. Therefore, it was decided that the parent and HCP studies would be better executed as a two-phase explanatory mixed-methods design.

An explanatory mixed-method design typically begins with a first phase quantitative foundation, followed by second phase qualitative text-based data. A combination of statistical and text-based data is effective in producing rich and robust conclusions, compensating for their own individual weaknesses.¹³⁵ For the purposes of this thesis, quantitative data will be

¹³⁰ Ibid, 50.

¹³¹ Ibid.

¹³² Ibid.

¹³³ Ibid.

¹³⁴ Ibid.

¹³⁵ Alexander C. Wagenaar and Scott Burris, (n17) 591.

necessary to reveal patterns between identified populations; this provides the necessary initial empirical foundation, and scope, to the parent and HCP studies. It will also unveil any patterns of behaviour, which need subsequent in-depth qualitative exploration.

Using both qualitative and quantitative tools for collecting data requires the requisite qualitative and quantitative analysis.¹³⁶ Depending on the research questions, socio-legal researchers may employ a range of different techniques, and tools, to analyse the data set.¹³⁷ The researcher may then compare or integrate the data collected.¹³⁸ Therefore, the qualitative data can be integrated with the quantitative data to explore, or confirm, the statistical foundation.

3.4.4 Experience of Practitioners of Informed Consent and Trisomy Screening

Before designing the research study, the researcher thought it was crucial that he investigated the perspectives of practising healthcare professionals, to gain a better understanding of the trisomy screening and testing pathway, and to identify any contentious areas between the law and medical practice. The researcher found it necessary to contact practising healthcare professionals, who perform a role on the trisomy screening and testing pathway,, to obtain their views, experiences and opinions, before any attempt was made to design a research project which could inform, or recommend, policy changes.

After a number of meetings with health care professionals (sonographers, midwives, obstetricians and neo-natal consultants) and healthcare academics, in 2017-2018, it was apparent that trisomy screening was currently in a period of transition and unrest. ES and PS had recently been introduced to the traditional 'Down's Syndrome screening programme', and NIPT had also been introduced to the pathway, in Wales. It was forecast, following my meetings with healthcare professionals and an extensive review of the relevant literature, that this could potentially lead to significant legal and policy implications, in the context of consent.¹³⁹ With the implementation, and impending implementation, of NIPT into existing

¹³⁶ Ibid, 592.

¹³⁷ Ibid.

¹³⁸ Ibid, 592.

¹³⁹ Caroline Silcock and others, 'Will the Introduction of Non-invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?', (2015) 18 International Journal of Public Participation in Health Care and Health Policy 1658.

English and Welsh antenatal trisomy screening programmes, it was forewarned that this could further exacerbate existing concerns for delivering and securing parent consent.

The researcher contacted the NHS Clinical Board of Director and consultant fetal medicine practitioners, from England and Wales, to discuss the contentious areas of law and practice, particularly in anticipation of the implementation innovative screening technology, such as NIPT. From there, the researcher was put in touch with many NHS healthcare professionals, including midwives, sonographers, fetal medicine consultants, and a neonatal consultant, from across England and Wales, as a means to scope the study.

In the design and execution of the research, the researcher worked particularly closely with an internationally renowned Consultant in Fetal Medicine, Dr Bryan Beattie MD FRCOG, over a 3-year period. He works for both the NHS, and private sector, and is the owner of the only private antenatal clinic in Wales. He is also the secretary for the Royal College of Obstetrician and Gynaecologists (RCOG) executive committee. Dr Beattie has a wealth of medico-legal knowledge, in the context of antenatal screening, and therefore his expertise for the research was invaluable. The researcher also worked closely with Dr Katie Morris, lead consultant in fetal medicine at Birmingham Women's NHS Foundation Trust, and senior member of the British Maternal and Fetal Medicine Society (BMFMS).

The researcher also arranged meetings with senior members, and coordinators, of ASW and FASP, to discuss the study. The members informed the researcher of current regulatory and systemic issues, emerging from the current trisomy screening pathway, that may require further empirical investigation. These discussions provided an invaluable insight into the topical issues facing HCPs, in the context of securing consent for trisomy screening.

The researcher also delivered a lecture – alongside Dr Bryan Beattie and Dr Samantha Leonard – for Obstetric trainees in Wales. A question and answer, following the lecture, allowed the researcher to obtain feedback for the study, and to keep pace with the recent developments, in the context of trisomy screening.

3.5 Parent Study

As discussed in chapters 1 and 2 – with *Mordel* and *Montgomery's* principles of self-determination and autonomy at the forefront of practice – a main objective of this thesis was

to explore the interests of parents for decision-making and consent, in light of the key areas of interest. Indeed, *Mordel* provided a timely illustration that consent for trisomy screening is not a 'one-off' event, as there are many decisions expectant parents will need to make across the chosen pathway. Thus, it was imperative, to the design of this study, that both qualitative and quantitative techniques were employed, to reflect the parents experiences, and elicit data to effectively explore parents' interests, in this regard.

The researcher obtained advice to conduct research into the interests of parents across England and Wales from April 2018, as this is when *both* NHS Wales and England had implemented the UK NSC recommendation: first introduced trisomy screening and testing into existing antenatal screening programmes. This would ensure standardisation of trisomy screening policy, and to limit systemic discrepancies of parents' experience, in this regard.

3.5.1 Research Population and Sampling

Researchers must consider the best means of recruiting the desired population sample to reach credible, reliable and representative conclusions for the study. Obtaining the relevant sample group also enables the researcher to accurately generalise the findings of the study.¹⁴⁰ The first stage of sampling is to clearly define the target population.¹⁴¹ The second stage is to select the sampling frame, which means the list of persons or cases which form the researcher's population of specific interest.¹⁴² The third stage is to choose an appropriate sampling technique, which are divided into two main types: probability or random sampling and non-probability or non-random sampling.¹⁴³

Under a quantitative study design, researchers typically utilise probability sampling.¹⁴⁴ Probability sampling or random sampling includes: simple random; stratified random; cluster sampling; systematic sampling; and multi-stage sampling.¹⁴⁵ In essence, probability sampling simply means that every entity or persons, in the population, have an equal opportunity of being included in the sample group.¹⁴⁶ This particular method of sampling reduces researcher

¹⁴⁰ Mike McConville and Wing Hong Chui, (n4) 54.

¹⁴¹ Hamed Taherdoost, '*Sampling Methods in Research Methodology: How to Choose a Sampling Technique for Research*', (2016) 5 International Journal of Academic Research in Management (IJARM), 19.

¹⁴² *Ibid*, 20.

¹⁴³ *Ibid*.

¹⁴⁴ Mike McConville and Wing Hong Chui (n4) 55.

¹⁴⁵ Hamed Taherdoost (n141) 20.

¹⁴⁶ *Ibid*, 21.

bias, increasing the study's credibility and representativeness.¹⁴⁷ The most favoured method of sampling are simple random, systematic and stratified, due to its associated strengths for representativeness, and for its ability to effectively generalise the findings.¹⁴⁸ On the other hand, this method of sampling requires a substantial amount of time, resources and cost.¹⁴⁹

Non-probability, or non-random, sampling is typically used under a qualitative research design, and is often employed where resources are limited.¹⁵⁰ Non-probability sampling includes: quota sampling; snowball sampling; purposive or judgemental sampling; and convenience sampling.¹⁵¹ Commonly, the aim of researchers, when employing a non-probability sampling method, is not to reach statistical inference or ensure representativeness, but to focus on small samples. Researchers must also aim to justify why this method was appropriate, and indeed necessary, for the study's explanatory or descriptive nature.¹⁵²

The fourth stage is to determine the sample size.¹⁵³ An adequate sample size is required for optimising the ability to generalise findings, and to reduce sampling error and bias.¹⁵⁴ The size of the sample population should correspond to the researcher's aims, the complexity of the population, and the intended method, to calculate the statistics and the analysis of the data set.¹⁵⁵ Therefore, a larger sample size does not necessarily correspond to more credible and representative research. It is imperative that the sample size is concluded based on the objectives of the research.

The final stage is to assess the response rate of the study.¹⁵⁶ Response rate corresponds to the number of individuals who agreed to take part in the research.¹⁵⁷ It is important to understand why respondents decide not to take part in the study, as it could subject the research to sample bias.¹⁵⁸ To reduce the presence of sample bias, it is vital that the

¹⁴⁷ Ibid, 21.

¹⁴⁸ Mike McConville and Wing Hong Chui (n4) 6.

¹⁴⁹ Hamed Taherdoost, (n141) 21.

¹⁵⁰ Mike McConville and Wing Hong Chui, (n4) 22.

¹⁵¹ Hamed Taherdoost, (n141) 20.

¹⁵² Ibid, 22.

¹⁵³ Ibid, 23.

¹⁵⁴ Ibid.

¹⁵⁵ Ibid, 24.

¹⁵⁶ Ibid.

¹⁵⁷ Ibid.

¹⁵⁸ Ibid.

researcher clearly defines the desired sample, whilst effectively managing and adapting the research design to integrate an appropriate, perhaps substitute, sampling technique.¹⁵⁹

Parent Groups: Purposeful Sampling

The two key variables of the study were whether the parent had a test positive diagnosis for a baby with either DS, ES or PS, and those who had not (but could either be low or high-risk following screening), and the country in which they received the screening. It is important to obtain the perspectives of parents from all demographics and populations, for a representative, comparative and unbiased analysis of the parents experience of decision-making and consent, across the pathway. Therefore, it was necessary to exercise *purposeful* sampling to recruit the desired demographic.

Purposeful sampling of parents who did not have a baby with a trisomy was recruited through online maternity support groups. Purposeful sampling techniques were used to recruit those parents who had, or had a higher-risk result, of having a baby with DS, ES or PS, through the Down's Syndrome Association (DSA) and Support for Trisomy 13 and 18 (SOFT).¹⁶⁰ Both SOFT and the DSA were also imperative to recruiting parents who had undergone NIPT screening, either on the NHS and/or privately. Recruitment of parents who had NIPT was executed by both purposeful and snowball sampling techniques.

3.5.2 Data Collection

Pilot Study

The design of the research questions required the input of members of the NHS R&D team, comprising of sonographers, midwives, obstetricians, and fetal medicine consultants, as an initial scoping exercise. Following these informative meetings, the researcher produced a set of questions that sought to gather data to meet the objectives of the research. The finalised questions were also sent to the DSA and SOFT to ensure that the questions were clear, employed appropriate language and terminology, and was ethically satisfactory.

¹⁵⁹ Ibid.

¹⁶⁰ SOFT work alongside national screening programmes across England and Wales (ASW and FASP) and provide parents with further information on the tested conditions, in particular, T18 and T13. SOFT created a close-knit network of trained volunteers who are able to deliver support to families through the screening journey. They also provide support to those parents who have ended or lost a pregnancy, as their baby had ES or PS. SOFT also work with HCPs and professional researchers to provide information, training and support.

The researcher conducted fifteen pilot studies with parents who had a positive diagnosis of having a baby with either DS, ES or PS, and others who had not (but received either a high or low-chance result), to see whether the questions were clear, accurate, balanced and reflective of their experience. From these pilot studies, in-depth feedback was gathered regarding the phrasing of the questions, and whether the questions reflected their experience(s). Consequently, the researcher was able to finalise the survey and interview questions to the highest possible standard, ensuring the reliability, credibility and validity of the data.

Surveys

Surveys and questionnaires are commonly employed as the appropriate method to collecting initial quantitative data for socio-legal studies. They are used to “understand people’s attitudes, beliefs, views and opinions on different aspects of social life”.¹⁶¹ Considering the lack of existing contemporary research on parents’ interests, in the context of consent and decision-making for trisomy screening, adopting surveys would provide the desired broad foundational (statistical) data, and allow the researcher to obtain patterns of behaviour between the study’s variables. This would produce the desired quantitative data, which could then be analysed using appropriate statistical tests, to determine whether the results are statistically significant between the populations.

When employing surveys, as a method of data collection, socio-legal researchers must first decide what needs to be measured in correspondence to any variables within specific populations. A closed question survey must incorporate a range of set multiple-choice responses, such as yes or no, or a psychometric ‘attitude scale’ – otherwise known as the ‘Likert’ scale – of positive or negative responses (i.e strongly agree, agree, neutral, disagree, strongly disagree), as a means of capturing and grouping the data, which could subsequently be analysed using a variety of statistical tests.¹⁶²

There are significant challenges, however, to conducting quantitative socio-legal research into public health.¹⁶³ It is challenging for socio-legal researchers to arrive at concluding

¹⁶¹ Mike McConville and Wing Hong Chui, (n4), 59.

¹⁶² Ibid, at pp.60.

¹⁶³ Scott Burris and Evan Anderson, (n15), 101.

explanations or evaluations of legal doctrines from statistics or numbers alone.¹⁶⁴ However, Burris and Anderson explain that quantitative studies in law are fundamental to assessing the effect of the law on population health, and could be “useful ... in facilitating clearer definitions and presentation of the characteristics of particular laws”.¹⁶⁵ The findings would then be objectively measured which can then be generalised into the corresponding demographics.

There are two types of administering surveys: self-administered and interviewer-administered.¹⁶⁶ Self-administered surveys are typically postal surveys, delivery and collection and online surveys.¹⁶⁷ Interviewer-administered surveys are typically face-to-face interviews and telephone surveys, and are far more structured in nature.¹⁶⁸ The researcher decided to perform interviewer-administered surveys. As participants were potentially vulnerable, building rapport with them was a key objective by creating an environment of trust and confidence. Thus, due to the sensitive and potentially demanding nature of the questions, interviewer-administered surveys were the best method to use. Furthermore, interviewer-administered surveys provided the researcher with the opportunity to clarify questions, if participants misinterpreted its meaning, and also to offer any additional communication support.

Table below demonstrates the original sample plan for the **surveys**:

	Wales	England
T21 (most common tested condition - 0.1% with 750 incidences in England and Wales each year)¹⁶⁹.	15 (CARIS = 78 a year)	30
T18 (second most common tested condition – 0.067%	4 (CARIS = 21 a year)	8

¹⁶⁴ Ibid.

¹⁶⁵ Ibid, 101.

¹⁶⁶ Mike McConville and Wing Hong Chui, (n4), 59.

¹⁶⁷ Ibid.

¹⁶⁸ Ibid, 60.

¹⁶⁹ Figure obtained from the Down’s Syndrome Association website. Whilst this is the recorded figure, the number of unrecorded incidences of Down’s Syndrome will be higher as terminations or miscarriages are not always recorded.

with 530-540 incidences in England and Wales each year) ¹⁷⁰		
T13 (Third most common tested condition – 0.025% with 230-240 incidences in England and Wales each year) ¹⁷¹	1 (CARIS = 8 a year)	2
No Positive Diagnosis (but received either a high or low-chance screening result)	30	60
NIPT (with any of the above demographics)	10	20
Overall	50	100

Table representing sample characteristics following **survey** responses:

	Wales	England
T21	8	16
T18	3	5
T13	2	4
No Positive Diagnosis	41	30
NIPT (with any of the above population)	14	12

¹⁷⁰ Primary figure obtained from the Nuffield Council Report on NIPT in 2017. However, it is not possible to guarantee that this figure accounts for every miscarriage or termination of a baby with Edwards' Syndrome.

¹⁷¹ Ibid.

Overall	54	55
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Interviews

Following the survey, parents were given the opportunity to participate in the interview. Interviews are often described as the bedrock of qualitative research. However, before conducting interviews, there are several key considerations which must be addressed by the researcher. Firstly, an appropriate sample group must be identified: focus must be on locating and selecting the desired demographic/sample group the researcher trying to reach, and why this sample is required.¹⁷² Secondly, the nature of the interview questions must be considered: focus should be on what questions will be asked, and why.¹⁷³ Thirdly, the researcher must take steps to learn how to phrase the interview questions, in conjunction with the context: focus must be on the appropriate use of language (removing the existence of jargon, offence and bias), particularly with vulnerable populations.¹⁷⁴

The main advantages of using interviews as a method of data collection are: they are very effective in terms of producing data which is rich in both quality and quantity; their relative flexibility and ability to tailor the questions to the responses and situation of each participant; they are useful where the research topic is new or if there is a lack of information due to the sensitive nature of the topic (exploring experiences, attitudes and behaviour); and are helpful to overcome communication or literacy barriers, which would otherwise impede on the collection of data using other methods.¹⁷⁵

The main disadvantage to interviews, as a method of data collection, is the sample size is typically much smaller than quantitative methods. It is also difficult to generalise the data findings to different populations and contexts, and interviewer bias can impact on the findings, which is either very difficult or impossible to eliminate (i.e the interviewer's physical appearance, age, ethnicity, religion and gender).

Semi-Structured Interviews

¹⁷² Edwin van Teijlingen and Karen Forrest, (n120), 172

¹⁷³ Ibid.

¹⁷⁴ Ibid.

¹⁷⁵ Ibid, at 172.

Interviews are divided into three main types: structured, unstructured and semi-structured.¹⁷⁶ The researcher decided that semi-structured interviews were the most appropriate means of collecting qualitative data for the parent study. Semi-structured interviews typically comprise of a combination of set open-ended questions, and flexible framework/interviewing schedule, to facilitate the desired discussion of relevant topics.¹⁷⁷

The researcher wanted interviewees to be able to provide a rich, nuanced and detailed account of their experience of decision-making and consent for trisomy screening, while retaining a degree of control over the structure of the interviewing process.¹⁷⁸ The flexible framework and compromising nature of semi-structured interviews allowed the researcher to elicit in-depth and insightful responses from participants.¹⁷⁹ Parents were able to divulge and convey particular areas of interest and concern. A degree of subjectivity was also utilised to further explore the initial patterns from quantitative findings among the populations. Indeed, semi-structured interviews operated as a “conversation with a purpose”.¹⁸⁰

However, utilising semi-structured interviews, as a method of data collection, relied on the truthfulness of the respondents’ accounts.¹⁸¹ The researcher had to manage and minimise the risk of responses that conflated or exaggerated accounts.¹⁸² Indeed, participants may have felt the need to alter or modify their responses to induce political, cultural or legal reform if they felt disadvantaged, frustrated or vulnerable. In addition to this, if the ethical considerations had not been sufficiently evaluated by the researcher, a risk existed of participants altering or concealing responses, if they felt that a breach of their confidentiality or anonymity may occur.

The researcher also had to be aware of ‘interviewer bias’ that may have potentially steered the discussion in a particular direction, impeding on the reliability and credibility of the

¹⁷⁶ Structured are often used in quasi-quantitative studies, due to their rigid and uncompromising nature; they do not permit the participant to elaborate on their answers. Unstructured are used to explore a limited number of research questions; often no more than five questions are introduced during the interview and are led by the participant.

¹⁷⁷ Edwin van Teijlingen and Karen Forrest, (n120), 172.

¹⁷⁸ Hamza Alshenqeeti, ‘*Interviewing as a Data Collection Method: A Critical Review*’, (2014) 3 English Linguistics Research, 40.

¹⁷⁹ Sarantakos, S, *Social research*, (Palgrave Macmillan 2012) at 276-290.

¹⁸⁰ Edwin van Teijlingen and Karen Forrest, (n120) 173.

¹⁸¹ Hamza Alshenqeeti, (n141) 45.

¹⁸² Ibid.

data.¹⁸³ Indeed, participants may have provided answers to the questions which they believe the researcher would want to hear. Therefore, drafting the research questions required a significant amount of input from HCPs and parents to ensure that these potential hinderances, to data collection, could be minimised.

Fifty-one participants decided to take part in the semi-structured interview. The interviews finished after fifty-one for two reasons. Firstly, the data became saturated; this means that the same themes kept reoccurring, and the researcher was not uncovering any new data. Secondly, the researcher successfully recruited an appropriate number of participants that were able to represent each demographic; this exceeded the original sample plan.

Table representing sample characteristics for **interviewing**:

	Wales	England
T21	5	4
T18	2	3
T13	1	3
No Positive Diagnosis	21	12
NIPT	10	8
Overall	29	22

Of the fifty-one who decided to take part in the interview, twenty interviews were conducted face-to-face, eight took place on Skype and twenty-three were conducted over the phone. This triangulation method was a useful tool to better understand the method which resulted to the most successful interviews; success, in this sense, was measured by the quality of the discussion and the richness of data collected.

Face-to-Face Interviews

¹⁸³ Edwin van Teijlingen and Karen Forrest, (n120), 171.

Pre-COVID, all interviews took place face-to-face. Face-to-face method of interviewing provided an effective means for collecting rich, detailed and high quality qualitative data.¹⁸⁴ The researcher was able to read body language and facial expression and provided the researcher with the ability to appreciate emotion and nuances, such as pauses and silences in conversation.¹⁸⁵ Furthermore, this method enabled the researcher to build rapport and trust with the participant, creating a safe and comfortable environment for participants to 'open up', in terms of sharing personal feelings and emotions.¹⁸⁶ Indeed, utilising face-to-face semi-structured interviews, whilst conducting research with vulnerable participants, provided a means to encourage an in-depth conversation, which explored personal and individual experiences.¹⁸⁷

On the other hand, a weakness of face-to-face semi-structured interviews was that it relied heavily on the skills of the interviewer. First impressions and gaining the trust of participants required skill and understanding, as vulnerable participants typically possess low levels of self-esteem and confidence.¹⁸⁸ In addition, a significant weakness of face-to-face semi-structured interviews was the impact of the interviewer.¹⁸⁹ Unwanted interviewer effect may also have influenced discussion: facial expressions and physical gestures of the interviewer to the responses of participants could influence the data. A negative consequence of this is the existence of bias, as participants may innocently provide responses based on what they think the interviewer wanted to hear.

Telephone and Skype Interviews

Following the outbreak of COVID, the interviews were conducted over the phone or via Skype. Historically, telephone interviews have been employed as a method of collecting quantitative data, with very few qualitative studies utilising this method.¹⁹⁰ In the context of telephone interviews, they were depicted as the "less attractive alternative to face-to-face

¹⁸⁴ Gina Novick, 'Is There a Bias Against Telephone Interviews in Qualitative Research?', (2008) 31 *Research in Nursing & Health*, 391.

¹⁸⁵ Sarantakos, S, (n179), 290.

¹⁸⁶ Ibid.

¹⁸⁷ Alan Bryman, *Social Research Methods*, (5th edn, OUP 2015) 301.

¹⁸⁸ Jacqueline H Watts, 'Emotion, Empathy and Exit: Reflections on doing Ethnographic Qualitative Research on Sensitive Topics', (2008) 3 *Medical Sociology Online*, 14.

¹⁸⁹ David Silverman, *Qualitative Research*, (Sage Publications 2016) 240.

¹⁹⁰ Gina Novick (n184) 391.

interviews”.¹⁹¹ There exists a bias against the use of telephone interviews in qualitative research.

The initial objective of conducting telephone interviews was to overcome geographical boundaries to increase access to participants, and to be more cost effective for both researcher and participant.¹⁹² The benefits to telephone interviews, following a review of the literature, was that participants can take part in the research in a comfortable, relaxed or familiar environment which could encourage “rich, vivid, detailed and high quality” discussion.¹⁹³ Other advantages include being able to facilitate confidentiality, privacy and anonymity of the participants if they so request, an increase in rapport, a decrease in social pressure and the ability to take notes without distracting participants when they are providing an answer.¹⁹⁴

The disadvantages of telephone interviews, according to the literature, is that researchers are unable to effectively collect nonverbal data¹⁹⁵, such as reading the body language or facial expressions of the participants. Researchers are not able to appreciate visual cues, which is reported to be a fundamental concern to telephone interviews, with researchers losing the ability to effectively collect informal communication or probe information based on nonverbal reactions.¹⁹⁶ Discussions over the phone were said to be typically shorter than face-to-face interviews, resulting in a data set which lacked sufficient depth or richness.¹⁹⁷ However, as Novick highlights, there is a scarcity of evidence which supports these claims, with research demonstrating that telephone interviews can last between 1-2 hours.¹⁹⁸ Furthermore, to overcome the commonly reported issue of researchers being unable to utilise visual aids with telephone interviews, the research information sheet and questions included diagrams to prompt their memory of the trisomy screening and testing pathway.

¹⁹¹ Ibid.

¹⁹² Ibid, 393.

¹⁹³ Ibid.

¹⁹⁴ Ibid.

¹⁹⁵ Ibid, 395.

¹⁹⁶ Ibid.

¹⁹⁷ Ibid.

¹⁹⁸ Ibid.

Skype is a relatively new tool for qualitative research, and its use sparks conflicting perspectives among academics and researchers, as to their usefulness and effectiveness, in this regard.¹⁹⁹

Similarly to telephone interviews, Skype was utilised as a tool to overcome geographical boundaries, and to overcome financial implications associated with travel for both researcher and participant. As with telephone interviews, participants are able to remain anonymous, if they so request, increasing trust and rapport with the participants. The literature also points to the relaxed and familiar environment Skype is able to facilitate, with participants being able to complete interviews from the comfort of their own homes/workspaces.²⁰⁰ Facilitating this type of environment is said to encourage rich and detailed data, as participants are willing to 'open-up' and trusting in this regard.²⁰¹ Researchers are also able to utilise the video function to incorporate visual aids and/or prompts, to maintain or enhance rich and detailed discussion.²⁰²

The principal disadvantage of Skype interviews is that this tool is ultimately dependant on whether the participant has access to the technology required.²⁰³ Another disadvantage, according to the literature, is that the researcher is not able to read body language, facial expressions or nonverbal cues, as effectively as face-to-face interviews.²⁰⁴ The literature also purports that maintaining rapport between researcher and participant is more difficult via Skype, as participants report feeling "distanced", with interviews typically being shorter than those face-to-face.²⁰⁵

The discussion often became disjointed during Skype interviews, as the technology was unpredictable and/or unreliable (either on the researcher's or participant's side), resulting to the loss of momentum, and trail of thought, between researcher and participant. Due to the unpredictability of the technology, this created anxiety from both participant and researcher,

¹⁹⁹ Lo Iacono, Valeria, Paul Symonds and David HK Brown, "Skype as a tool for qualitative research interviews", (2016) 21 Sociological Research Online 103, at 104.

²⁰⁰ Ibid, at 105.

²⁰¹ Ibid.

²⁰² Ibid, at 106.

²⁰³ Mirick Rebecca G and Stephanie P. Wladkowski, "Skype in Qualitative Interviews: Participant and Researcher Perspectives", (2019) 24 The Qualitative Report 3061.

²⁰⁴ Ibid, at 3062.

²⁰⁵ Ibid, at 3065.

which may have had a negative impact on the quality and richness of the qualitative data collected.

Of the three methods of conducting the interview discussions, telephone interviews elicited the most honest, rich and informative discussions. At the end of the interviews, the researcher asked all participants if there were aspects of the research questions that they would change. Almost all the participants were satisfied with the research questions and felt that they were able to convey their experiences fully.

3.6 HCP Study

Exploring the interests of HCPs, in the context of supporting parent decision-making and securing consent for trisomy screening, was a key objective in this thesis. Indeed, *Montgomery* underlined the need for HCPs to facilitate a shared decision-making model of care, to protect patients' right to self-determination and autonomy. More specifically, however, *Mordel* threw into question the possible disconnect and frailty of established systems for securing consent, for trisomy screening. *Mordel* also reassessed the individual role HCPs play (namely midwives and sonographers), when supporting parent decision-making and consent, along the trisomy pathway. Thus, it was imperative that the study explored, firstly, HCPs experience of securing parent consent under existing systems of consent – including the impact the decision in *Mordel* had on clinical practice – and, secondly, key considerations going to supporting parent decision-making across the pathway, outlined in *Montgomery* and *Mordel*. The key areas of interests and themes, identified from the clinical guidelines on trisomy screening and testing, formed the framework for the exploration, in this regard.

3.6.1 Research Population and Sampling

Purposeful Sampling

To gain a better understanding of systemic considerations going to securing consent for trisomy screening, it was vital that HCPs – who performed a key role on the pathway – were recruited for this study. Sample group consisted of midwives, sonographers and consultants (obstetricians and fetal medicine).

The sample was primarily recruited by the British Maternal and Fetal Medicine Society (BMFMS), led by Dr Katie Morris; this proved to be effective for recruiting the desired population. The British Journal of Midwifery (BJM) effectively recruited midwives and sonographers for the study. The Society of Radiographers (SoR), UK Audit & Research Collaborative in Obstetrics & Gynaecology (UK ARCOG) and the International Society of Ultrasound in Obs & Gyn were effective for targeting ultrasonographers, led by Dr Mike Rimmer. The BMFMS were also instrumental for gathering a contextual understanding of the trisomy programmes, as their executive committee work closely with the UK National Screening Committee (UKNSC) and the Fetal Anomaly Screening Programme (FASP).

3.6.2 Data Collection

Pilot Study

This study was designed with the assistance of Dr Katie Morris and Dr Bryan Beattie. The research questions were drafted to best gather data on the key areas of interest for the aims and objectives of this thesis. The questions were piloted by the BMFMS which saw an uptake of ten midwives and eight sonographers. Feedback was received from these HCPs and amendments were made to enhance the quality of the research questions, producing data that was more credible and reliable.

Closed and Open-Ended Surveys

Due to the ongoing COVID outbreak, a two-stage closed and open-ended survey was used to gather quantitative and qualitative data. LimeSurvey programme was used to design and distribute the surveys.²⁰⁶ This method allowed HCPs to complete the questions around their workload, during the pandemic.

The closed survey consisted of ten questions. The survey sought to identify patterns in the data to enable the researcher to further explore these areas in greater depth using qualitative techniques.

Table representing sample characteristics following closed-survey responses:

²⁰⁶ This programme allows researchers to design and distribute survey questions. There are also options on the programme to quickly and efficiently analyse both qualitative and quantitative data through various coding systems. The programme produces tables and 'coding trees' for both qualitative and quantitative use.

	Wales	England
Sonographer	16	37
Midwife	23	49
Consultant	4	9
Overall	43	95

The open-survey consisted of ten questions. This survey sought to gather rich and meaningful qualitative data during the COVID pandemic. Ideally, the researcher would have focussed exclusively on an interviewing method to gather qualitative data. However, the researcher understood that the pandemic meant HCPs were not able to engage with lengthy semi-structured interviews, and completion of the open-ended survey could fit around the HCPs unprecedentedly busy schedules.

Table representing sample characteristics following open-ended survey responses:

	Wales	England
Sonographer	12	22
Midwife	15	31
Consultant	3	7
Overall	30	60

Zoom Semi-Structured Interviews

The option was also given to HCPs to participate in a semi-structured interview if they were able to, with the ongoing pandemic. The interview questions were identical to those on the open-survey. The purpose of the semi-structured interviews was to contribute further depth to the existing qualitative data, and to provide HCPs with the opportunity to engage into a dialogue with the researcher. Due to local and national lockdowns, the interviews with HCPs were conducted over Zoom. This allowed the researcher to access the sample group without

having to break COVID rules (gain access to hospital sites or leave the local area). It also transpired that this was an effective method of interviewing, as HCPs were able to participate at home or around shift patterns.

Table representing sample characteristics following interview responses:

	Wales	England
Sonographer	4	5
Midwife	6	8
Consultant	3	2
Overall	13	15

The diverse sample of midwives, sonographers and consultants allowed the researcher to employ quantitative and qualitative techniques, to explore data in search of delineating areas of particular concern, pertaining to the question of whether a ‘reasonable’ system to secure parent consent, is currently implemented between professionals and maternity units. It also allowed the researcher to identify potential challenges HCPs face, when supporting parent decision-making, along the trisomy pathway.

3.7 Private Sector Study: Market for Trisomy Screening and Non-Invasive Prenatal Testing (NIPT)

While not central to the key objectives of the thesis, the researcher found it necessary to conduct an exploration into the private NIPT and trisomy screening market, to determine whether this had any impact on the interests of NHS HCPs and parents, for delivering and securing consent, along the trisomy screening pathway. Despite the Nuffield Council raising early concerns for the provision of private care, namely for utilising NIPT technology for trisomy screening, research into decision-making and consent, in this context, was scant.

With the private market growing at an exponential rate, particularly in the context of NIPT, concerns emerged pertaining to its impact on NHS services. Concerns extended to the provision of parent support, going to decision-making and consent for trisomy screening, and the use of NIPT. The private market also raised questions surrounding its impact on

established NHS systems for securing parent consent, for trisomy screening, and whether HCPs should account for this consideration, particularly in the context of NIPT screening.

For this study, as very little is understood of the impact the private NIPT and trisomy screening market may have on existing NHS systems of care, a phenomenological methodology was deemed most appropriate, for the purposes of acquiring meaningful data. Phenomenological approaches typically employ qualitative techniques to gather data, such as observational methods, interviews, open-ended surveys or a combination of these.

3.7.1 Sampling

Purposeful Sampling

The sample was recruited at the 'Innermost Healthcare Clinic', in Cardiff, owned by Dr Bryan Beattie. The researcher identified the private NIPT market as an under-researched area from the literature, particularly on its impact on NHS systems for securing consent, for trisomy screening. Gaining access to parents, who engage with private NIPT screening, would provide the researcher with a better understanding of the intersection between the private sector and the NHS, for the purpose of this thesis.

3.7.2 Data Collection

Pilot Study

The researcher was granted permission, by the clinic manager, to attend the clinic for meetings, and to discuss the latest developments in NIPT technology. The purpose of this study was to: provide the researcher with an understanding of the private NIPT screening experience; why parents access NIPT on the private market; and the potential impact this may have NHS provisions of support for decision-making and consent.

This study also allowed the researcher to identify and understand the latest developments in NIPT technology, and the panel of conditions parents were able to screen for, under the private market: 'Natera' was the provider of NIPT for Innermost, offering the widest panel of conditions of all the pharmaceutical giants.

Both the researcher and Dr Beattie discussed the design of the study, before commencing the research. Dr Beattie thought the researcher would benefit from obtaining an understanding

of the clinical environment, as a foundation, providing access to the NIPT technology, equipment, and clinical rooms. Following this, the researcher designed five research questions, as a means to explore the researcher's experiential assumptions. The questions were reviewed by supervisors and Dr Beattie, before commencing the survey.

Interactions with HCPs at the Clinic

The researcher gained invaluable experience and knowledge from interacting with the HCPs at the clinic. With funding from Natera – provider of NIPT to the clinic – Dr Beattie, Dr Leonard and the researcher created an educational tool for NHS HCPs, specifically addressing the influence of the private NIPT market on the provision of NHS support for decision-making and consent, along the trisomy pathway. This experiential learning also provided the researcher with a contextual understanding, and insight, into the operation of private NIPT clinics: how parents were drawn to private care; what technology was available to parents; and where the developments in NIPT screening were heading in the future. The researcher attained an understanding of the potential impact and influence of the private market on NHS provisions, which support parents' decision-making and consent, for trisomy screening and testing. This experience also left the researcher with questions, pertaining to the influence of the private market on established NHS systems for securing consent.

Open-Ended Survey

Initially, it was deemed that interviewing methods would be appropriate for the purpose of this study. However, upon reflection during the outbreak of COVID, open-ended surveys were considered the most pragmatic and ethical means of collecting the desired qualitative data. Ensuring that the parents were comfortable was a key objective in this research. Allowing parents to complete the research questions, at their own leisure, provided the researcher with a rich and rounded data set.

Participants were given the option to participate in the qualitative online survey, via LimeSurvey. The uptake number of parents was small, with only five participating. While the study began before the COVID pandemic, it was significantly interrupted by the outbreak, with the number of parents accessing private care significantly decreasing, possibly due to financial or logistical challenges. While the sample size was not a significant number, particularly in the context of coding for in-depth qualitative analysis, it provided original and

meaningful data on the potential impact the private NIPT market could have on NHS services, and the provision of parent support going to decision-making and consent for trisomy screening.

3.8 Data Preparation and Analysis

3.8.1 Qualitative data

The researcher had to carefully consider the most effective and appropriate means of preparing and analysing the qualitative data, collected from the parent, HCP and private clinic studies, within the research paradigm.

All interviews, from the parent and HCP studies, were recorded and transcribed. During transcription, participants' details were stripped and given a 'research number' to ensure confidentiality and anonymity. Each participant was given the opportunity to review the transcriptions, to ensure they were satisfied with the data, upholding the highest ethical standards.

Interview length varied between thirty-minutes to two hours. Each interview took between one to three hours to transcribe. During the process of transcribing, the researcher became very familiar with the dataset, which was beneficial to gain an initial understanding of early themes, that may require further exploration using appropriate analytical techniques. The transcriptions were uploaded to Nvivo12 software to begin the process of coding the qualitative data.

The open-ended HCP survey responses were also uploaded to Nvivo12, in preparation for qualitative analysis. The responses were taken from LimeSurvey, and the data was separated from the interview data to be analysed using the Nvivo12 software. All responses were read, and re-read, to check for any information that could reveal the identity of the participant; any indicators of identity were removed before engaging with the process of coding.

As the researcher adopted a pragmatic worldview, Thematic Analysis (TA) was deemed to be the best means of effectively analysing and coding the dataset, from both the parent and HCP studies. Thematic analysis is not bound to a particular ontological and epistemological perspective, nor is it bound by a worldview; it is merely a means of analysing qualitative

data.²⁰⁷ Thematic analysis is the process of systematically and methodically identifying frequent patterns or occurrences, within the dataset.

Thematic analysis is performed in various stages.²⁰⁸ The researcher familiarises themselves with the data through the process of transcription; this process allows the research to identify initial themes and ideas.²⁰⁹ A systemic identification of initial codes is then conducted from the data collected. Defining codes, and arranging them into the requisite themes, forms the foundation to the framework. Patterns emerge from the dataset, which is often cross-checked to the codes and themes. The themes and codes are further refined to reflect the key areas of interest.

Themes and patterns were identified by the occurrence, or frequency, in which they emerged from the data. Nvivo12 allowed the researcher to construct a table of themes or 'nodes', and the frequency in which they occurred. While there was some overlap between themes, the researcher was able to successfully refine the data gathered, into their requisite themes, for in-depth analysis.

Thematic analysis has been criticised for lacking the rigour of other analytical methods, for effective qualitative analysis: the technique is subject to the discretion of the researcher's perception and interpretation of the dataset.²¹⁰ Nevertheless, the flexibility of this method has been consistently commended, by social researchers, for enabling researchers to produce and define clear themes that emerge from the requisite dataset, without adhering to predetermined and regimented confines of other analytical methods, such as Grounded Theory (GT) (see discussion of this under 'research paradigm').

Initially, a deductive coding approach was first used by the researcher to analyse the dataset. As highlighted in chapter 2, a narrative synthesis technique was undertaken by the researcher on the relevant clinical guidelines and literature, producing broader deductive themes for further exploration. This meant that the researcher had designed and defined broader initial themes from the literature, before exploring the dataset itself. However, coding and analysis, following the empirical gathering of data, was primarily inductive, meaning that the

²⁰⁷ Alan Bryman (n187) 370.

²⁰⁸ Ibid.

²⁰⁹ Ibid.

²¹⁰ Ibid, 375.

researcher methodically identified patterns and themes, as they emerged from the dataset. A combined deductive and inductive approach is often cited as facilitating the desired rigour, for the purpose of qualitative thematic coding.

The researcher also considered Interpretative Phenomenological Analysis (IPA) as a means to analyse the dataset. IPA follows a very similar process to Thematic Analysis: familiarisation of the data through transcription; initial identification and labelling of themes; systemic grouping of major and minor themes; production and refinement of themes to reflect key areas of interest.²¹¹ However, the difference between the two methods of analysis, is that IPA focusses on capturing idiosyncratic experiences, as opposed to identifying patterns in the dataset. IPA is employed with very small sample sizes, often no more than ten participants.²¹²

However, while it was decided that Thematic Analysis would be an appropriate and effective means of analysing the larger parent and HCP dataset, over that of IPA, IPA was an appropriate tool for analysing the private study qualitative data. A combination of the open-ended surveys and small sample size meant that the researcher had an opportunity to explore the experiences of individual cases, in greater detail. This proved to be very effective for capturing idiosyncratic experiences and key considerations pertaining to decision-making and consent, under the private market.

3.8.2 Quantitative Data

All survey responses, from the parent and HCP studies, were collected and uploaded to SPSS (Statistical Package for the Social Sciences) and Excel, in preparation for quantitative analysis. While SPSS is a reputable and reliable software, statistical coding – using specific equations and formulas – was performed by the researcher, on Excel, to ensure internal validity when analysing large datasets; comparisons were then drawn between the data, uploaded to SPSS and Excel, to ensure consistency in this regard.

When analysing the quantitative data, researchers must evaluate the appropriateness statistical tests, to effectively explore the research questions. The quantitative data can be subdivided into three main categories; that is, univariate descriptive, bivariate descriptive and

²¹¹ Ibid, 380.

²¹² Ibid.

explanatory analysis.²¹³ These are employed to assess the characteristics of a particular phenomenon, and to explore, explain and understand a range of relationships between the study's variables.²¹⁴

Univariate descriptive analysis merely provides a picture of the data at that moment in time, generating a superficial and basic conclusion of each variable.²¹⁵ Bivariate analysis requires the researcher to assess the study's variables together, meaning that statistical tests are performed to measure whether a significant difference, or an association, exists between the relevant variables.²¹⁶ Statistical testing is also used to establish relationships between the variables. Explanatory analysis requires the researcher to broaden the interpretation of the data, beyond establishing significance and relationships between the variables, to answering why these outcomes have been produced.²¹⁷

The researcher felt that bivariate analysis would be the most appropriate means of analysing the quantitative data, for both parent and HCP studies. A bivariate analysis would allow the researcher to identify and locate any significant differences (or similarities) between the responses of the individual parent/HCP groups, with the aim of measuring any interactions, or relationships, which relate to the key themes and areas of interest, for this thesis. This bivariate procedure of analysis would best contextualise the patterns of behaviour between the individual HCP and parent groups, leading to a more robust and comprehensive understanding of how to approach the subsequent qualitative exploration, of the quantitative dataset.

The first phase of data analysis for quantitative data requires the researcher to consider whether the dataset is normally distributed. A Shapiro-Wilks test was performed to determine whether the data set was normally distributed. It is recommended that the data is plotted in the form of a histogram, and deciding whether it generates a bell-curve distribution. A Shapiro-Wilks test of normality was performed for *every* question from the surveys, under both sets of data, to determine whether the data is normally distributed. When determining whether the data is normally distributed, focus rests on the 'p value' produced from the test.

²¹³ Mike McConville (n4), 61.

²¹⁴ Ibid.

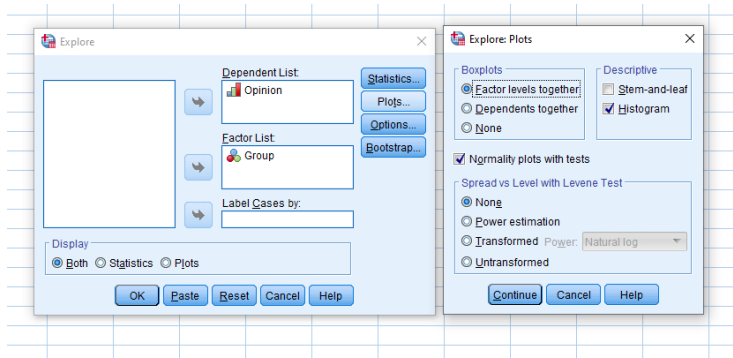
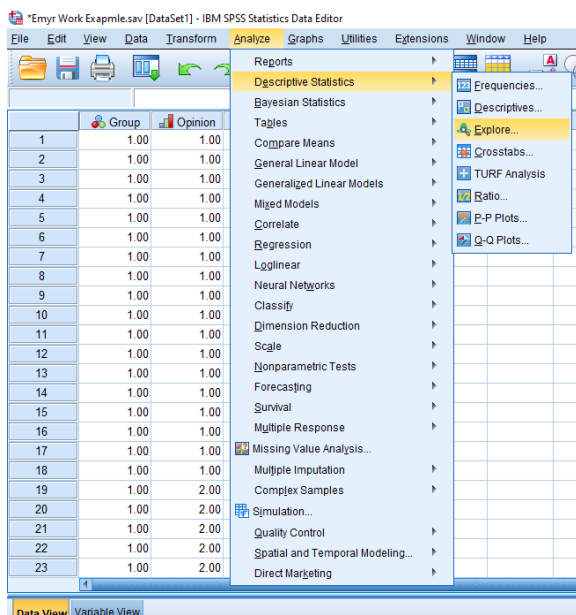
²¹⁵ Ibid.

²¹⁶ Ibid, 62.

²¹⁷ Ibid.

The p value represents the calculated ‘probability’ of a given incident. If the p value is above 0.05, then we are able to determine that the data is normally distributed, requiring the application of parametric tests. If it below this score, then the data is not normally distributed, requiring the application of non-parametric tests.

Non-parametric are different to parametric tests, as “the model structure is not specified a priori but determined from the data”.²¹⁸ Non-parametric tests are also known as free-distribution tests which do not follow a normal distribution.²¹⁹



²¹⁸ Nadim Nachar, “The Mann-Whitney U: A Test for Assessing Whether Two Independent Samples Come from the Same Distribution”, (2008) 4 Tutorials in Quantitative Methods for Psychology, 2008, 13.

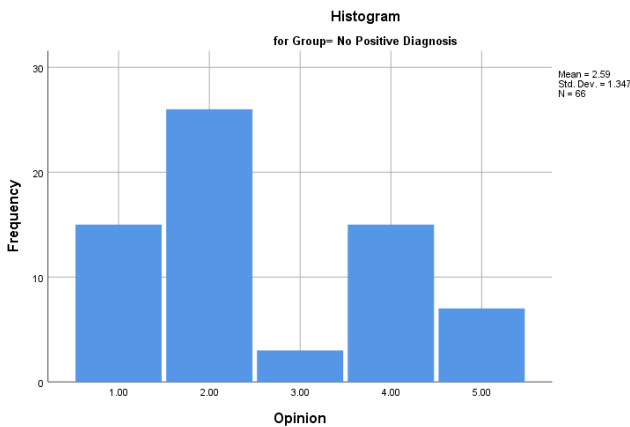
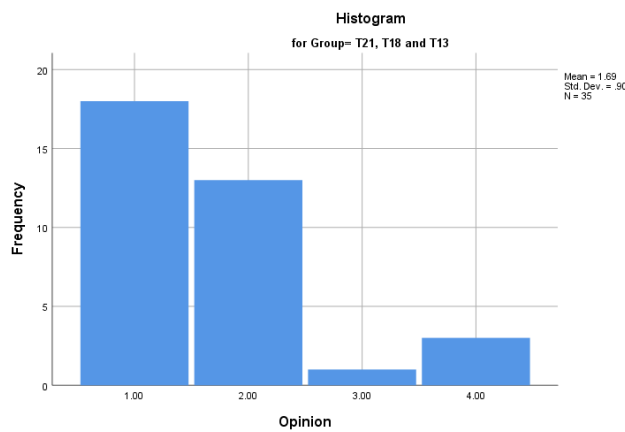
²¹⁹ Ibid.

Tests of Normality

Group	Kolmogorov-Smirnov ^a			Shapiro-Wilk			
	Statistic	df	Sig.	Statistic	df	Sig.	
Opinion	T21, T18 and T13	.291	35	.000	.716	35	.000
	No Positive Diagnosis	.291	66	.000	.845	66	.000

a. Lilliefors Significance Correction

Histograms



Following the performance of the test of normality on all questions, across both parent and HCP data sets, in every event, the data was not normally distributed; the p value was consistently below 0.05, and the histogram did not generate a bell-curve distribution.²²⁰ Therefore, it was necessary to run a non-parametric test for each question to determine whether there was a 'significant difference', in relation to the responses (opinions) between the selected groups.

²²⁰ Andy Field, 'Discovering Statistics Using IBM SPSS Statistics', (Sage Publishers 2013) 478.

Deciding between statistical tests can be a complex, but very important, task. Under this research paradigm, both T-tests and a Mann-Whitney U test were considered, to measure the significance between the populations. Both tests measure whether there is a relationship between two numeric variables. The T-test assumes that the variables conform to, what is called, a normal distribution (normality).²²¹ Therefore, to use the T-test, it must be decided first whether the variables are, in fact, normally distributed. The T-test is generally preferred, as it is more efficient in identifying a significant relationship. If the data is not normally distributed, then the Mann-Whitney U test is the recommended test to run on the data set.²²² The objective of running these tests is to provide a comparison between the responses from each population, and their significance; highlighting any statistical significance, or patterns, is necessary to provide a foundation for the qualitative data, to build upon and explore through qualitative means.²²³

Standard deviation is a measure of variability, and this must also be considered by the researcher, when performing the statistical tests.²²⁴ The standard deviation, in this research, related to the grouping of the answers from the sample population. It quantifies the grouping of the responses from the sample population and assesses its variability.²²⁵

The researcher decided that a two-tailed Mann-Whitney U test provided the most appropriate means of exploring the quantitative data. The 'null hypothesis' was measured – for each survey question response – following the application of the Mann-Whitney U test; that is, whether the two independent groups possess the same distribution, and were analogous.²²⁶ The 'null hypothesis' simply relates to whether the average response of the two sample populations differ significantly.

At this stage, consideration of the 'p value' is key. If the p value is greater than 0.05, the null hypothesis should not be disregarded, meaning the response of the two sample populations do not differ significantly. If it's less than 0.05, the null hypothesis can be disregarded, in favour of an alternative hypothesis, meaning the average response of the two samples

²²¹ Ibid, 480.

²²² Ibid, 481.

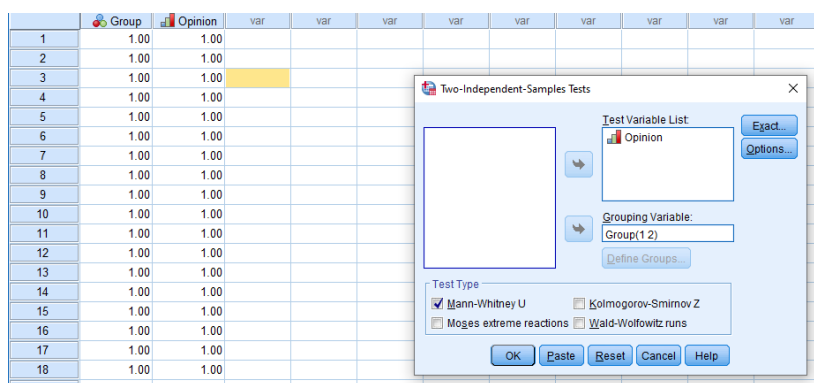
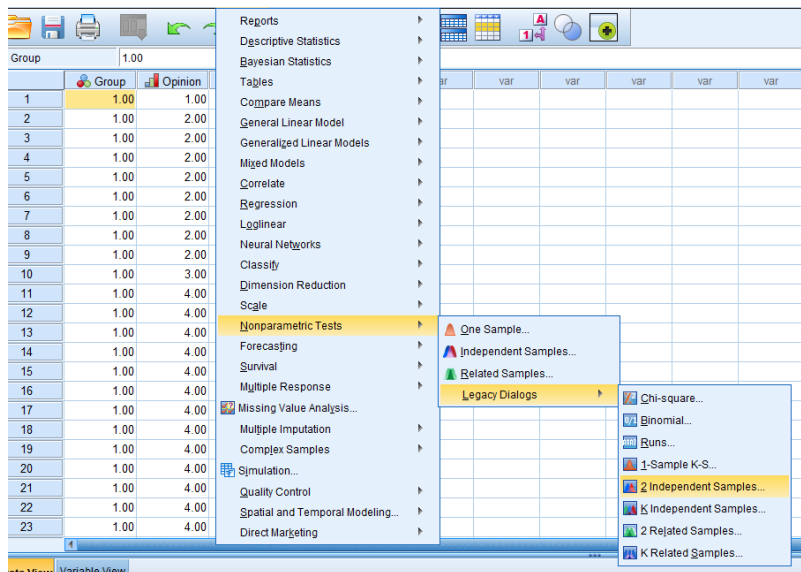
²²³ Ibid.

²²⁴ Ibid, 482.

²²⁵ Ibid.

²²⁶ Nadim Nachar, *'The Mann-Whitney U: A Test for Assessing Whether Two Independent Samples Come from the Same Distribution'*, (2008) 4 Tutorials in Quantitative Methods for Psychology, 19.

Figure 2: Data view showing the inputted data set. The researcher's objective was to explore whether there was a significant difference in responses between the two groups.



→ NPar Tests

Mann-Whitney Test

Ranks

	Group	N	Mean Rank	Sum of Ranks
Opinion	T21, T18 and T13	35	37.89	1326.00
	No Positive Diagnosis	66	57.95	3825.00
Total		101		

Test Statistics^a

	Opinion
Mann-Whitney U	696.000
Wilcoxon W	1326.000
Z	-3.450
Asymp. Sig. (2-tailed)	.001

a. Grouping Variable: Group

3.9 Ethical Considerations

It was imperative, as a socio-legal research, that the researcher upheld all key ethical and moral principles. Unethical practices, in research, may include: using data obtained from participants, without their consent or knowledge; coercing or deceiving the participants, during the course of the research; failing to disclose why the research is taking place; and exposing the participants to emotional or physical harm (or both).²²⁷ Ensuring the participants confidentiality, privacy and anonymity is also fundamental, particularly when conducting research into sensitive issues, on potentially vulnerable participants. These issues were thoroughly evaluated and assessed, in-line with professional advice, which formed the design of the research.

3.9.1 Research Information Sheet for Participants

All participants received a research information sheet, which highlighted the key practical and ethical considerations for the study. The participant information sheet consisted of two sections; section (a) and section (b).²²⁸

Section (a) answered questions pertaining to the implications of the study and its components: (i) why I am conducting the study, (ii) why they had been invited to take part in the research, (iii) whether they have to take part in the research, (iv) what is required from them if they decided to take part, (v) whether there are any risks or disadvantages to taking part in the research, (vi) any further support needed, (viii) what the possible benefits of taking part were, (ix) what happens once the research has finished, (x) what if there is a problem and (xi) if their taking part in the research would be kept confidential.

Section (b) answered questions pertaining to any further information: (i) contact details, (ii) complaints procedure, (iii) whether they are able to withdraw from the study, (iv) whether the information and data will be kept confidential at all times, (v) who has reviewed the

²²⁷ Mike McConville (n4) 63.

²²⁸ Please see appendix.

research study, (vi) financial/organisational elements of the research, (vi) whether it has any effect on the data and (viii) any further details.

3.9.2 Consent Forms

Participants were provided with a consent form to sign, which had to be returned before conducting the study. Six statements were included on the consent form, which participants had to read and sign individually (with their initials beneath each statement). Written consent was provided by participants at the end of the consent form, before taking part in the study. Verbal consent was given, again, before conducting the follow-up interviews. As consent is a dynamic process, the researcher reminded participants that they were able to withdraw from the study at any time, regardless of the initial written and oral consent.

3.9.3 Down's Syndrome Association (DSA) and Support Organisation for Trisomy 13 and 18 (SOFT) Ethical Review Process

The researcher approached the services development manager of the DSA²²⁹, and arranged a face-to-face meeting, to discuss the research study. Together, they discussed the research questions and design, ethical considerations, and the requirements the researcher must comply with, to work with the DSA. Following the meeting, the researcher was required to fill out the DSA research proposal form, that included questions on the project title, description, objectives, design (methods), populations (age range/sex/comorbid conditions), main outcome measure(s), name and status of the researcher and Swansea University's ethical approval letter. Once the researcher had filled out and sent the proposal, it was returned with feedback, which was subsequently incorporated into the research design. After a second meeting with the service development manager, the research study was advertised by the DSA. The researcher sent over the research information sheets and consent forms for the DSA, to review and distribute to the relevant sample population. The DSA were very supportive of all aspects of this PhD study.

²²⁹ The DSA is an organisation that exists to support individuals with Down's Syndrome and their families, and is available to provide information, advocacy and support. They also work alongside national screening programmes across England (FASP) and Wales (ASW) to offer support to parents who have had a high-risk or positive diagnosis of DS, and offer up to date, accurate training to researchers, HCPs and screening co-ordinators on Down's Syndrome.

The Chair of SOFT²³⁰ was also very supportive of the research. The researcher had to complete a similar form to that of the DSA, outlining the study's details, purpose, and ethical considerations. Following the study, all SOFT families, who participated in the research, relayed very positive feedback to the Chair, assuring the researcher that the study was executed sensitively, and adhered to ethical standards.

3.9.4 Private Clinic Study

Many ethical considerations had to be addressed, before engaging with this study. With the advice of both supervisors and Dr Beattie, open-ended survey questions were deemed most ethically and logistically appropriate, particularly in light of the developing COVID outbreak. All participants were briefed by the researcher, Dr Beattie, and the clinical manager, before allowing the participants to engage with the study. A consent form and participant information sheet, created by the researcher, was distributed to the parents, before engaging with the study. The researcher did not have any access to patients' private medical records or history.

Upon reflection, the researcher decided that the intended interviewing methods of data collection was not desirable, as parents were early in the pregnancy, and questioning may induce anxiety and stress. Interviewing may also impact on the relationship between parent and professional, if they deemed that consent, at this stage, was less than informed.

3.9.5 Anonymity and Confidentiality

To ensure that the participants were eligible to participate in the research, they were asked whether they would be happy to state their country, the year they had screening, and whether they had a baby with a trisomy, or not. However, the research questions were carefully designed, so that they could not reveal any information which could be linked to them personally, or expose their identity. Participants' age, name, gender and ethnicity were stripped, to ensure that participants remained anonymous. Their identity was replaced with a study number and letter (i.e A1, A2, A3). If participants accidentally disclosed information

²³⁰ SOFT work alongside national screening programmes across England and Wales (ASW and FASP) and provide parents with further information on the tested conditions, in particular, T18 and T13. SOFT created a close-knit network of trained volunteers who are able to deliver support to families through the screening journey. They also provide support to those parents who have ended or lost a pregnancy, as their baby had ES or PS. SOFT also work with HCPs and professional researchers to provide information, training and support.

that could have potentially revealed their identity, the researcher took the appropriate steps to remove these, once their permission had been granted.

Every participant, who decided to take part in the follow-up interview, was anonymised. The researcher irrevocably stripped the data of any direct identifiers, such as their name, age, race and gender, during the transcribing process. With face-to-face or Skype/Zoom interviews, the researcher assured participants that only he would be aware of their identity.

The researcher assured participants that all ethical duties were being upheld, to keep participants' information confidential, throughout the contingent stages: initial collection of information; use of information; dissemination of the findings; the storage of the information; and, importantly, the disposal of any material or records, which include personal information.

3.9.6 Storing Data

The researcher understood that information, which contains personal or identifiable data, falls within the ambit of the Data Protection Act. The researcher ensured participants that he followed the University's protocols for data protection. The data obtained from the surveys and interviews, were held on file (computerised and non-computerised) at the University, in a location which is only accessible by the researcher and supervisors. All paper information or data relating to the participants were placed in a secure locked filing system on the security-controlled Swansea University premises. The information obtained via email, and any data stored on computerised systems (recordings from interviews), were filed appropriately on the University's computer system. All files were encrypted, and password protected, which is only accessible to the researcher and agreed members of staff. Firewalls, anti-virus software, and other measures were undertaken, to ensure data protection. The researcher did not store any data on personal computer systems. As the researcher is funded by the ESRC, a requirement was that the data will be given to the UK Data Archive (UKDA). As such, this will include the materials collected (transcribed interviews and field notes), from participants. However, all potential identifiers have been stripped from the data, so that participants entirely unidentifiable and anonymised.

3.9.7 Limitations

Parent and HCP Studies

The primary limitation to the parent's study, was that participants would ultimately have to retrospectively recall their experiences, relying on memory. This could result in participants forgetting crucial aspects of their experience, or misremembering the various stages of trisomy screening and testing, impeding the credibility and reliability of the data. To reduce the existence of recall bias, the researcher decided to include parents who had trisomy screening/testing within 12-months of commencing the research. Recall bias recognises that research participants may not be able to accurately remember a past experience or are unable to recall particular details from the event in question, which could reduce the validity and reliability of the dataset. This also ensured that *all* parents experienced screening and/or testing since the implementation of the trisomy screening pathway (post-2018).

Another limitation was that the researcher appropriated the role of a counsellor, at times, particularly when interviewing parents who lost a baby with a trisomy. These discussions were often very emotional, and required the researcher to reflect on his own well-being during the course of the study.

The missing dimension, for both HCP and parent studies, was the observational method of data collection, with the NHS. After completing lengthy NHS ethics forms, with the researcher's R&D lead for this study, this process ended following the outbreak of COVID. While an observational dimension would have strengthened the validity and reliability of the data set, this was not possible, due to COVID restrictions: all booking appointments went online/telecommunication, and researchers were not able to gain access to hospital sites, due to national and local lockdowns.

Conducting surveys and interviews *after* parents have left the NHS system was for the benefit of the participants' well-being, transpiring to a high quality and rich data set. Conducting research on participants, who were still under the NHS trisomy screening programme, was not desirable, due to a number of professional and ethical reasons.

To many parents, screening and testing is an entirely new and unfamiliar experience. Thus, due to the nature of this research, it would not have been ethical, to survey and interview parents, who are currently going through trisomy screening. Indeed, if participants were to realise that their consent was less than perfect, it may impact on the doctor-patient relationship, throughout the remainder of the pregnancy.

Furthermore, parents may receive an unexpected result, inducing feelings of anxiety or distress. Conducting the research, after parents have left the pathway, would allow them to reflect on their experience. Some of the participants terminated a baby, based on an unexpected result. While a parent would never fully recover from this experience, conducting the study, once parents have disembarked from the testing pathway, allows time to regather and stabilise emotions, before sharing their experience.

While observational methods could have strengthened the credibility of the parent and HCP studies, 'Hawthorn Effect' could potentially provide a significant barrier to obtaining reliable data; this means that the behaviour of participants may change, in the presence of the researcher, to portray an ideal 'reality'. Indeed, particularly for the purposes of this thesis, it was foreseeable that parent and HCP may modify behaviour, to reflect an intended outcome. Observational methods, particularly in this field, could also impede on the parent-HCP relationship – particularly if parents suspect their consent is less than informed – impacting the depth and richness of the data.

Many studies, that focus on parents' decision-making and consent, have done so by evaluating ability of participants to recall specific information, given at consultations.²³¹ Consequently, this fails to provide an insight into parent and professional interests, during the process of providing and securing consent, for trisomy screening. Indeed, following the decisions in *Mordel* and *Montgomery*, consent is not a 'one-off' event: it extends across the entire pathway, which is not appreciated by many existing studies, in this area. The aim of this study was not focused on the ability of participants to recall, verbatim, the conversations HCPs had with them, as this merely assesses the participants' memory.

Private Clinic Study

The research had to finish, following the second COVID lockdown. The number of parents accessing private care reduced dramatically, and thus the recruitment of parents significantly decreased. While this was not ideal, the researcher had captured a satisfactory understanding

²³¹ Susan Michie, Elizabeth Dormandy and Theresa M Marteau, 'The Multi-Dimensional Measure of Informed Choice: A Validation Study', (2002) 48 *Patient Education Counselling*, 91. This was amended by, Celine Lewis, Melissa Hill and Lyn S Chitty, "Development and Validation of a Measure of Informed Choice for Women Undergoing Non-Invasive Prenatal Testing for Aneuploidy", (2016) 24 *European Journal of Human Genetics*, 186. This still relies heavily on the recall of specific information.

of parents' experiences, and initial areas of interests, in terms of why and how the private NIPT market may impact on the provision of NHS parental support, for decision-making and consent, for trisomy screening. Ultimately, the purpose of this small study was to highlight initial areas of interest, for future researchers to explore, in greater depth.

Summary and Conclusion of Methods and Methodology

The aim of this chapter was to outline and explain the research paradigm for this thesis. Following a contextualisation of other socio-legal work on consent in healthcare, and empirical bioethics research, this chapter has explored and located an appropriate research methodology, to best answer the key research questions, for this thesis. A range of both quantitative and qualitative methods, and subsequently procedures for data analysis, have also been outlined for gathering the desired data, in this regard.

Chapter 4: HCP and Parent Study Quantitative Results and Analysis

4.1 Justifying the Use of Surveys for Framing an Initial Empirical Response

As discussed previously in chapter 3, quantitative methods are utilised to allow the researcher to map the terrain of a given area of study. The use of surveys allows researchers to map and locate initial themes, that could be further explored, using qualitative methods and techniques.

Following the pragmatic research paradigm set out in chapter 3, the researcher conducted surveys to fulfil the 'first quantitative stage', of the two-phase, explanatory mixed methods research design. As there was a lack of existing contemporary research on parent and professional interests, in the context of consent and decision-making for trisomy screening, the aim, at this stage in the research, was to map the terrain; this process would provide a means to uncover lacunae in legal coverage, and locate initial broader themes and patterns of behaviour, for further qualitative exploration. To answer the research questions, it was necessary to explore HCP and parent responses, on the following key areas of initial interest, identified from chapters 1 and 2: provision of information; supporting understanding; supporting choice; communication and relationship between parent and HCP; understanding of 'risk'; and understanding of alternative methods of treatment.

Using appropriate statistical testing (a Mann-Whitney U 2-tailed test – see chapter 3), this first quantitative stage would provide an initial empirical foundation to scope phenomenological socio-legal issues, which *could* be statistically significant to parent and professional interests, in the context of informed consent and trisomy screening.

To frame an initial empirical response from the key considerations, identified in chapters 1 and 2, on parent and professional interests for delivering and securing consent, the surveys were carefully designed to achieve this aim and objective. Indeed, the survey for parents sought to explore their experiences of delivering consent for trisomy screening, and whether any initial patterns or behaviours were identifiable and/or comparable, between the variables. The survey, for HCPs, was designed to explore their roles and experiences of securing consent for trisomy screening, and also to identify any themes or areas of interest, in relation to the systemic considerations, underlined in *Mordel*.

The purpose of this chapter is to present the quantitative findings, from the parent and HCP surveys. The variables (discussed below) are addressed in turn, to demonstrate whether any statistically significant differences between responses are identifiable. The purpose of this is to locate any initial areas of significance, or to uncover conceptual relationships, between populations. Using qualitative techniques, these would subsequently be explored in further depth, for a rich and comprehensive understanding of the key themes and areas of interest.

4.2 Variables in the Quantitative Study

Parent Groups

The key variables, in the parent study, compared responses of those parents who received trisomy screening in England and Wales, parents who had a high-and low-risk result following screening, and parents who had a baby with a tested condition (either DS, ES or PS) and those who did not.

HCP Groups

The key variables, in the HCP study, compared the responses of the different professions (midwives, sonographers and consultants), and their respective nations (England and Wales).

The Populations Considered in this Research:

Parent Study	HCP study
<i>Parents from Wales</i>	<i>Midwives from Wales</i>
<i>Parents from England</i>	<i>Sonographers from Wales</i>
<i>Parents who were high-risk following screening</i>	<i>Consultants from Wales</i>
<i>Parents who were low-risk following screening</i>	<i>Midwives from England</i>
<i>Parents Who Had a Baby with a Tested Condition (DS, ES or PS).</i>	<i>Sonographers from England</i>

Parents Who Did Not Have a Baby with a Tested Condition (DS, ES or PS).	Consultants from England
--	---------------------------------

4.3 Parent Study Survey Results

After receiving information (paper-based, online and/or verbal) from the healthcare professional (HCP) at first contact or the booking appointment, do you feel they provided the information you needed on the purpose of trisomy screening and testing?

<u>Question 1</u>	England	Wales
Response		
Strongly Agree	17.42%	15%
Agree	24.24%	17.42%
Neutral	8.33%	6.43%
Disagree	36.36%	37.86%
Strongly Disagree	13.64%	23.57%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.04$).

<u>Question 1</u>	Low-Risk	High-Risk
Response		

Strongly Agree	17.29%	12.07%
Agree	18.69%	27.59%
Neutral	7.01%	8.62%
Disagree	38.32%	32.76%
Strongly Disagree	18.69%	18.97%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.878$).

<u>Question 1</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		
Strongly Agree	18.42%	15.81%
Agree	21.05%	20.51%
Neutral	0%	8.55%
Disagree	42.11%	36.32%
Strongly Disagree	18.42%	18.80%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.972$).

The majority of parents disagreed, or strongly disagreed, that they were provided with the information they needed on trisomy screening. There was a statistically significant difference between the English and Welsh groups. This response was also particularly prominent among parents who had a baby with a trisomy.

After receiving information from the HCP(s) on trisomy screening and testing at first contact or the booking appointment, did the information help support your understanding of the conditions being screened for, that is, Down’s Syndrome and/or Edwards’ Syndrome and Patau’s Syndrome?

<u>Question 2</u>	England	Wales
Response		
Strongly Agree	9.85%	13.57%
Agree	24.24%	17.86%
Neutral	6.06%	6.43%
Disagree	33.33%	35.71%
Strongly Disagree	26.52%	26.43%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 (p = 0.955).

<u>Question 2</u>	Low-Risk	High-Risk
Response		
Strongly Agree	13.55%	5.17%
Agree	21.50%	18.97%
Neutral	7.01%	3.45%

Disagree	32.71%	41.38%
Strongly Disagree	25.23%	31.03%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.076$).

<u>Question 2</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		
Strongly Agree	5.26%	12.82%
Agree	21.05%	20.94%
Neutral	2.63%	6.84%
Disagree	34.21%	34.62%
Strongly Disagree	36.84%	24.79%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.100$).

The majority of parents disagreed, or strongly disagreed, that the information helped support their understanding of the trisomies. This response was particularly prominent among parents who had a baby with a trisomy, and those who were high-risk.

After receiving information from the HCP(s) on trisomy screening and testing at first contact or the booking appointment, did the information help support your understanding of the methods of screening and testing?

<u>Question 3</u>	England	Wales

Response		
Strongly Agree	12.12%	12.86%
Agree	18.18%	14.29%
Neutral	6.06%	3.57%
Disagree	39.39%	38.57%
Strongly Disagree	24.24%	30.71%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.302$).

<u>Question 3</u>	Low-Risk	High-Risk
Response		
Strongly Agree	14.49%	5.17%
Agree	17.29%	12.07%
Neutral	4.67%	5.17%
Disagree	39.25%	37.93%
Strongly Disagree	24.30%	39.66%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.009$).

<u>Question 3</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		

Strongly Agree	10.53%	12.82%
Agree	10.53%	17.09%
Neutral	2.63%	5.13%
Disagree	34.21%	39.74%
Strongly Disagree	42.11%	25.21%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.064$).

The majority of parents disagreed, or strongly disagreed, that the information helped support their understanding of the methods of screening and testing. This response was particularly prominent among parents who had a baby with a trisomy, and those who were high-risk.

Did you understand that the decision to have trisomy screening and/or testing was entirely your choice?

<u>Question 4</u>	England	Wales
Response		
Strongly Agree	14.39%	9.29%
Agree	22.73%	22.14%
Neutral	7.58%	7.86%
Disagree	37.12%	39.29%
Strongly Disagree	18.18%	21.43%

N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.264$).

<u>Question 4</u>	Low-Risk	High-Risk
Response		
Strongly Agree	11.21%	13.79%
Agree	23.83%	17.24%
Neutral	7.48%	8.62%
Disagree	37.85%	39.66%
Strongly Disagree	19.63%	20.69%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.791$).

<u>Question 4</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		
Strongly Agree	10.53%	11.97%
Agree	21.05%	22.65%
Neutral	2.63%	8.55%
Disagree	39.47%	38.03%
Strongly Disagree	26.32%	18.80%

N/A		
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The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.328$).

The majority of parents disagreed, or strongly disagreed, that they understood the decision to have trisomy screening and/or testing was their choice. This response was particularly prominent among those parents who were high-risk, and those who had a baby with a trisomy.

Did the HCPs make you aware that you were able to refuse trisomy screening or testing at any time?

<u>Question 5</u>	England	Wales
Response		
Strongly Agree	12.12%	10.71%
Agree	26.52%	20.71%
Neutral	7.58%	7.86%
Disagree	37.12%	37.14%
Strongly Disagree	16.67%	23.57%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.162$).

<u>Question 5</u>	Low-Risk	High-Risk
Response		
Strongly Agree	11.68%	10.34%
Agree	23.83%	22.41%
Neutral	7.01%	10.34%
Disagree	36.92%	37.93%
Strongly Disagree	20.56%	18.97%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.985$).

<u>Question 5</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		
Strongly Agree	7.89%	11.97%
Agree	21.05%	23.93%
Neutral	7.89%	7.69%
Disagree	47.37%	35.47%
Strongly Disagree	15.79%	20.94%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.717$).

The majority of parents disagreed, or strongly disagreed, that they were made aware of the fact they could refuse trisomy screening and/or testing, at any time. This response was

particularly prominent among those parents who were low-risk, and those who had a baby with a trisomy.

Do you feel that the HCP(s) adopted an unbiased approach when supporting your decision-making throughout the pathway?

<u>Question 6</u>	England	Wales
Response		
Strongly Agree	10.61%	7.86%
Agree	15.15%	15%
Neutral	7.58%	9.29%
Disagree	45.45%	41.43%
Strongly Disagree	21.21%	26.43%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.435$).

<u>Question 6</u>	Low-Risk	High-Risk
Response		
Strongly Agree	10.28%	5.17%
Agree	14.95%	15.52%
Neutral	8.88%	6.90%

Disagree	44.86%	37.93%
Strongly Disagree	21.03%	34.48%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.085$).

<u>Question 6</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		
Strongly Agree	2.63%	10.26%
Agree	13.16%	15.38%
Neutral	0%	9.83%
Disagree	42.11%	43.59%
Strongly Disagree	42.11%	20.94%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.004$).

The majority of parents disagreed, or strongly disagreed, that the HCP(s) adopted an unbiased approach, when supporting decision-making, throughout the pathway. This response was particularly prominent among parents who were high-risk, and those who had a baby with a trisomy. Statistically, there was a significant difference between those parents who had a baby with a trisomy, and those who did not.

Do you feel that the HCP(s) effectively communicated with you to support your decision-making for trisomy screening?

<u>Question 7</u>	England	Wales
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Response		
Strongly Agree	15.15%	17.86%
Agree	19.70%	21.43%
Neutral	9.09%	2.86%
Disagree	30.30%	29.29%
Strongly Disagree	25.76%	28.57%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.001$).

<u>Question 7</u>	Low-Risk	High-Risk
Response		
Strongly Agree	17.24%	20.56%
Agree	29.31%	26.17%
Neutral	7.01%	6.90%
Disagree	24.77%	18.97%
Strongly Disagree	22.90%	27.59%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a low-risk result following

screening, and those who had a high-risk result, as the p value was more than 0.05 (p = 0.464).

<u>Question 7</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		
Strongly Agree	15.79%	20.94%
Agree	7.89%	22.65%
Neutral	5.26%	5.98%
Disagree	44.74%	27.35%
Strongly Disagree	26.32%	23.08%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 (p = 0.024).

The majority of parents disagreed, or strongly disagreed, that the HCP(s) effectively communicated with them, in the context of supporting decision-making, for trisomy screening and/or testing. This response was particularly prominent among the parents who were high-risk, and those who had a baby with a trisomy. Statistically, there was a significant difference between the English and Welsh groups.

Did you understand what high-chance (high-risk) and low-chance (low-risk) meant in relation to your trisomy screening result?

<u>Question 8</u>	England	Wales
Response		
Strongly Agree	12.88%	12.14%
Agree	23.48%	22.86%

Neutral	7.58%	5%
Disagree	36.36%	35%
Strongly Disagree	19.70%	25%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.442$).

<u>Question 8</u>	Low-Risk	High-Risk
Response		
Strongly Agree	14.49%	5.17%
Agree	22.90%	24.14%
Neutral	5.61%	8.62%
Disagree	35.05%	37.93%
Strongly Disagree	21.96%	24.14%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.288$).

<u>Question 8</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		

Strongly Agree	5.26%	13.68%
Agree	28.95%	22.22%
Neutral	0%	7.26%
Disagree	44.74%	34.19%
Strongly Disagree	21.05%	22.65%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.532$).

The majority of parents disagreed, or strongly disagreed, that they understood what high-risk and low-risk meant, in relation to their screening result. This response was particularly prominent among those parents who were high-risk, and those who had a baby with a trisomy.

Do you feel that the HCP(s) placed *equal* importance on supporting your understanding of both the advantages and disadvantages of your chosen methods of screening and/or testing?

<u>Question 9</u>	England	Wales
Response		
Strongly Agree	9.09%	15%
Agree	19.70%	17.86%
Neutral	8.33%	7.86%
Disagree	41.67%	35%

Strongly Disagree	21.21%	24.29%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.699$).

<u>Question 9</u>	Low-Risk	High-Risk
Response		
Strongly Agree	14.02%	5.17%
Agree	19.16%	17.24%
Neutral	8.41%	6.90%
Disagree	37.85%	39.66%
Strongly Disagree	20.56%	31.03%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.038$).

<u>Question 9</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		
Strongly Agree	2.63%	13.68%
Agree	10.53%	20.09%
Neutral	5.26%	8.55%
Disagree	39.47%	38.03%

Strongly Disagree	42.11%	19.66%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.000$).

The majority of parents disagreed, or strongly disagreed, that the HCP(s) placed equal importance on supporting their understanding of the advantages and disadvantages, of trisomy screening and/or testing. This response was particularly prominent among those parents who were high-risk, and those who had a baby with a trisomy. Statistically, there was a significant difference between all variables.

Do you feel that there were any barriers to communication between you and the HCPs before consenting to decisions on trisomy screening and/or testing?

<u>Question 10</u>	England	Wales
Response		
Strongly Agree	12.12%	12.86%
Agree	21.21%	21.43%
Neutral	10.61%	8.57%
Disagree	35.61%	39.29%
Strongly Disagree	20.45%	17.86%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.830$).

<u>Question 10</u>	Low-Risk	High-Risk
Response		
Strongly Agree	14.95%	3.45%
Agree	21.50%	20.69%
Neutral	9.81%	8.62%
Disagree	35.51%	44.83%
Strongly Disagree	18.22%	22.41%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a low-risk result following screening, and those who had a high-risk result, as the p value was more than 0.05 ($p = 0.055$).

<u>Question 10</u>	Trisomy (DS, ES or PS)	No Trisomy
Response		
Strongly Agree	5.26%	13.68%
Agree	21.05%	21.37%
Neutral	10.53%	9.40%
Disagree	39.47%	37.18%
Strongly Disagree	23.68%	18.38%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from those parents who had a baby with DS, ES or PS, and those who did not, as the p value was more than 0.05 ($p = 0.230$).

The majority of parents disagreed, or strongly disagreed, that there were barriers to communication between them and the HCP(s), before consenting to trisomy screening and/or testing. This response was particularly prominent among those parents who were high-risk, and those who had a baby with a trisomy.

Concluding Comments on Parent Quantitative Findings

A strong theme that emerged from the data was the clear dissatisfaction of the majority of *all* parent groups – particularly those who were high-risk and/or had a baby with a trisomy – with their experience of decision-making and consent along the trisomy pathway. This was evident across all identified areas of interest, such as: provision of information; support provided for their understanding of the trisomies, methods of screening and testing, screening results, and the associated advantages and disadvantages; supporting parent choice or refusal; communication between parent and HCP; and whether the HCPs remained unbiased. Of those parents who were low-risk or did not have a baby with a trisomy, the data suggests their experience was significantly better, in this regard.

In the second phase of this explanatory research paradigm, qualitative techniques will be employed to further explore the apparent dissatisfaction, and to confirm, or refute, the quantitative data. This has also provided the researcher with a clearer understanding of the terrain, and of key areas of significant interest.

4.4 HCP Study Survey Results

Do you believe there are appropriate systems in place to secure parents’ consent?

<u>Question 1</u>	English Midwives	Welsh Midwives
Response		
Strongly Agree	10.53%	14.53%

Agree	23.68%	24.36%
Neutral	2.63%	8.12%
Disagree	28.95%	33.76%
Strongly Disagree	34.21%	19.23%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.131$).

<u>Question 1</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	15.91%	12.14%
Agree	24.24%	24.29%
Neutral	7.58%	7.14%
Disagree	29.55%	36.43%
Strongly Disagree	22.73%	20%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.688$).

<u>Question 1</u>	English Consultants	Welsh Consultants
Response		

Strongly Agree	14.95%	10.34%
Agree	24.30%	24.14%
Neutral	7.94%	5.17%
Disagree	33.64%	31.03%
Strongly Disagree	19.16%	29.31%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.166$).

The majority of midwives, sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, that there were appropriate systems in place to secure parent consent. This response was prominent among all identified HCP groups. Statistically, there was no significant difference between each population.

Are you clear on your role for securing parents' consent for trisomy screening?

<u>Question 2</u>	English Midwives	Welsh Midwives
Response		
Strongly Agree	13.16%	14.10%
Agree	10.53%	18.38%
Neutral	0%	9.40%
Disagree	39.47%	38.46%
Strongly Disagree	36.84%	19.66%

N/A		
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The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.034$).

<u>Question 2</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	14.39%	13.57%
Agree	15.91%	18.57%
Neutral	7.58%	9.26%
Disagree	40.15%	37.14%
Strongly Disagree	21.97%	22.14%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.838$).

<u>Question 2</u>	English Consultants	Welsh Consultants
Response		
Strongly Agree	15.42%	8.62%
Agree	17.76%	15.52%
Neutral	8.41%	6.90%
Disagree	40.19%	32.76%
Strongly Disagree	18.22%	36.21%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.018$).

The majority of midwives, sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, that they were clear on their role for securing parents' consent for trisomy screening. This response was prominent among all identified HCP groups. Statistically, there was a significant difference between the response of English and Welsh midwives, and English and Welsh consultants.

Are you confident supporting parents' informational needs on trisomy screening?

<u>Question 3</u>	English Midwives	Welsh Midwives
Response		
Strongly Agree	5.26%	16.24%
Agree	18.42%	20.94%
Neutral	5.26%	7.69%
Disagree	44.74%	35.47%
Strongly Disagree	26.32%	19.66%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.067$).

<u>Question 3</u>	English Sonographers	Welsh Sonographers

Response		
Strongly Agree	12.12%	17.14%
Agree	18.18%	22.86%
Neutral	7.58%	7.14%
Disagree	37.12%	36.43%
Strongly Disagree	25%	16.43%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 (p = 0.054).

<u>Question 3</u>	English Consultants	Welsh Consultants
Response		
Strongly Agree	15.89%	10.34%
Agree	20.56%	20.69%
Neutral	7.01%	8.62%
Disagree	36.92%	36.21%
Strongly Disagree	19.63%	24.14%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 (p = 0.366).

The majority of midwives, sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, that they were confident supporting parents' informational needs on trisomy screening. This response was prominent among midwives and

sonographers. Statistically, there was a significant difference between English and Welsh midwives.

Are you confident supporting parents' understanding of trisomy screening?

<u>Question 4</u>	English Midwives	Welsh Midwives
Response		
Strongly Agree	18.42%	16.67%
Agree	18.42%	20.51%
Neutral	5.26%	8.55%
Disagree	31.58%	34.19%
Strongly Disagree	26.32%	20.09%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.665$).

<u>Question 4</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	17.42%	16.43%
Agree	18.94%	21.43%
Neutral	6.82%	9.29%

Disagree	35.61%	32.14%
Strongly Disagree	21.21%	20.71%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.777$).

<u>Question 4</u>	English Consultants	Welsh Consultants
Response		
Strongly Agree	17.76%	13.79%
Agree	18.22%	27.59%
Neutral	8.41%	6.90%
Disagree	34.58%	31.03%
Strongly Disagree	21.03%	20.69%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.827$).

The majority of midwives, sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, that they were confident supporting parents' understanding of trisomy screening. This response was prominent among all HCP groups. Statistically, there was no significant difference between each population.

Do you feel confident supporting parents' choices along the trisomy pathway?

<u>Question 5</u>	English Midwives	Welsh Midwives

Response		
Strongly Agree	7.89%	19.66%
Agree	23.68%	24.79%
Neutral	7.89%	10.26%
Disagree	44.74%	29.91%
Strongly Disagree	15.79%	15.38%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.112$).

<u>Question 5</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	17.42%	18.57%
Agree	24.24%	25%
Neutral	9.58%	7.14%
Disagree	30.30%	33.57%
Strongly Disagree	18.18%	12.86%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.518$).

<u>Question 5</u>	English Consultants	Welsh Consultants
Response		
Strongly Agree	14.02%	6.90%
Agree	29.91%	24.14%
Neutral	10.28%	8.62%
Disagree	24.77%	39.66%
Strongly Disagree	21.03%	20.69%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.013$).

The majority of midwives, sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, that they were confident support parents' choices along the trisomy pathway. This response was prominent among midwives and sonographers. Statistically, there was a significant difference between English and Welsh midwives, and English and Welsh consultants.

Do you feel confident supporting parents' understanding of 'risk' in the context of their screening results?

<u>Question 6</u>	English Midwives	Welsh Midwives
Response		
Strongly Agree	10.53%	15.38%

Agree	18.42%	24.79%
Neutral	7.89%	12.68%
Disagree	34.21%	30.34%
Strongly Disagree	28.95%	19.23%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.108$).

<u>Question 6</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	15.15%	14.29%
Agree	21.97%	25.71%
Neutral	12.88%	7.14%
Disagree	29.55%	32.14%
Strongly Disagree	20.45%	20.71%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.925$).

<u>Question 6</u>	English Consultants	Welsh Consultants
Response		

Strongly Agree	16.36%	8.62%
Agree	24.30%	22.41%
Neutral	9.81%	10.34%
Disagree	29.91%	34.48%
Strongly Disagree	19.63%	24.14%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.151$).

The majority of midwives, sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, that they were confident supporting parents' understanding of 'risk', in the context of their screening result. This response was prominent among all HCP groups. Statistically, there was no significant difference between the populations.

Do you feel that there are barriers to communication between professional and parent along the trisomy pathway?

<u>Question 7</u>	English Midwives	Welsh Midwives
Response		
Strongly Agree	13.16%	20.51%
Agree	10.53%	23.08%
Neutral	2.63%	10.68%
Disagree	42.11%	29.49%

Strongly Disagree	31.58%	16.24%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.012$).

<u>Question 7</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	14.39%	17.14%
Agree	25%	28.57%
Neutral	11.36%	7.86%
Disagree	26.52%	25%
Strongly Disagree	22.73%	21.43%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.468$).

<u>Question 7</u>	English Consultants	Welsh Consultants
Response		
Strongly Agree	20.09%	6.90%
Agree	22.43%	18.97%
Neutral	10.28%	6.90%
Disagree	28.97%	37.93%
Strongly Disagree	18.22%	29.31%

N/A		
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The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.002$).

The majority of midwives, sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, that there are barriers to communication between professional and parent, along the pathway. This response was prominent among midwives. Statistically, there was a significant difference between English and Welsh midwives, and English and Welsh consultants.

Do you feel you have received up to date training on securing consent since the implementation of the trisomy pathway?

<u>Question 8</u>	English Midwives	Welsh Midwives
Response		
Strongly Agree	13.16%	14.53%
Agree	21.05%	24.36%
Neutral	10.53%	10.68%
Disagree	42.11%	33.76%
Strongly Disagree	13.16%	16.67%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.800$).

<u>Question 8</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	15.15%	13.57%
Agree	23.48%	24.29%
Neutral	12.88%	8.57%
Disagree	35.61%	34.29%
Strongly Disagree	12.88%	19.29%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.356$).

<u>Question 8</u>	English Consultants	Welsh Consultants
Response		
Strongly Agree	15.42%	10.34%
Agree	24.30%	22.41%
Neutral	10.28%	12.07%
Disagree	34.58%	36.21%
Strongly Disagree	15.42%	18.97%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.319$).

The majority of midwives, sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, that they received up to date training on securing consent,

since the implementation of the trisomy pathway. This response was prominent among all HCP groups. Statistically, there was no significant difference between all populations.

Do you feel that you have received up to date training on supporting parent understanding of DS, ES and PS since the implementation of the pathway?

<u>Question 9</u>	English Midwives	Welsh Midwives
Response		
Strongly Agree	13.16%	19.66%
Agree	18.42%	25.64%
Neutral	7.89%	9.40%
Disagree	34.21%	26.92%
Strongly Disagree	26.32%	18.36%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.120$).

<u>Question 9</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	14.39%	15.71%
Agree	22.73%	26.43%
Neutral	9.85%	8.57%

Disagree	27.27%	28.57%
Strongly Disagree	25.76%	20.71%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was a significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.02$).

<u>Question 9</u>	English Consultants	Welsh Consultants
Response		
Strongly Agree	18.69%	13.79%
Agree	25.23%	27.59%
Neutral	9.81%	6.90%
Disagree	26.17%	29.31%
Strongly Disagree	20.09%	22.41%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.447$).

The majority of midwives, in Wales, either agreed, or strongly agreed, that they received up to date training on supporting parent understanding of the trisomies. However, the majority of midwives in England disagreed, or strongly disagreed, with the question. The majority of sonographers and consultants, across England and Wales, either disagreed, or strongly disagreed, with the question. This response was prominent among midwives. Statistically, there was significant difference between English and Welsh sonographers.

Do you feel that you have received up to date training on non-invasive prenatal testing (NIPT) for the purpose of supporting parent decision-making?

<u>Question 10</u>	English Midwives	Welsh Midwives
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Response		
Strongly Agree	9%	8%
Agree	23%	27%
Neutral	0%	5%
Disagree	23%	21%
Strongly Disagree	45%	39%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the responses from the English and Welsh populations, as the p value was less than 0.05 ($p = 0.078$).

<u>Question 10</u>	English Sonographers	Welsh Sonographers
Response		
Strongly Agree	7%	7%
Agree	16%	20%
Neutral	9%	7%
Disagree	18%	33%
Strongly Disagree	50%	33%
N/A		

The result of the Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.198$).

<u>Question 10</u>	English Consultants	Welsh Consultants
Response		
Strongly Agree	20%	12%
Agree	24%	27%
Neutral	15%	6%
Disagree	21%	25%
Strongly Disagree	20%	30%
N/A		

The result of the SPSS Mann-Whitney U (2-tailed) test demonstrates there was no significant difference between the English and Welsh populations, as the p value was more than 0.05 ($p = 0.577$).

The majority of midwives and sonographers, across England and Wales, either disagreed, or strongly disagreed, that they received up to date training on NIPT, for the purpose of supporting parents' decision-making. This response was prominent among midwives and sonographers. The majority of English consultants agreed, or strongly agreed, with the question, while the majority of Welsh consultants disagreed, or strongly disagreed. Statistically, there was no significant difference between populations.

Concluding Comments on HCP Quantitative Findings

A strong theme that emerged from the HCPs data was the clear dissatisfaction of all HCP groups – particularly midwives and sonographers – with their experience operating under current systems of consent for trisomy screening and/or testing. This was evident across all the identified areas of interest: appropriateness of systems for securing parent consent; HCP understanding of role for securing parent consent for trisomy screening; confidence

supporting parents' informational needs on trisomy screening; confidence supporting parents' understanding of trisomy screening; confidence supporting parents' choices along the trisomy pathway; confidence supporting parents' understanding of 'risk' for screening results; training on supporting parents understanding of the trisomies; and training on non-invasive prenatal testing (NIPT).

Using qualitative techniques, it will be necessary for the researcher to explore this apparent dissatisfaction, in more depth. This will also allow the researcher to confirm, or refute, the quantitative data, providing a clearer understanding of the terrain surrounding these key areas of interest.

4.5 Summary and Conclusion on Quantitative Data

This chapter has outlined the quantitative data gathered from the parent and HCP surveys. The selected procedure of data analysis (bivariate descriptive) has also identified significance and correlations, pertaining to patterns of behaviour and experience of consent and trisomy screening, between the populations. This first phase has also provided the researcher with a contextual understanding of initial themes from the key areas of interest, to further explore using qualitative methods.

Chapter 5 – Parent Groups Qualitative Findings

5.1 Justifying the Use of Qualitative Interviews for Framing an Empirical Response

The purpose of this chapter is to systematically outline the qualitative findings from the parent study, providing the second stage to the explanatory research paradigm. Following the initial themes identified from the quantitative data (see chapter 4 ‘concluding comments’ section), it was necessary for the researcher to probe into the themes, pertaining to the key areas of interest. The aim of this second phase, under the explanatory research paradigm, was to gain an in-depth understanding of parent interests and experiences, regarding the process of delivering consent, across the trisomy pathway. This qualitative exploration was particularly significant when comparing and/or integrating the data collected, which either confirmed, or refuted, initial observations and assumptions.

Inevitably, themes overlapped, reflecting their interesting relationships and interactions, in this regard. However, the researcher attempted to distinguish (as far as possible) between themes, to provide clarity for the reader. The researcher also drew upon previous empirical research (primarily those on trisomy screening and consent), legal scholarship, and case law, to support the study’s findings.

While the researcher obtained an abundance of rich qualitative data, quotations were carefully selected to best represent the key considerations and areas of interest, within each identified theme. Subject to the considerations raised in *Montgomery* and *Mordel*, compounded by the initial quantitative themes, the aim of this chapter was to present parents’ account of their experiences decision-making and delivering consent, across the trisomy pathway. The aim was also to uncover any additional themes or key areas for consideration, providing the researcher with a rich contextual understanding and insight into the identified key areas of interest.

5.2 Provision of Information on Trisomy Screening

5.2.1 Parent Dishonesty

Dishonesty, and a lack of transparency between parents and HCPs, emerged as a prominent theme, among parent groups. Parents admitted to falsely confirming that they had read the trisomy screening information packs, before attending the appointments when, in fact, they had not. The reason for this was to appear as a ‘responsible’ parent:

*“Being completely honest, I did not bother reading the booklets they gave us. I put them in the kitchen drawer and did not go back to them. Obviously, that is irresponsible of me not to read it before seeing the midwife. I did tell the midwife I read it, though; I did not want to look irresponsible. I wanted to be a good mum (laughing)”.*¹

This finding is consistent with Dormandy *et al.*, who explained that parents may falsely confirm that they have read the information materials, to appear as “responsible” and “good” parents.² It has also been widely cited that parents seldom deliberate on the screening information, before making a decision to engage with the screening pathway.³ Another explanation for this, according to John *et al.*, is that HCPs may not be sufficiently directing parents to the information materials, following the initial consultation.⁴ This finding is concerning, as dishonesty, in this regard, may impede on the HCP’s ability to effectively support parents’ informational needs.⁵

5.2.2 Preferred Delivery of Information

A preference to receive information face-to-face, with the HCPs, was demonstrated by all parent groups. Parents also stated that the HCPs relied too heavily on the paper-based materials, and failed to go into sufficient depth, during pre-screening consultations:

¹ low-chance, baby without trisomy, Wales.

² Dormandy E, Michie S, Hooper R, Marteau T.M, ‘Low uptake of prenatal screening for Down syndrome in minority ethnic groups and socially deprived groups: a reflection of women’s attitudes or a failure to facilitate informed choices?’ (2005) 34 *International Journal of Epidemiology*, 346.

³ Ahmed, S., Bryant, L.D., Tizro, Z. & Shickle, D, ‘*Interpretations of informed choice in antenatal screening: A cross-cultural, Q-methodology study*’, (2012) 74 *Soc Sci Med*, 997.

⁴ Sophie John, Maggie Kirk, Emma Tonkin and Ian Stuart-Hamilton, ‘*Influence of midwife communication on women’s understanding of Down Syndrome screening information*’, (2019) 27 *British Journal of Midwifery*, 1.

⁵ *Ibid.*

*“We got the information (on trisomy screening) with the handheld pregnancy notes. She (midwife) did rely a lot on the fact that we had already read this information, before we went to the appointment. I’d much rather we discussed the information more face-to-face, at the clinic”.*⁶

This finding may explain the initial theme, identified from the quantitative data, that the majority of parent groups felt that the HCP did not provide the information they required on the purpose of screening and testing, at first contact. Previous research has established that, this parent preference, rests on the expectation that HCPs will cover *all* of the information, during the booking appointment.⁷ John *et al.* state that, while face-to-face methods have proven to be the most effective means of imparting information on pregnancy care, NHS time constraints, limited access to resources, and cost – both in time and financially – mean that this expectation may be unrealistic and unreasonable.⁸ Sole reliance on a face-to-face approach to care management, may also encourage infantilisation of parents.⁹

5.2.3 The Desire to Tailor/Personalise Information

Receiving tailored and personalised information was desired by all parent groups. However, they also revealed that, due to shame for not reading the information, and fear of appearing irresponsible, parents understood that this would be difficult for HCPs to fulfil, during consultations:

*“I do think this job is hard for the midwife because it does rely on the fact you have actually read the information, before going to the appointment. I did not tell her (midwife) I had not read the stuff, so I guess that makes it hard for the midwife to know what is best for me, at that time, when I don’t even know what is best”.*¹⁰

This finding suggests the initial theme, identified from the quantitative data, that parents did not feel the HCP(s) provided the information they needed to support their understanding of screening’s purpose, may be misleading, as parents indicate they also have a responsibility to

⁶ low-chance, baby without trisomy, England.

⁷ Dormandy E and others (n2), 346.

⁸ Sophie John (n4) 2.

⁹ Beulen L and others, ‘The effect of a decision aid on informed decision-making in the era of non-invasive prenatal testing: a randomised controlled trial’ (2016) 24 European Journal of Human Genetics, 2016, 1409.

¹⁰ low-chance, baby without trisomy, Wales.

be honest and transparent, and to take steps to read the information provided. This finding is consistent with Hartwig *et al.*, who found that a lack of transparency, between HCPs and parents, interferes with the HCP's ability to effectively tailor the information, subject to patients' needs.¹¹ The study found that building rapport, with the parents, is paramount to effectively executing this task.¹²

Parents suggested that constructing a set of questions could assist HCPs, to efficiently personalise the information, subject to the particular patient. They also explained that this would better focus their attention on the information they consider relevant:

"I think there should be a list of standard questions women get on why we want screening, what would we do if we had a low or high-risk result, and if we would have the amnio if we were high-risk. I think that would have encouraged me to think for myself about what information I wanted. It would also help the midwife know what information we find most important to our situations".¹³

This is supported by John *et al.*, who found that constructing a predetermined set of questions for parents to answer during counselling, could provide the desired efficiency and outcomes, under current time constraints.¹⁴ This method has also been hailed as a means for HCPs to effectively build the desired rapport with the parents, under the pressures of clinical practice.¹⁵

5.2.4 Preference for Online Resources

The popularity of utilising the internet and online resources for obtaining information on screening and obstetric care, emerged as a prominent theme. Parents revealed that they often supplemented the NHS information packs on trisomy screening with online sources. Many of the internet sources referenced were unreliable, unvalidated or unregulated:

¹¹ Tanja Schlaikjaer Hartwig and others, 'High risk – What's next? A survey study on decisional conflict, regret, and satisfaction among high-risk pregnant women making choices about further prenatal testing for fetal aneuploidy', (2019) 39 *Prenat Diagn*, 635.

¹² *Ibid.*

¹³ low-chance, baby without trisomy, England.

¹⁴ Sophie John (n4) 3.

¹⁵ *Ibid.*

“We used the internet for everything: it was just far more convenient. We found ourselves on the strangest websites looking at the information for the syndromes and things. The pictures we found on Patau were scarring. Photos of dead babies with Patau were shown on these bizarre sites, you know, with eulogies beneath them; it was absolutely terrifying. I think we need to be pointed in the right way, you know, the reliable information websites.”¹⁶

This finding is supported by the work of Mercer *et al.* and Lagan *et al.* on the use of internet sources for maternity care.¹⁷ In *Mordel*, the Claimant also admitted that she accessed online sources, rather than reading the information packs the NHS had provided, to inform her decision.¹⁸ This may speak to the tedium parents experience of expertise, preferring to explore internet sources for information, at their own leisure and convenience.

5.2.5 Parents’ Previous Experience of Screening and Provision of Information

Parents, who had previous experience of screening, admitted that they did not feel the need to read the information packs or materials, before attending the appointments, due to their prior knowledge of the pathway:

“This was my third pregnancy, so I was comfortable going ahead with the combined test. I didn’t really pay attention to the booklets they gave me, because I did not feel the need to. I remember cutting the midwife off when she was explaining it, just said ‘yes, yes, I’ll have it all’ (laughing). That was naïve of me, but at the time I didn’t realise that they offered screening for those two other conditions, since my last one (pregnancy).”¹⁹

This is supported by Carroll *et al.*’s study of parents in this category, demonstrating that parents will have varying appetites for information.²⁰ While this finding is unsurprising, failure

¹⁶ low-chance, baby without trisomy, Wales.

¹⁷ Mercer M.B, Agatista P.K, Farrell R.M, ‘What patients are reading about non-invasive prenatal testing: an evaluation of Internet content and implications for patient-centred care’, (2014) 34 *Prenatal Diagnosis*, 986, and Lagan B.M, Sinclair M, Kernohan W.G, ‘A web-based survey of midwives’ perceptions of women using the Internet in pregnancy: A global phenomenon’, (2011) 27 *Midwifery*, 273.

¹⁸ *Mordel v Royal Berkshire NHS Trust* (2019) EWHC 2591.

¹⁹ high-chance, baby with trisomy (DS), England.

²⁰ F. E. Carroll, A. Owen-Smith, A. Shaw, and A. A. Montgomery, ‘A qualitative investigation of the decision-making process of couples considering prenatal screening for Down syndrome,’ (2012) 32 *Prenatal Diagnosis* 63; see also, Emily Jackson, ‘Challenging the Comparison in Montgomery Between Patients and ‘Consumers Exercising Choices’, (2021) 00 *Medical Law Review*, 15.

to read the information subject to the recent amendments – addition of ES/PS and NIPT – could mean that key considerations for decision-making are being overlooked.

5.2.6 Information Preferences on the Trisomies

Parents revealed they did not know what ES and PS were, before entering and *after* leaving the trisomy pathway. They explained they left the trisomy screening pathway with little, if any, awareness of ES and PS, in terms of what they are or what they mean for the baby. During the research interviews, parents explained that ES and PS are overlooked in the paper-based materials, as DS takes centre stage, primarily due to the purported rarity of ES and PS:

“I had, and still have, no idea what Edwards’ and Patau were. All I could say for certain was that Edwards’ and Patau were less common than Down’s, but that was it. Everyone has heard of Down’s, so it takes that focus, I think. I also think because I thought they were so rare, I did not bother worrying, so did not feel the need to read the information on it. Just put it to the back of my mind if I am honest.”²¹

They suggested that a review of the order and layout of the information materials on the trisomies may also be necessary, to support parents’ informational needs. All parent groups explained that the presentation of the ‘trisomy’ pathway as a genetic model, in the information materials, as opposed to distinguishing between individual phenotype, misled them to believe that the three conditions possess the *same* physical characteristics and health outcomes:

“I thought they were all the same thing because it was presented in that way; it was called ‘trisomy’ screening so, naturally, I thought the conditions were all the same. I think those other two (ES and PS) need individual attention if that makes sense; separate them out to show us they are very different things, instead of the same thing.”²²

This finding is significant, as it could explain the initial theme from the quantitative data, that the majority of parents felt the information, provided by HCPs, did not help support their understanding of the trisomies, particularly in relation to ES and PS. Indeed, these findings are significant, as the implications of obtaining a higher-risk or positive diagnosis for ES and

²¹ high-chance, baby without trisomy, England.

²² high-chance, baby without trisomy, Wales.

PS, and the subsequent decisions parents would be required to make, are substantially different to that of DS, due to their aetiology and pathogenesis. The decisions and care pathways, available to parents, substantially differ subject to each trisomy, and parents must be informed in this regard.

5.2.7 Information on Practicalities of Screening and/or Invasive Testing

Parents revealed they tended to not read the information on the methods of screening and testing, preferring “just to leave all that stuff to them (HCPs)”. Reasons for not reading the information on the methods of screening and testing, commonly related to stress and fear. Participants also demonstrated a considerable amount of trust towards HCPs, and their ability to advise them appropriately on such matters:

“I had no idea what the different tests were, or what I needed to do for the test. I quickly looked at the booklet on that stuff, but it all sounded pretty intimidating and scary. That needle one (amnio) really scared me. I just thought why stress myself out when I can just leave all that stuff in their (HCPs) hands; they know what they’re doing.”²³

This finding suggests the initial theme identified from the quantitative data, that parents did not feel that the information provided by the HCP(s) supported their understanding of the methods of screening and testing, may be misleading, as parents revealed that they did not read the information provided. This finding is supported by John *et al.*, who found that feelings of fear and intimidation were experienced by parents, due to their unfamiliarity with screening and testing. This fear may also be explained by the study of Heyman *et al.* – and the expert witness statement of the defendant in *Mordel* – both asserting that parents often fail to distinguish between the practicalities of screening and invasive testing.²⁴

Parents explained they were only provided with the more detailed information on invasive testing, and its implications, following a higher-risk screening result.²⁵ They preferred to receive this information earlier in the pathway, as a means to prepare for such an eventuality:

²³ High-chance, baby without trisomy, England.

²⁴ Heyman B and others, ‘On being at higher risk: A qualitative study of prenatal screening for chromosomal anomalies’, (2006) 62 *Social science & medicine*, 2360.

²⁵ Sophie John and others, ‘Influence of midwife communication on women’s understanding of Down Syndrome screening information’, (2019) 27 *British Journal of Midwifery*, 3.

“We did not discuss the amnio(centesis) at the booking appointment, and we were not encouraged to look at the information on it, before giving our consent to the combined screening. We should get reminded that most people won’t need the amnio, but we still need to think about this type of testing and what it means before we get the high-risk letter.”²⁶

This finding may provide an explanation for the theme identified in the quantitative study, that the majority of parent groups did not feel that the information provided supported their understanding of the methods of screening and testing, as parents are particularly dissatisfied with the information provided on diagnostic testing. This is supported by John *et al.*, who found that the midwives, in their study, did not adequately counsel parents on invasive testing at the booking appointment, with some even *refusing* to discuss invasive testing, unless the results are high-risk.²⁷ Heyman *et al.* found that review of the post-screening options, before undertaking screening, is crucial to parents’ decision-making, as the parents, in their study, declined screening to avoid obtaining a ‘risk’ status, due to the possibility of having to undertake invasive testing.²⁸

5.2.8 Overwhelmed with Information

Being overwhelmed with information, in early pregnancy, was a strong theme among all parent groups. They found that they were not able to absorb and reflect on the information they received, during the booking appointment, as trisomy screening represents only a fraction of the information they receive on antenatal and obstetric care:

*“You have to remember that we do not just get the information about the Down’s screening, we get so much more information about lots of different things about the pregnancy like nutrition, food, breastfeeding, and other tests. The sheer volume of information we get is so unrealistic; Down’s screening was only a small part of the stuff we need to read”.*²⁹

This feeling of being overwhelmed is also consistent with the initial theme from the quantitative data, that the majority of all parent groups did not feel the provision of

²⁶ High-chance, baby with trisomy (DS), England.

²⁷ Sophie John (n25) 3. See also, Marie-Anne Durand, Mareike Stiel, Jacky Boivin and Glyn Elwyn, ‘Information and decision support needs of parents considering amniocentesis: interviews with pregnant women and health professionals’, (2010) 13 Health Expectations, 126.

²⁸ Heyman B and others, (n24) 2360.

²⁹ low-chance, baby without trisomy, England.

information supported their understanding of the components of the trisomy pathway. This finding is supported by John *et al.* and Saunders *et al.* of parents in this population.³⁰ These academics suggested that the risk of overwhelming parents will only intensify in the future, accounting for the ever-increasing amount of information HCPs are required to review with parents, during clinical appointments.³¹

Engaging into a process of prioritisation of information was a suggestion, made by parents, to negate the risk of being overwhelmed, enhancing their ability to absorb and retain the requisite information, early in the pathway. They suggested that the information on screening should take precedent early in gestation, with information on perinatal and postnatal care being reviewed at a later stage.

"I loved my midwife, well I had two, but the first one I had did talk far too long about breastfeeding and other things, at the appointment, when I could have had more conversation on the immediately relevant information on the Down's screening. I just think issues like breastfeeding can wait until afterwards. I just think this will relieve them (HCPs) of the pressure their under, to cover all the information in forty-odd minutes".³²

This suggestion was also raised by Beulen *et al.*, in their research, to overcome such risks.³³ However, this recommendation begs the question whether its curative potential, to overcome the risk of overwhelming parents, is limited in this regard, as the overall quantity of information has not been reduced, nor has it changed in nature. It may be that the prioritisation of information must also be accompanied by palatable summarisations.³⁴

5.2.9 Preparedness to Ask Questions to Obtain Information

Parents reported that their preparedness to ask questions, depended on the HCP's role. They explained that midwives were typically the first port of call, in relation to questions on matters of screening. On the other hand, parents perceived sonographers as technicians, whose role

³⁰ Sophie John and others (n2), 2; and, Sanders J, Hunter B, Warren L, 'A wall of information? Exploring the public health component of maternity care in England', (2016) 34 *Midwifery*, 253.

³¹ *Ibid.*

³² low-chance, baby without trisomy, Wales.

³³ Beulen L, and others, 'The effect of a decision aid on informed decision-making in the era of non-invasive prenatal testing: a randomised controlled trial', (2016) 24 *European Journal of Human Genetics*, 1409.

³⁴ Bret Asbury, 'Fostering Informed Choice: Alleviating the Trauma of Genetic Abortions', (2015) 25 *Cornell Journal of Law and Public Policy*, 305.

does not entail answering questions; they described sonographers as being there ‘just to do the scan’:

“I did not really feel the need to ask the sonographer any questions because I did think that was part of their job. I think the midwife was more open to questions from us. The scan with the sonographers was very much in and out, no hanging around asking questions.”³⁵

This finding is supported by the research of Steen *et al.*, suggesting that both varying degrees of confidence, and the perception of the HCP’s role, are leading factors in this regard.³⁶ Di Mattei *et al.* underlined that more rigorous research is required to explore the interaction of parents’ characteristics, and the dynamic between parents and HCPs, in the context of prompting questions, during counselling.³⁷

Confidence was another factor going to parents’ preparedness to ask questions. They viewed HCPs as authoritative and omniscient figures, and confirmed they did not possess the confidence to ask questions, during counselling, due to the power dynamic. Feelings of intimidation, stress, feeling overwhelmed and not knowing what questions to ask, were all common reasons preventing parents asking questions:

“Screening is all new to most of us. I had loads of questions I wanted to ask, but I just did not have that confidence. It is all so intimidating because they (HCPs) obviously know a lot more than us, so I did not want to look stupid. At that stage, you are just so overwhelmed by it all”³⁸

Steen *et al.* explained that rapport, between parent and HCP, is fundamental to increasing parents’ confidence to ask questions, during counselling.³⁹

Parents felt asking questions was “inappropriate”, as they feared appearing antagonistic or undermining the HCP’s quality of counselling. They outlined that they were willing to *reserve* their right to ask questions, to ensure preservation of the relationship between HCPs and parents:

³⁵ low-chance, baby without trisomy, England.

³⁶ Sanne L. van der Steen and others, ‘Offering a choice between NIPT and invasive PND in prenatal genetic counselling: the impact of clinician characteristics on patients’ test uptake’, (2019) 27 European Journal of Human Genetics, 235-243.

³⁷ Valentina Di Mattei and others, “Decision-making factors in prenatal testing: A systematic review”, (2021) Health Psychology Open, 20.

³⁸ low-chance, baby without trisomy, England.

³⁹ Sanne L. van der Steen and others, (n36) 235.

“It's difficult isn't it because you don't want to ask the midwife too many questions because when I started to ask questions, I felt that she (midwife) got the impression I doubted what she was saying. I did not want to appear antagonistic, because she (midwife) got slightly defensive when we did ask questions.”⁴⁰

This finding is supported by the research of Hartwig *et al.*, who confirmed that preserving rapport was a key objective of parents, and that they were unwilling to jeopardise this relationship, by challenging the ‘authority’ of HCPs.⁴¹ This also speaks to managing and effectively moderating the power dynamic, between parents and HCPs, to restore the desired balance, in this regard. Employing open-ended questions may empower parents to elaborate on any concerns they have, and to encourage those less confident parents to engage into an open and honest dialogue.⁴²

⁴⁰ low-chance, baby without trisomy, England.

⁴¹ Tanja Schlaikjaer Hartwig and others, ‘High risk – What’s next? A survey study on decisional conflict, regret, and satisfaction among high-risk pregnant women making choices about further prenatal testing for aneuploidy’, (2019) 39 *Prenat Diagn*, 635.

⁴² Sanne L. van der Steen and others, (n36) 235.

5.3 Parent Understanding

5.3.1 Shame and Embarrassment for Failing to Understand Information

Parents explained that they felt reluctant, or embarrassed, to reveal that they did not understand the information on trisomy screening, during counselling. They exhibited feelings of self-blame for not understanding the information, and said they “gave up” trying to understand it. Thus, they admitted to falsely confirming that they had understood the information when, in fact, they had not:

*“She (midwife) did ask me if I understood what she had said about the screening, and I just said, ‘yes’, when really I did not understand. I am sure most mums are too proud or ashamed to admit that they did not understand all the information, on the Down’s testing”.*⁴³

This is supported by Wiel who explained that embarrassment, due to not understanding the information, typically prevents parents from making informed decisions, as this issue is often not raised by the parents, at the appropriate time.⁴⁴ Asbury underlined that parents will often feel self-blame for not understanding the information on screening, which may also diminish their confidence, in this regard.⁴⁵

5.3.2 Understanding the Trisomies

During the research interviews, it was apparent that parents’ understanding of ES and PS was limited, superficial, or incorrect.⁴⁶ They commonly described ES and PS as “the other ones they offer” with DS, or “like Down’s but worse”. Parents also revealed that because screening was offered as ‘trisomy’ screening, this gave the impression that all trisomies shared the same, or had similar, health characteristics and prognosis:

⁴³ low-chance, baby without trisomy, England.

⁴⁴ Jon Wiel, *Psychosocial Genetic Counselling*, (OUP 2000) 298.

⁴⁵ Bret Asbury (n34) 306.

⁴⁶ Meyer, R.E and others, ‘*Survival of Children with Trisomy 13 and Trisomy 18: A Multi-State Population Based Study*’, (2015) *American Journal of Medical Genetics*, 13.

*“I can’t really believe I screened for them (ES and PS), without really understanding what they were. I thought they were just bad versions of Down’s. I think it is clear, from this conversation, that they (HCPs) need to make sure that we understand what these conditions are before we consent to screening. I could not tell you what they are even today”.*⁴⁷

This is supported by Leuthner and Acharya, who observed that many myths and misconceptions exist, surrounding the prognosis and health characteristics of ES and PS, often being confused with DS.⁴⁸ This misunderstanding risks interfering with parents’ understanding of the unique health characteristics and prognosis of the individual trisomies, and the requisite care pathways or interventions they would need to consider, in the event of a high-risk result or positive diagnosis.⁴⁹

Parents, who had a baby with a trisomy, felt that generalisations were made by HCPs, during counselling, in terms of their health characteristics and prognosis, following a positive diagnosis. This theme was particularly prominent among those parents who had a baby with ES or PS. They reported that HCPs used the phrase “incompatible with life” to describe ES and PS, along with associating the condition as being “deadly” or “lethal”:

*“I resigned myself to the fact my baby was going to die. Not once did I hear of the possibility of (names baby) being sat here on my lap today, babbling away and giggling. The consultant said she was incompatible with life, and that the condition was deadly. It is not true for all babies; it’s such a damaging misconception.”*⁵⁰

This finding is consistent with the study by Janvier and Watkins, that highlighted “three hundred and third-two parents who had a baby with ES or PS reported being told by HCPs that their child was incompatible with life”, amounting to eighty-seven percent of the research population.⁵¹ Janvier and Watkins stated that “by declaring that T13 and T18 are universally lethal conditions with ‘unacceptable outcomes’ for which lifesaving intervention

⁴⁷ low-chance, baby without trisomy, Wales.

⁴⁸ Steven R Leuthner and Krishna Acharya, ‘Perinatal Counseling Following a Diagnosis of Trisomy 13 or 18: Incorporating the Facts, Parental Values, and Maintaining Choices’, (2020) 20 Adv Neonatal Care, 204.

⁴⁹ Emre Yanikkerem, Semra Ay, Alev Ciftci, Sema Ustgorul and Asli Goker, ‘A survey of the awareness, use and attitudes of women towards Down syndrome screening’, (2012) 22 Journal of Clinical Nursing, 1748.

⁵⁰ high-chance, baby with trisomy (ES), England.

⁵¹ Annie Janvier and Andrew Watkins, “Medical interventions for children with trisomy 13 and trisomy 18: what is the value of a short disabled life?”, (2013) 102 Acta Paediatrica, 1112.

are not indicated, HCPs have harmed this population”.⁵² Indeed, as Coleman and Janvier assert, “lethal language leads to lethal decisions” and “use of the word lethal in counselling often proves to be a self-fulfilling prophecy”.⁵³ Lyneham concludes that HCPs’ “thoughtless words and actions” result in parents experiencing preventable harm.⁵⁴

Parents, who had a baby with a trisomy, expressed concern towards the information on the trisomies in the current pregnancy packs, reporting that they were outdated. This theme was particularly prominent among those parents who had a baby with ES and PS. Parents, in this population, found that the information on the trisomies referred to research dating as far back as the sixties and seventies, and did not provide an accurate account of the conditions:

*“The information was out-of-date. The information in the leaflets, on Edwards’, referenced research from the sixties. Medical science has come a long way since then, so they must refer to recent studies.”*⁵⁵

This finding is consistent with the Nuffield Council report which found that information produced by individual NHS hospitals included information on the trisomies which were out-of-date.⁵⁶ However, previous studies concluded that NHS materials on ES and PS must be based on current research and experiential knowledge.⁵⁷ In accordance with clinical guidelines, the provision of accurate, balanced and *current* information, with reference to the relevant support groups and programmes, is integral to support decision-making.

These findings may provide an explanation for the initial theme in the quantitative study, that the majority of all parent groups (particularly those who had a baby with a trisomy), did not have a sufficient understanding of the trisomies. This also supports the quantitative data, that parents, particularly those who had a baby with a trisomy, did not feel that the information *supported* their understanding of the trisomies. The findings also suggest that this theme is

⁵² Ibid, 1111.

⁵³ Annie Janvier and Andrew Watkins (n51), 1111, and Priscilla Coleman, ‘*Diagnosis of Fetal Anomaly and the Increased Maternal Psychological Toll Associated with Pregnancy Termination*’, (2012) 30 Issues in Law and Medicine, 17.

⁵⁴ Joy Lyneham, ‘*Is there harm in silence?*’, (2010) 36 Journal of Medical Ethics, 642.

⁵⁵ high-chance, baby with trisomy (ES), Wales

⁵⁶ Nuffield Council Report (2017), 48.

⁵⁷ Gabriel Macias and Cheryl Riley, ‘*Trisomy 13: Changing Perspectives*’, (2016) 35 Neonatal Netw, 31; and Brian G Skotko, ‘*Postnatal diagnosis of Down syndrome: synthesis of the evidence on how best to deliver the news*’, (2009) 124 Paediatrics, 751.

particularly pertinent to parents' understanding of ES and PS, and what these conditions mean for the baby's prognosis.

5.3.3 Understanding Trisomy Screening and Testing

Parents explained that their understanding of the difference between screening (combined/quadruple screening) and diagnostic testing (amniocentesis/CVS), was poor. However, they also revealed that they were focussed on amniocentesis, due to the physical risks, overlooking the other methods of screening. Indeed, during the research interviews, parents' understanding of the distinction between these methods of screening and testing, was limited or incorrect:

"Yeah, well, I went to the appointment and signed in, but I thought I was getting the needle test, with the ultrasound. It sounds incredibly stupid now, but I turned up on the day full of adrenaline thinking I was getting the needle test."⁵⁸

This finding is consistent with the expert witness statement in *Mordel*, that parents frequently attend the examination appointment (for the purposes of the 12-week NT measurement), under the assumption of having invasive testing.⁵⁹ This is also supported by John *et al.*, who observed that parents assumed combined screening involved invasive techniques, underlining a concerning lack of understanding of the practicalities of screening and testing.⁶⁰ The study, by John *et al.*, revealed that midwives proffered the option of screening, before reviewing the information on the practical differences, between *both* screening and testing.⁶¹

Parents explained they were unclear as to the purpose and of invasive testing, and its practicalities, before consenting to screening:

"I have to say I did ask her (midwife) about the amnio at the first (booking appointment), but she said, 'we'll cross that bridge when we come to it'. That was the extend of our conversation on that; it was just focussed on the combined test at that point, you know, the ultrasound scan."

⁵⁸ low-chance, baby without trisomy, England.

⁵⁹ Mordel (n12) 56.

⁶⁰ Sophie John and others, 'Influence of midwife communication on women's understanding of Down Syndrome screening information', (2019) 27 British Journal of Midwifery, 1.

⁶¹ Ibid.

This finding may explain the initial theme from the quantitative data, that the majority of parent groups did not have a clear understanding of the methods of screening and/or testing. It is clear, from the findings, that the differentiation between ‘screening’ and ‘diagnostic’ (invasive) is problematic for parent understanding, and could be a probable cause of this initial theme. This is supported by the work of Marteau *et al.*, that highlighted parents’ complete lack of understanding of invasive testing.⁶² John *et al.* revealed that only a small percentage of the midwives, in their study, reviewed the information on invasive testing at the booking appointment, with some *refusing* to discuss the information, unless the parents obtained a higher-risk result.⁶³ Previous studies also highlighted that parents were not provided with sufficient information on invasive testing, and were unable to make informed decisions for this option.⁶⁴

5.3.4 Use of Medical Jargon

The use of medical jargon or terminology posed a significant barrier to parents’ understanding of trisomy screening. Jargon was reported in the context of describing the trisomies, and its use on the maternity serum report letters. The use of abbreviations, medical terms and unexplained complex statistics, increased anxiety and hindered interactivity, between parent and professional. Parents suggested that such terms should be explained during counselling, or clearly signposted in the information materials:

*“The word ‘trisomy’ was thrown around without actually reminding us of what this term referred to. The screening midwife and sonographer kept using the phrase ‘NT measurement’, NT this, NT that; we did not have a clue what ‘NT’ meant. My maternity report letter was full of abbreviations I did not understand; if I just get my letter now (participant pulls out letter), it says: CRL 56.9mm, hCGb 66 ng/mL, PAPP-A 3383 mU/L, NT 1.5mm, hCGb MoM 1.99 and so on (puts letter away). I just thought, ‘what does this all mean going forward?’”.*⁶⁵

This may support the initial quantitative finding, that the majority of parents, particularly those who had a high-risk result or baby with a trisomy, did not feel HCP(s) effectively

⁶² Mateau T.M, “Towards informed decision about prenatal testing: a review”, (1995) 15 Prenatal Diagnosis, 1215.

⁶³ Sophie John (n60), 3.

⁶⁴ Green JM, Hewison J and others, ‘Psychosocial aspects of genetic screening of pregnant women and newborns: a systematic review’, (2004) 8 Health Technology Assessment, 23.

⁶⁵ low-chance, baby without trisomy, Wales.

communicated with them to support decision-making. This finding is consistent with Asbury, who observed that HCPs often fail to appropriately simplify complex medical language and concepts during counselling, concluding that a “bureaucratic style of communication” impedes parents’ ability to make informed decisions.⁶⁶

⁶⁶ Bret Asbury (n34), 306.

5.4 Parent Decision-Making and Choice

5.4.1 Parents' Predetermined Decision-Making and Choice

Parents revealed that they often made a decision whether to undertake screening, or not, *before* engaging with the information and/or counselling. They explained that once a decision had been made, they did not consider or engage with *any* of the information on screening, at the booking appointment:

"I went off my gut and we just decided that we wanted screening; that was it. I knew when I fell pregnant I wanted all the tests and things, so the information they gave us did not make any difference to my decision. I knew I wanted it and that was that".⁶⁷

This is consistent with the facts of *Mordel*, as the Claimant appeared to have made an instinctive or intuitive decision to undertake screening, before obtaining the information materials. Crombag *et al.* and Skirton *et al.* asserted that parents will typically make a decision on screening, either before or just after conception, having not received any information on it, to inform their decision.⁶⁸ Jackson noted that, "patients do not necessarily take medical decisions after having received information about a treatment ... it is not uncommon for patients to have already made the decision to be treated before encountering the doctor" basing their decisions on instinct or intuition.⁶⁹

5.4.2 Access to Trisomy Screening

Free access to screening, on the NHS, was a key motivator for parents to engage with trisomy screening. They underlined that due care and attention would have been exercised on consideration of whether the option to screen was the right choice for them, if they were required to pay for it:

"I knew I wanted to check if my baby was healthy: screening is free, so it makes sense to just have it. That is the main reason why I had it (screening). Do I think the number of mothers

⁶⁷ low-chance, baby without trisomy, England.

⁶⁸ Neeltje M Crombag and others, 'Determinants affecting pregnant women's utilization of prenatal screening for Down syndrome: a review of the literature', (2013) 26 J Matern Fetal Neonatal Med, 1676; and, Skirton H, Barr O, 'Antenatal screening and informed choice: a cross-sectional survey of parents and professionals', (2010) 26 Midwifery, 596.

⁶⁹ Emily Jackson, 'Challenging the Comparison in Montgomery Between Patients and 'Consumers Exercising Choices'', (2021) 00 Medical Law Review, 14.

having screening would drop if they had to pay for it? Absolutely, no question; you do not give it a second thought.”⁷⁰

Davis also identified that the uptake of screening may be influenced by its free availability, suggesting an interplay with opportunism, and parent choice.⁷¹ This academic felt that the presence of opportunism, in screening, may hinder informed choice, without effective care management by HCPs.⁷²

The decision to screen for ES and PS was considered as an *accompaniment* to the decision to screen for DS, by all parents, describing it as a ‘screening package’. They recommended a fragmentation of the pathway to ensure that counselling is bespoke to each condition which, they explained, is not operational under the current ‘genetic model’ of ‘trisomy’ screening:

*“I just thought, you know, if they are already going to test me for Down’s Syndrome, they may as well test for the other two (ES and PS,) while they are there. I think I screened for those other ones (ES and PS) because I could, it was being offered, so why not go for it? The NHS would not offer screening for the Patau and Edward ones, unless they wanted you to screen for it”.*⁷³

Furthermore, parents alleged that they were *unaware* that they had opted to screen for ES and PS with DS. They explained that they were under the impression that screening targeted DS, and that only upon the return of their screening maternity report, were they made aware of the fact that they were also screened for ES and PS:

“I have spoken to a few others in our group (SOFT) who had a higher-risk result for trisomy 13 and trisomy 18 and we all had no idea that they were also being screened for with Down’s. I only realised that it (screening) picks up T13 and T18 after I had the phone call from my screening midwife explaining this.”⁷⁴

This finding may support the initial theme from the quantitative data, that parents, particularly those who had a baby with a trisomy, did not feel that the decision to have

⁷⁰ low-chance, baby without trisomy, England.

⁷¹ Dena S Davis, ‘*Opportunistic Testing: the Death of Informed Consent?*’, (2013) 23 Health Matrix: Journal of Law-Medicine, 35.

⁷² Ibid.

⁷³ low-chance, baby without trisomy, England.

⁷⁴ high-chance, baby with trisomy (ES), England.

screening was entirely their choice. While this oversight may be due to the fact that parents failed to read the information packs, this finding is indicative of consent being less than informed to screen for *all three* trisomies. This may further justify a fragmentation of the trisomy pathway, to further support parents' decision-making.

5.4.3 Expectation to Undertake Trisomy Screening

All parent groups reported a sense of expectation to undertake trisomy screening. They underlined that, even if they were aware that screening was optional, 'conformity' ultimately influenced their decision to screen. During the research interviews, parents revealed that conforming to the notion of a 'responsible parent' – in the eyes of *both* HCPs and family – meant they felt an expectation to have all the 'routine' tests. They also underlined that the construction of the trisomy pathway, and design of the programme, further compounds this sense of conformity, and to embrace 'routine' care:

"All my friends had the screening done when they were pregnant, so I thought it was the right thing to do, and the expected thing to do. It is the default position to have screening, and I would have been causing an issue if I did not have it. The way it is set up feels like there is much more support to have screening, than not".⁷⁵

This finding may provide another explanation for the initial theme identified in the quantitative data, that the majority of all parent groups did not feel that screening was ultimately their own autonomous choice. Hawthorn and Ahern also found that parents felt pressured to provide their consent for screening, as they wanted to appear as 'responsible parents', conforming with perceived social norms.⁷⁶ Research by Berg *et al.* and Gottfredsdottir *et al.*, found that, this apparent social norm, is the primary factor which motives parents to engage with screening, borne from a societal perception of DS, as a severe or serious disability.⁷⁷ It may also be that the implementation of antenatal screening

⁷⁵ low-chance, baby without trisomy, Wales.

⁷⁶ Fiona Hawthorne and Kathy Ahern, "*Holding our breath': the experience of women contemplating nuchal translucency screening*", (2009) 22 Applied Nursing Research, 236.

⁷⁷ van den Berg, M and others, 'Understanding pregnant women's decision making concerning prenatal screening' (2008) 27 Health Psychology, 430; and Gottfredsdóttir, H., Björnsdóttir, K. & Sandall, J, '*How do prospective parents who decline prenatal screening account for their decision? A qualitative study*', (2009) 69 Social science & medicine, 274.

programmes, within the institution, places an unconscious pressure on parents to engage with screening.⁷⁸

Parents reported being swept away by the screening programme, being “easier to engage with screening than resist it”. They felt that, because the narrative surrounding screening underlines its benefits and advantages to both mother and baby, this conveyed the impression that screening was the ‘right’ choice to make. Furthermore, parents reported that an abundance of information was available on the pathway to engage with screening, but a lack of information was available on the alternative pathway to refuse it:

*“Even if I knew I could have said ‘no’ to it, it would have been a step in the dark, because I thought everyone had it. I think it boils down to reassuring mums that they won’t be causing a stir if they do not want the testing, and will still be looked after properly by the NHS. It is so easy to get swept away by it all”.*⁷⁹

This finding may explain the initial theme identified from the quantitative data, that the majority of all parents groups were unaware that they were able to refuse trisomy screening and/or testing, at any time, during the pathway. This is consistent with Tsouroufli, who reported an over-emphasis, by HCPs, on the choice to accept screening, through providing more information on this option, rather than the care pathway to refuse screening.⁸⁰ These academics found that an over-emphasis on the option to accept screening, significantly hinders the ability of parents to consider both options, subject to their values and beliefs, and risks a standardisation of screening programmes.⁸¹

5.4.4 ‘Routine’ Care

All parent groups were under the assumption that trisomy screening was a part of their routine care, and it was not optional. Indeed, a significant majority of participants were unaware that screening was optional even *after* leaving the pathway, or that they could opt to disembark from the pathway, once they had engaged with it:

⁷⁸ Clare Williams, Priscilla Alderson and Bobbie Farsides, ‘*Is nondirectiveness possible within the context of antenatal screening and testing?*’, (2002) 54 Soc Sci Med, 339.

⁷⁹ high-chance, baby with trisomy (DS), England.

⁸⁰ Tsouroufli M, ‘*Routinisation and constraints on informed choice in a one-stop clinic offering first trimester chromosomal antenatal screening for Down’s syndrome*’, (2011) 27 Midwifery, 27, 431, 436.

⁸¹ Ibid.

"I honestly did not know that I had a choice; I thought it was a routine screening test? I did not feel the need question it because doesn't everyone have it? It was not presented as an option, but just part of the care I would get at the beginning".⁸²

This finding may also provide another explanation to the initial theme from the quantitative data, that the majority of all parent groups did not feel it was entirely their 'choice' to undertake screening and/or testing. Research studies by Di Mattei *et al.*, Berg *et al.*, Ngan *et al.* and Reid *et al.*, all outline that parents view screening as a routine and compulsory part of their antenatal care, without having made a real or conscious choice.⁸³ John *et al.*, underlined that only *half* of the midwives, in their study, presented screening as a choice to parents, at the booking appointment.⁸⁴ While trisomy screening intends to promote and empower parents' reproductive choices, these findings may call for a review of the screening programme's design and presentation, as the routinisation and standardisation of screening is at risk of hindering deliberate choice.⁸⁵

5.4.5 Perception of the Trisomies

Parents, who had a baby with a trisomy, strongly expressed that the information, provided by the NHS, portray overly pessimistic, biased and unbalanced generalisations of the trisomies. They all demonstrated concern towards generalisations, which reportedly extend from the medical community, on parent uptake for screening. Of those who had a baby with ES and PS, all reported receiving little, if any, positive information on their child's future, health characteristics and prognosis:

⁸² low-chance, baby without trisomy, England.

⁸³ Valentina Di Mattei, and others, 'Decision-making factors in prenatal testing: A systematic review', (2021) *Health Psychology Open*, 17; see also, van den Berg M, Timmermans D.R.M, ten Kate L.P, van Vugt J.M.G & van der Wal, '*Informed decision making in the context of prenatal screening*', (2006) 63 *Patient Education and Counselling*, 110, 117; and Olivia Miu Yung Ngan and others, 'Parental expectations of raising a child with disability in decision-making for prenatal testing and termination of pregnancy: A mixed methods study', (2020) 103 *Patient Education and Counselling*, 2373; and, Reid B, Sinclair M, Barr O, Dobbs F and Crealey, G, "*A meta-synthesis of pregnant women's decision-making processes with regard to antenatal screening for Down syndrome*", (2009) 69 *Social Science and Medicine*, 1561.

⁸⁴ Sophie John and others, (n2), 12.

⁸⁵ Panagiota Nakou, '*Is routine prenatal screening and testing fundamentally incompatible with a commitment to reproductive choice? Learning from the historical context*', (2021) 24 *Medicine, Health Care and Philosophy*, 73, 83.

“All the information on the trisomies is so overwhelmingly negative. There is a serious imbalance of information on Edwards’ Syndrome. We had to actively look for other cases which showed a more positive outcome; this did not come from the healthcare staff.”⁸⁶

This finding may support the quantitative data, which suggested that parents – particularly those who had a baby with a trisomy – believed HCPs remained ‘unbiased’ across the trisomy pathway. This is supported by Lou *et al.*, who found that the HCPs, in their study, focussed *excessively* on the negative aspects of the condition, and portrayed an overly pessimistic depiction of the conditions, possibly due to a lack of experiential knowledge.⁸⁷ This finding is also consistent with Arthur and Gupta, who found that HCPs demonstrated an over-reliance upon the “dire prognostic data”, in relation to ES and PS, during pre-screening counselling.⁸⁸ However, Tsukada *et al.*, Patterson *et al.*, and Silberberg *et al.*, confirmed that the prognosis of babies with ES and PS have dramatically improved with access to specialist care, and that babies respond well to innovative neonatal treatments.⁸⁹

5.4.6 Intervention

Parents alleged an imbalance of information on available care pathways, post-diagnosis of a trisomy, with a reported ‘abundance’ of information and support on terminating the pregnancy, compared to the limited information and support on continuing with the pregnancy.⁹⁰ They explained that termination was offered as the first choice:

“There is a definitely an imbalance of information, as it currently stands: termination is the first option given to you. There is an abundance of information and support on ending the pregnancy in the booklets, but there was a huge absence of information for continuing with

⁸⁶ high-chance, baby with trisomy (ES), England.

⁸⁷ Lou S, Lanther MR and others, ‘*This is the child we were given: A qualitative study of Danish parents’ experiences of a prenatal Down syndrome diagnosis and their decision to continue the pregnancy*’, (2020) 23 Sex Reprod Healthc, 480; see also, Crombag NM and others, ‘*Receiving the news of Down syndrome in the era of prenatal testing*’, (2020) 182 Am J Med Genet A, 385.

⁸⁸ Joshua D Arthur and Divya Gupta, ‘*You Can Carry the Torch Now: A Qualitative Analysis of Parents’ Experience Caring or a Child with Trisomy 13 or 18*’, (2017) 29 HEC Forum, 223.

⁸⁹ Tsukada K, Imataka G, Suzumura H, and Arisaka O, ‘*Better prognosis in newborns with trisomy 13 who received intensive treatments: a retrospective study of 16 patients*’, (2012) 63 Cell Biochem Biophys, 191; and, Agustin Silberberg and others, ‘*Ethical issues about the paradigm shift in the treatment of children with trisomy 18*’ (2020) 179 Eur J Pediatr, 493; and Jacquelyn Patterson and others, ‘*Transitions in Care for Infants with Trisomy 13 or 18*’, (2017) 34 Am J Perinatol, 887.

⁹⁰ Shelly Haug, Mitchell Goldstein and others, ‘*Using Patient-Centred Care After a Prenatal Diagnosis of Trisomy 18 or Trisomy 13: A Review*’, (2017) 171 JAMA Pediatr, 382; see also, Crombag NM and others, ‘*Receiving the news of Down syndrome in the era of prenatal testing*’, (2020) 182 Am J Med Genet A, 374.

the pregnancy. The impression we got from the information, was a presumption that parents should end the pregnancy, in our situation."⁹¹

Parents, who had a baby with a trisomy, explained that obtaining information on the phenotypic variability of the detected trisomy, is key to inform decision-making. Participants reminded the researcher that the trisomies have three different forms: mosaic, translocation and full. Being in receipt of this information, they noted, is key to informing their decision; whether to continue or end the pregnancy:

"An issue that did come up for us when we had the amnio, it came back from the lab, and the report said, 'there is an observation of Edwards' Syndrome', and that was it. The three different forms of the condition can have different outcomes for the baby. Also, the sex was important because the outcomes; for males, is much worse than females with Edwards'. So, what were the chances of having a stillborn baby? We did not get information on this, but it was crucial for us to make that ultimate – and timely - decisions."⁹²

Crombag *et al.* and Coleman found that the choice to end the pregnancy was disclosed as the *first* choice, by HCPs.⁹³ Indeed, information on ending the pregnancy, in the NHS booklets, currently succeeds introductions to the trisomies.⁹⁴ Previous research has highlighted that attitudes on terminations will vary, depending on the HCP's perception of fetal conditions, and the level of knowledge parents' possess of the condition.⁹⁵ Winn *et al.* observed that, because most HCPs told parents that their baby with ES and PS would die in utero, or during the birthing process, many decided they would opt to end the pregnancy.⁹⁶

A significant concern of parents was an overemphasis on the option to end the pregnancy, following a positive diagnosis of a trisomy, in the paper-based materials, and by HCPs, during counselling. Parents explained that over-repetition by HCPs of the option to end the

⁹¹ high-chance, baby with trisomy (ES), England.

⁹² high-chance, baby with trisomy (ES), England.

⁹³ Crombag NM and others, '*Receiving the news of Down syndrome in the era of prenatal testing*', (2020) 182 *Am J Med Genet A*, 374; and Priscilla Coleman, '*Diagnosis of Fetal Anomaly and the Increased Maternal Psychological Toll Associated with Pregnancy Termination*', (2019) 30 *Issues in Law and Medicine*, 16.

⁹⁴ Both in the FASP and ASW booklets.

⁹⁵ Olivia Miu Yung Ngan, '*Parental expectations of raising a child with disability in decision-making for prenatal testing and termination of pregnancy: A mixed methods study*', (2020) 103 *Patient Education and Counselling*, 2373.

⁹⁶ Phoebe Winn and others, '*Prenatal counselling and parental decision-making following a fetal diagnosis of trisomy 13 or 18*', (2018) 23 *Journal of Perinatology*, 7.

pregnancy, at the follow-up clinical appointments, risks undermining the reproductive autonomy of parents. Parents also reported “losing count” of how many times the option of termination was offered, despite stating, at the first instance, that this was not an option for them. This theme was particularly prominent among those parents who had a baby with ES or PS. A significant minority of parents were offered a termination only weeks before the baby’s due date; this caused significant distress, as they felt their baby was not valued in the eyes of the profession:

“I lost count of how many times I was offered a termination for (names baby). It was at every appointment, and we attended both hospitals every couple of weeks for check-ups and extra scans. I felt her kicking, so abortion was not an option for us. I know they have to double-check, but it felt like our choice was being undermined in the end, and that we did not know what we were doing.”⁹⁷

These findings are consistent with Crombag *et al.* and Haug *et al.*’s studies of parents in this category.⁹⁸ They reported that the option to end the pregnancy was repeated by HCPs, in excess of six times, during post-diagnosis counselling.⁹⁹ Haug *et al.* concluded that over-repetition of the option to end the pregnancy, points to a directive model of care.¹⁰⁰ Asbury explained that parents, who underwent counselling following a diagnosis of a trisomy, frequently felt that the information they obtained was “incomplete or one-sided”, focussing on the negative aspects of the condition, rather than the unknown or positive aspects.¹⁰¹ As Asbury concludes, if parents exit counselling appointments with “inaccurately grim prognoses” for their child, non-directiveness can only be described as aspirational.¹⁰²

Parents revealed they experienced feelings of vulnerability, isolation and irresponsibility, following the decision to continue with the pregnancy, post-diagnosis of a trisomy; this theme was particularly prominent among those parents who had a baby with ES and PS. Parents alleged that such feelings extended from being treated differently, or even ostracised, by the

⁹⁷ high-chance, baby with trisomy (DS), England.

⁹⁸ Crombag NM and others (n93); and Shelly Haug and others, ‘Using Patient-Centred Care After a Prenatal Diagnosis of Trisomy 18 or Trisomy 13: A Review’, (2017) 171 JAMA Pediatr, 382.

⁹⁹ Ibid.

¹⁰⁰ Shelly Haug and others (n90), 382.

¹⁰¹ Bret Asbury (n34), 296.

¹⁰² Ibid.

institution, reporting that the pregnancy becomes medicalised, and the baby becomes a diagnosis:

“I did doubt myself. I wanted to know if it was normal to carry on with the pregnancy, and if other mums had done it successfully. When we found out he had trisomy 13, they (HCPs) started to refer to him as a ‘fetus’, rather than a ‘baby’. On that phone call I had to break the news, we asked what the sex of the baby was, and she (HCP) said, ‘you still want to know the sex?’, as if I was crazy. My baby died 6 hours after he was born, but we cherished those 6 hours we had with him (participant begins crying). Afterwards, the helpline asked me, ‘didn’t you see the bereavement midwife?’, and I said, ‘I didn’t even know there was one?’.”¹⁰³

This finding is supported by Walker *et al.* and Lou *et al.*, who found that parents, in this category, felt ostracised, and sought more information on whether their decision to continue with the pregnancy was the norm.¹⁰⁴ Guon *et al.* and Javier observed subtle modifications in the manner HCPs communicated with the parents conveyed this impression; for example, post-diagnosis, HCPs began referring to the child as a ‘fetus’, rather than ‘the baby’, or by their condition, causing significant distress for parents.¹⁰⁵

These findings may explain the initial theme identified in the quantitative data, that a majority of all parent groups, particularly those who had a baby with a trisomy, felt that HCPs did not remain unbiased, across the trisomy pathway.

5.4.7 Duty to Report an Increased NT Measurement

Parents, who initially *refused* screening, drew attention to the HCP’s overriding duty to report an increased NT measurement, to the requisite fetal medicine unit. They outlined that their decision, to refuse screening, was futile, as their HCPs identified and reported their increased NT measurement to fetal medicine, during the 12-week dating scan. Indeed, parents felt that this duty infringed on their right to reproductive autonomy:

¹⁰³ high-chance, baby with trisomy (PS), England.

¹⁰⁴ Lou S, Lanther MR and others (n87) 102, and Walker LV, Miller VJ, and Dalton VK, “*The health-care experiences of families given the prenatal diagnosis of trisomy 18*”, (2008) 28 J Perinatol, 19.

¹⁰⁵ Guon J and others, ‘*Our children are not a diagnosis: the experience of parents who continue their pregnancy after a prenatal diagnosis of trisomy 13 or 18*’, (2014) 164 Am J Med Genet A, 318; and, Annie Janvier and Andrew Watkins, ‘*Medical interventions for children with trisomy 13 and trisomy 18: what is the value of a short disabled life?*’, (2013) 102 Acta Paediatrica, 1116; see also, Pauline Thiele, ‘*He was my son, not a dying baby*’, (2010) 36 J Med Ethics, 646.

*“I had absolutely no idea they (HCPs) could go over my head and still take a reading, even if I said no to the screening. I learnt afterwards that they (HCPs) have to act on an increased nuchal fold, but no one explained that, at the time. They (fetal medicine) asked me if I wanted my bloods done to complete the test, and I obviously said, ‘yes’; could not leave it like that”.*¹⁰⁶

Ahman *et al.*'s study also underlined that parents felt their autonomy was undermined, expressing they “were shocked by the unexpected and sometimes unwanted information on elevated risk”, suggesting that HCPs take the necessary steps to inform parents this possibility, before performing the scan.¹⁰⁷ This finding points to the battle between informing parents of material risks, and their right to exercise discretion over their reproductive options.

5.4.8 Partners

Participants, who had partners present during screening, outlined that decisions on screening would be made jointly. Parents, in this population, reminded the researcher of the importance of the partner's role for decision-making, viewing the midwife's role as subservient, in this regard:

*“We made decisions about screening together, always. We both had an equal say in the matter because it is both our baby. I would never go ahead with making a decision without consulting (names partner).”*¹⁰⁸

This finding is consistent with Watterbjork *et al.*, who underlined the influential role partners' played during the decision-making process, which was not reflected in the clinical setting.¹⁰⁹ A study by Laberge *et al.* identified that the person whose input was considered *most* important to the pregnant person, was the partner.¹¹⁰

¹⁰⁶ high-chance, baby with trisomy (DS), England.

¹⁰⁷ Annika Ahman, Karin Runestam and Anna Sarkadi, 'Did I really want to know this? Pregnant women's reaction to detection of soft marker during ultrasound screening', (2010) 81 Patient Education and Counselling, 87.

¹⁰⁸ low-chance, baby without trisomy, Wales.

¹⁰⁹ Wätterbjörk, I. and others, 'Decision-making process of prenatal screening described by pregnant women and their partners' (2015) 18 Health Expectations: An International Journal of Public Participation in Health Care & Health Policy, 1582.

¹¹⁰ Anne-Marie Laberge and others, 'Canadian Pregnant Women's Preferences Regarding NIPT for Down Syndrome: The Information They Want, How They Want to Get It, and With Whom They Want to Discuss It', (2019) 41 J Obstet Gynaecol Can, 782.

Despite these findings, partners reported feeling excluded or marginalised from the decision-making process, for trisomy screening:

*“We are not being intentionally excluded by the midwife, but I did feel excluded. You know, it was always asking what (names wife) wanted and how she was coping, rather than dividing the focus fifty-fifty, if that makes sense. Sounds really selfish, I know, but I think decisions like these should be made together. A spare part is the right term to use for us, I think (laughing)”.*¹¹¹

Laberge *et al.* revealed that partners typically feel marginalised during counselling, and they are not valued in the context of decision-making.¹¹² Fenton *et al.* found that the HCPs failed to consider the partners’ contributions or opinions during counselling, causing them to disengage with discussions on decision-making.¹¹³ Watterbjork *et al.* also highlighted that the partners demonstrated a lack of confidence to engage with the discussions on screening, as the focus fell almost solely on the pregnant person.¹¹⁴

¹¹¹ low-chance, baby without trisomy, Wales.

¹¹² Anne-Marie Laberge and others, ‘Canadian Pregnant Women’s Preferences Regarding NIPT for Down Syndrome: The Information They Want, How They Want to Get It, and With Whom They Want to Discuss It’, (2019) 41 J Obstet Gynaecol Can, 782, 784.

¹¹³ SK Fenton, Joscelyne T, Higgins S, ‘Part 1: exploring views from fathers and perinatal practitioners on the inclusion of fathers by perinatal services’, (2021) 29 British Journal of Midwifery 1, 8.

¹¹⁴ Wätterbjörk, I., Blomberg, K., Nilsson, K., & Sahlberg-Blom, E, ‘Decision-making process of prenatal screening described by pregnant women and their partners’, (2015) 18 Health Expectations: An International Journal of Public Participation in Health Care & Health Policy 1582, 1592.

5.5 Relationship and Communication Between HCP and Parent

5.5.1 Parents' Preferred Approach to Communication

During the research interviews, it became evident that parents were divided in their opinion of how they prefer HCPs to approach communication, and the HCP-parent relationship, throughout the trisomy pathway. A majority of parents revealed that they preferred the HCPs to provide information in a 'neutral' manner, distancing themselves from decision-making, in this regard. Indeed, parents explained that HCPs should merely act as a source of information to inform parent decision-making, rather than adopting a shared decision-making model of care; this theme was much stronger among those parents who had a baby with a trisomy:

"They (HCPs) should just give us the information and that is it. Supporting particular decisions can be seen as encouraging decisions. We just need them (HCPs) to pass over the information, and leave us to it".¹¹⁵

Indeed, Carroll *et al.* and Garcia also found that parents' perceived the HCP's role as a neutral information provider, confined to the purpose of communicating the information to parents, allowing them to make the ultimate choice.¹¹⁶ Williams *et al.* noted that HCPs often experience difficulty managing directiveness when communicating with parents, and that the boundary between coercion/directiveness, and supporting choice, was commonly blurred.¹¹⁷ This finding speaks to the preference of a consumerist model of care, as parents feel that the NHS should aim to respond to the needs and preferences of its service-users.¹¹⁸

Parents, who fell into the shared decision-making group, interpreted the HCP's role as advisory in nature, preferring HCPs to communicate in a *directive* manner. They perceived the HCP's education and experience as being invaluable for decision-making, exhibiting high levels of trust towards the profession, in this regard; this theme was much stronger among those parents who were lower-risk:

¹¹⁵ high-chance, baby with ES, England.

¹¹⁶ Fran E Carroll and others, 'A qualitative investigation of the decision-making process of couples considering prenatal screening for Down Syndrome', (2012) 32 *Prenat Diagn*, 57; and, Elisa Garcia and others, 'Rethinking autonomy in the context of prenatal screening decision-making', (2008) 28 *Prenat Diagn* 115.

¹¹⁷ Williams C, Alderson P and Farsides B, 'Is nondirectiveness possible within the context of antenatal screening and testing?' (2002) 54 *Social Science & Medicine*, 339.

¹¹⁸ T. T. Arvind and Aisling M. McMahon, 'Responsiveness and the Role of Rights in Medical Law: Lessons from Montgomery', (2020) 28 *Medical Law Review*, 445.

*“They (HCPs) know far more than we do about all this stuff, because they see so many women on a weekly basis, so of course we need to make the decisions with the midwife, or other staff, who have got specific training in Down’s screening. I was prepared, and actually wanted them (HCPs) to tell me what to do (laughing)”.*¹¹⁹

This is consistent with Ahmed *et al.*, who found that parents typically sought advice from the HCPs, and preferred directiveness when communicating information on screening, to inform their decision-making.¹²⁰ Ende *et al.* concluded that “patient’s preference for decision-making in general are weak”, and that patients did not have a strong desire for sole decision-making responsibility, preferring to leave the final decision into the hands of the HCPs.¹²¹

This research found that, during pressured or stressful situations, parents may feel *compelled* to rely upon the advice of HCPs. Parents, who had a baby with ES and PS, conveyed that, due to the uncharted territory they find themselves in following a higher-risk or positive diagnosis, the imbalance of knowledge between parents and HCPs induces them to trust the recommendations and suggestions of HCPs, during counselling:

*“I was just in floods of tears; I did not understand what was going on. I had never heard of Edwards’ before I got my diagnosis, so I did not have a clue what it meant for me or (names baby). I just had to put all my faith in them (HCPs) to do the right thing for her because I was completely clueless.”*¹²²

This finding ostensibly conflicts with the initial theme from the quantitative data, that among the majority of all parent groups – particularly those who had a baby with a trisomy – they believed that HCPs did not effectively communicate with them, to support decision-making, across the pathway. A degree of reliance is exhibited by parents – particularly those who are high-risk and/or had a baby with a trisomy – which may impact on the dynamic of a desired shared decision-making model of communication. Indeed, Janvier *et al.* found that forty-two percent of parents who had a baby ES or PS “did not want to make – or be part of – life-and-

¹¹⁹ low-chance, baby without trisomy, Wales.

¹²⁰ Ahmed, S and others, ‘*Interpretations of informed choice in antenatal screening: A cross-cultural, Q-methodology study*’, (2012) 74 *Social science & medicine*, 997.

¹²¹ Jack Ende, Lewis Kazis, Arlene Ash, & Mark Moskowitz, ‘*Measuring patients’ desire for autonomy*’ (1989) 4 *Journal of general internal medicine*, 27.

¹²² high-chance, baby with ES, England.

death decisions”.¹²³ This finding may point to a change in dynamic within the professional-parent relationship, due to an imbalance of expertise and knowledge, in this regard.¹²⁴

Parents, in this study, admitted asking unrealistic or unreasonable questions to HCPs which, in their opinion, was indicative of an increase in anxiety, stress or vulnerability, during screening and testing. Parents revealed that they asked, ‘what do you think I should do?’, when faced with decisions on screening and/or testing:

*“Look, when you are in a position like we were, you are so vulnerable. Looking back, I think because I was so stressed, I just expected the neonatologist to answer everything straight away, which is unfair. I asked questions like, ‘do you think I should go for the amnio?’, ‘what will happen if I have a baby with Down’s’, and ‘what would you do if you were me’. I imagine they get that all the time, and I do really feel for them”.*¹²⁵

This finding conflicts with the initial theme identified from the quantitative data, that parents – particularly those who were high-risk and/or had a baby with a trisomy – did not feel HCPs effectively communicated with them to support decision-making. The qualitative findings reveal an admission of unreasonableness, in the context of parent expectation of HCPs’ role to ‘support’ decision-making. This finding may explain the initial theme identified in the quantitative data, that the majority of all parent groups felt barriers to communication were overcome by the HCPs, possibly demonstrating a degree of sympathy toward HCPs. This finding may be explained by the work of Hertig *et al.*, who concluded that questions of this nature are becoming increasingly common, due to the growing complexity of medical science.¹²⁶ Hertig *et al.* and Asbury both explain that parents are becoming increasingly dependent on professional involvement to support decision-making, with one of the most common questions for HCPs being ‘what would you do?’, demonstrating a preference for a directive model of care.¹²⁷ Moller *et al.* also clarified that questions of this nature are

¹²³ Annie Janvier and others, ‘Building trust and improving communication with parents of children with Trisomy 13 and 18: A mixed-methods study’, (2020) 34 Palliat Med, 262.

¹²⁴ Brian G Skotko, “Postnatal diagnosis of Down syndrome: synthesis of the evidence on how best to deliver the news”, (2009) 124 Paediatrics, 751, and Brian Skotko, ‘Mothers of children with Down syndrome reflect on their postnatal support’, (2005) 115 Paediatrics, 64.

¹²⁵ high-chance, baby without trisomy, Wales.

¹²⁶ Solene Gouihers Hertig and others, ‘Doctor, what would you do in my position?’ Health professionals and the decision-making process in pregnancy monitoring’, (2014) 40 J Med Ethics, 310.

¹²⁷ Ibid, see also Bret Asbury (n34), 304.

commonplace among those parents who are experiencing stress and anxiety, when confronted with an unexpected outcome.¹²⁸

5.5.2 Time Pressures

Time limitations were identified as the most obstinate barrier to communication among all parent groups. Parents found that increasing time pressures on HCPs, during clinical practice, mars the HCP-parents relationship, by interfering with the building of rapport and/or the ability to enter into an interactive, meaningful and supportive dialogue. Indeed, parents compared the execution of trisomy screening to a 'conveyor belt' and 'revolving door', underlining the lack of interactivity, in this regard:

"You are in and out like a revolving-door. The appointment lasted 45 minutes, and 2-3 minutes were dedicated to Down's screening; so unrealistic. It all moves along very quickly too, so I think it is the almost factory-type nature of the whole thing which gets in the way of meaningful communication. I did not want to keep her (midwife) from the other women she had, so I did not bother asking about our concerns. She (midwife) must be very busy; we felt incredibly sorry for her when we left".¹²⁹

This finding supports the initial theme from the quantitative data, that parents – particularly those who were high-risk and/or had a baby with a trisomy – did not feel HCPs effectively communicated with them to support decision-making. However, the qualitative data reveals that parents feel a degree of sympathy toward HCPs, demonstrating concern toward the increasing time pressures and constraints HCPs are required to operate, in clinical practice. Consistent with McCourt, this finding underlines the crushing time limitations HCPs experience, in the clinical setting.¹³⁰ John *et al.* revealed that midwives typically spent between *fifteen-seconds* to nine minutes (a mean of two-minutes) counselling parents on trisomy screening, during a one-hour booking appointment.¹³¹ These findings suggest that time constraints, in addition to an apparent lack of interactivity between HCPs and parents,

¹²⁸ Aune, I. and Möller, A., *"I want a choice, but I don't want to decide" – A qualitative study of pregnant women's experiences regarding early ultrasound risk assessment for chromosomal anomalies"*, (2012) 28 Midwifery, 14.

¹²⁹ low-chance, baby without trisomy, Wales.

¹³⁰ McCourt C, *'Supporting choice and control? Communication and interaction between midwives and women at the antenatal booking visit'*, (2006) 62 Social Science & Medicine, 986.

¹³¹ Sophie John and others (n2), 1.

has created a perception of trisomy screening as a revolving-door or conveyor belt of care, resulting to HCPs disclosing the information on trisomy screening in a “run through” fashion.¹³²

5.5.3 Disagreements Between Parents and HCPs

Parents, particularly those who were high-risk and/or had a baby with a trisomy, reported that adversarial interactions with HCP(s) hindered communication, leading to a breakdown in trust and rapport. Adversarial interactions and distrust, between HCPs and parents, was a common theme in the context of provision of counselling and care management on the trisomies; in particular, approaches to care management for ES and PS:

*“She (midwife) was really nice and everything, so we haven’t got any complaints on that front, but it was almost becoming confrontational when I questioned why the figures and information I was getting on Edwards’ from them (NHS), was different to other research I found. I really had to back off and stop asking these questions, because I knew if I pushed it any harder, I knew I was going to upset her (midwife)”.*¹³³

This finding supports the themes identified from the quantitative data, that parents – particularly those who were high-risk and/or had a baby with a trisomy – felt that communication was ineffective with the HCPs for supporting decision-making. This finding may be explained by the study of Janvier *et al.*, who reinforced that the high pressured and emotive nature of trisomy screening, creates tension between HCPs and parents, which can often lead to an interruption or breakdown in rapport.¹³⁴ With the introduction of ES and PS requiring HCPs to communicate highly emotive, and sometimes distressing, information to parents, it may be that CPD include additional specialist training to support HCPs, in this regard. Maintaining honesty and transparency, between HCPs and parents, is crucial for a patient-centred approach to care management.¹³⁵

¹³² McCourt C, (n130), 986.

¹³³ high-chance, baby with trisomy (ES), England.

¹³⁴ Annie Janvier and others, ‘Building trust and improving communication with parents of children with Trisomy 13 and 18: A mixed-methods study’, (2020) 34 Palliat Med, 262.

¹³⁵ Johan Christiaan Bester, ‘Defensive practice is indefensible: how defensive medicine runs counter to the ethical and professional obligations of clinicians’, (2020) 23 Medicine, Health Care and Philosophy, 413.

Adversarial interactions between parents and HCPs also extended from discussions surrounding the decision to withhold treatment, surgical intervention or resuscitation, following the birth of a baby with ES or PS. Parents, in this category, explained that after deliberating with the Trust's ethics committee, they typically received recommendations from HCPs to *withhold* treatment, surgical intervention, or not to resuscitate the baby following the birth, as the health risks meant the baby would not have a quality of life.¹³⁶ Parents revealed that they often disagreed with the opinion of the healthcare team, and felt they needed to 'battle' to preserve the life of their child:

*"I'm getting emotional, I'm so sorry. It is bringing it all back, you know, sitting there asking, 'why won't you resuscitate my baby if he stops breathing when he's born, because the ethical committee are saying he's incompatible with life?'. I kept getting told 'the pregnancy is not viable', but I did not understand what that meant. I mean, you know, I felt my baby was written off from 15-weeks old. I carried my gorgeous boy and delivered him."*¹³⁷

"We were told there was a hole in the heart, so our question was, naturally, would that require surgery? You know, what would our scenario be like if the baby's condition was not too bad after the birth, when we first see it? Is there any community support available to us? How many hospital visits should we anticipate? We both really struggled to picture the outlook for our child; we had to cling onto something. It felt that they put the statistics before the baby."

This is consistent with Siegel and Janvier and Watkins' studies of parents in this category.¹³⁸ Andrews *et al.* explain that there are tensions between two schools of thought regarding caring for a baby with ES or PS: comfort care, or medical and surgical intervention.¹³⁹ Withholding medical and surgical interventions, for ES and PS, were longstanding and conventional approaches; however, this is beginning to change, in the wake of contemporary

¹³⁶ Alaina Pyle and others, 'Management options and prenatal voice in the treatment of trisomy 13 and 18' (2018) 38 *Journal of Perinatology*, 1135.

¹³⁷ high-chance, baby with trisomy (PS), England.

¹³⁸ Linda Siegel, 'When staff and parents disagree: decision making for a baby with trisomy 13', (2006) 73 *Mt Sinai J Med*, 590; and Annie Janvier and Andrew Watkins, 'Medical interventions for children with trisomy 13 and trisomy 18: what is the value of a short disabled life?', (2013) 102 *Acta Paediatrica*, 1112.

¹³⁹ Sasha Andrews and others, 'Shared decision making and the pathways approach in the prenatal and postnatal management of the trisomy 13 and trisomy 18 syndromes', (2016) 172 *American Journal of Medical Genetics*, 257.

medical knowledge and technology.¹⁴⁰ The justification for this rested on “not to prolong or increase suffering, when there is no potential for long-term benefit to the child”.¹⁴¹

Parents recommended that information on stillbirth or miscarriage should be withheld, following a positive diagnosis for ES or PS, if parents choose to continue with the pregnancy. They believed that this information exacerbates the severe stress and anxiety they experienced, after obtaining a positive diagnosis. Parents, in this category, explained that their decision to continue with the pregnancy warrants the non-disclosure, of said information, for therapeutic purposes:

“You just want to cherish that time you have with your baby, at the end of the day, not to be continuously reminded of depressing information and statistics. Like I said before, he lived for six-hours, but that was the happiest six hours of my life. My suggestion would be not to ruin other mums’ time with their baby, if they have Patau’s, by being negative; you only get that time with them once, so we do not need to know how long they (HCPs) think the baby will live for.”¹⁴²

Guon *et al.* and Janvier explained that, once parents decide to continue with the pregnancy – post-diagnosis of ES or PS – HCPs continue to disclose information on the risk of stillbirth and miscarriage, information that parents did not wish to receive.¹⁴³ This finding suggests that *Montgomery’s* therapeutic privilege exception, in these circumstances, may afford parents the time to grieve, and permits HCPs to respect the parent’s remaining time with their baby.

¹⁴⁰ Alaina Pyle and others, ‘Management options and prenatal voice in the treatment of trisomy 13 and 18’, (2018) 38 *Journal of Perinatology*, 1135; and, John Carey, ‘Perspectives on the care and management of infants with trisomy 18 and trisomy 13: striving for balance’, (2012) 24 *Curr Opin Pediatr*, 672.

¹⁴¹ *Ibid.*

¹⁴² high-chance, baby with trisomy (PS), England.

¹⁴³ Guon J, Wilfond BS, Farlow B, Brazg T, and Janvier A, ‘Our children are not a diagnosis: the experience of parents who continue their pregnancy after a prenatal diagnosis of trisomy 13 or 18’, (2014) 164 *Am J Med Genet A*, 308; and, Annie Janvier and Andrew Watkins, ‘Medical interventions for children with trisomy 13 and trisomy 18: what is the value of a short disabled life?’, (2013) 102 *Acta Paediatrica*, 1112.

5.6 Disclosure of Risk and Results

5.6.1 Prevalence of the Trisomies

During the research interviews, parents felt that the risk of ES and PS is trivialised, primarily due to its purported rarity. Parents found that they overlooked the risk of having a baby with ES and PS, due to their recent introduction to the pathway, and their purported rarity:

“I thought my only real risk was Down’s Syndrome; Edwards’ Syndrome never crossed my mind. I was not encouraged to think about the risk for Edwards’ and Patau’s at all. I read afterwards that these conditions are rare, being only one in five-thousand babies, but someone has to be that one (participants begins crying).”¹⁴⁴

This finding could explain the provenance of the initial theme identified from the quantitative data, that parents – particularly those who were high-risk and/or had a baby with a trisomy – did not understand what their risk-score meant, in relation to their trisomy screening result. This finding is consistent with Arthur and Gupta who found that HCPs routinely fail to accurately disclose the risks of ES and PS during counselling.¹⁴⁵ An explanation for this finding may extend to HCPs’ unfamiliarity with the conditions, and a lack of appropriate training.¹⁴⁶

5.6.2 Interpretation of Population Risk

Younger parents, in this study, reported that they were categorised as low-risk before entering screening, believing that *only* older parents were capable of being high-risk for the trisomies. Consequently, younger parents explained they overlooked risk of having a baby with a trisomy, experiencing a significant amount of distress and confusion upon discovering they were high-risk, following screening:

“I am 24, and I did question the midwife about my age and the likelihood of being at risk. She said, ‘don’t worry too much; you’re young and fit’. I had my 12+5-week test (combined), and

¹⁴⁴ high-chance, baby with trisomy (ES), England.

¹⁴⁵ Joshua D Arthur and Divya Gupta, ‘You Can Carry the Torch Now: A Qualitative Analysis of Parents’ Experience Caring or a Child with Trisomy 13 or 18’, (2017) 29 HEC Forum, 223.

¹⁴⁶ Sasha Andrews and others, “Shared decision making and the pathways approach in the prenatal and postnatal management of the trisomy 13 and trisomy 18 syndromes”, (2016) 172 American Journal of Medical Genetics, 257.

*the result came back as high. So, can you imagine how bewildered I was when I got that phone call?”.*¹⁴⁷

Crombag *et al.* underlined that “advanced maternal age and age related risk perception” significantly influence parents engagement in screening.¹⁴⁸ Engels *et al.* found that uptake of screening remains low among younger women, due to an overemphasis by HCPs that younger women are low-risk of having a baby with a trisomy.¹⁴⁹ Interestingly, Asbury observed that HCPs will typically spend two-minutes discussing potential diagnosis of a trisomy with younger parents, compared to seven minutes with older parents.¹⁵⁰

5.6.3 Interpretation of Individual Risk

Parents revealed that they translated a high-risk screening result as being diagnostic, and a low-risk result as the baby does not have a trisomy. They explained they experienced difficulty comprehending the concept of risk/chance in relation to their screening results, and therefore instinctively searched for ‘certainty among the uncertainty’ to rationalise their result:

*“I think my massive worry about the entire process are the words high-risk and low-risk. You take it either as a ‘yes’ or a ‘no’. Your mind naturally translates low-chance as “no”, and high-chance as “yes”; you want to find that certainty in all the uncertainty, you know? Everyone is guilty of it: it is human behaviour to jump to conclusions. You are under so much pressure, so your brain does not think logically when you are engrossed in it all; you want to believe it is yes or no. It is a coping mechanism, I guess.”*¹⁵¹

This finding may explain the initial theme from the quantitative study, that the majority of all parent groups— particularly those who were high-risk and/or had a baby with a trisomy – did not have a clear understanding of ‘risk’, in the context of their screening result. This finding is also supported by Pilnick *et al.*, who underlined that parents interpret their screening result

¹⁴⁷ high-chance, baby without trisomy, Wales.

¹⁴⁸ Crombag NM and others, ‘Determinants of first trimester combined test participation within the central region of the Netherlands’, (2015) 35 Prenat Diagn, 486.

¹⁴⁹ Melanie A J Engels and others, ‘Evaluation of the introduction of the national Down syndrome program in the Netherlands: age-related uptake of prenatal screening and invasive diagnostic testing’, (2014) 174 Eur J Obstet Gynecol Reprod Biol, 59.

¹⁵⁰ Bret Asbury (n34), 301.

¹⁵¹ low-chance, baby with trisomy (DS), England.

as being a *definitive* diagnosis.¹⁵² Heyman *et al.*'s study that found parents interpret a higher-risk screening result as a positive diagnosis, and will treat the pregnancy as such until invasive testing is performed, or following the birth of the child.¹⁵³ Furthermore, Ohman *et al.* underlined that a misinterpretation of risk commonly induces unnecessary anxiety among parents, as their interpretation of 'risk' does not always correlate to the *actual* risk.¹⁵⁴ This misunderstanding may be explained by John *et al.*, who reinforced that in only sixty percent of the booking appointments, the risk statistics were disclosed, with some midwives incorrectly quoting the statistical interpretation or "cut-off" of the categories.¹⁵⁵

Parents explained that a risk exists of being lulled into a false sense of security from false-negative results, or experience unnecessary anxiety from false-positive results. A lower-risk result was understood as being very ambiguous among participants, with comparisons being drawn to a 'lottery', or 'gambling' with the possibility that the baby does not have a trisomy:

*"I was low-risk after the combined screening test, but I had a baby with Down's Syndrome. My risk score was over one in a thousand. The letter I got from my antenatal clinic midwife said (participants pulls out letter), 'The screening test you had for Down's Edwards and Patau's Syndromes showed you have a low chance of your baby having one of these conditions. No further tests are recommended'. After I had (names baby) I just thought, well, what the hell was the bloody point in having the screening in the first place (laughing). It is a lottery at the end of the day; it is a bit of a gamble really."*¹⁵⁶

This is consistent with Dahl *et al.* and Lou *et al.*, who found that parents were unaware of false-positive or negative screening results, experiencing high levels of anxiety upon discovery of this possibility.¹⁵⁷ Due to increasing time pressures in clinical practice, Beulen *et al.* explain that HCPs poorly communicate the possibility of discordant – false-positive or false-negative

¹⁵² Pilnick A.M, Fraser D.M, and James D.K, 'Presenting and discussing nuchal translucency screening for fetal abnormality in the UK', (2004) 20 Midwifery, 82.

¹⁵³ Heyman, B. and others, 'On being at higher risk: A qualitative study of prenatal screening for chromosomal anomalies', (2006) 62 Social Science & Medicine, 2360.

¹⁵⁴ Susanne Georgsson Ohman, Charlotta Grunewald and Ulla Waldenstrom, 'Perception of risk in relation to ultrasound screening for Down's syndrome during pregnancy', (2009) 25 Midwifery, 264.

¹⁵⁵ Sophie John and others (n2), 1.

¹⁵⁶ low-chance, baby with trisomy (DS), Wales.

¹⁵⁷ Katja Dahl, Ulrik Kesmodel, Lone Hvidman and Frede Olesen, 'Informed consent: attitudes, knowledge and information concerning prenatal examinations', (2006) 85 Acta Obstet Gynecol Scand, 1414; and Stina Lou and others, "Does screening for Down's syndrome cause anxiety in pregnant women? A systematic review", (2015) 94 Acta Obstet Gynecol Scand, 15.

– results, often referring parents to information sources that include complex medical jargon or technical medical concepts.¹⁵⁸

5.6.4 Perceptions of Risk

Parents explained that their perception of ‘high-risk’ differed to that of the profession, set by the majority of Trusts in England and Wales at 1:150, or higher. These conflicting definitions of ‘risk’ rested primarily on the parents’ perception of the trisomies, focussing on their nature and subsequent consequences for decision-making. They felt that their perception of risk must take priority over that of the profession, warranting the opportunity to undertake alternative methods of treatment, in the form of further investigative screening, even if they are categorised as ‘low-risk’:

“I felt that my risk of Edwards’ and Patau’s, being one in two-hundred, was not ‘low-risk’. Because of what the condition meant for my baby and my own health, that was high, in our opinion. And because I fell outside of what they (HCPs) consider high-risk, (names baby) condition was only picked up on the 20-week scan, so I missed having the amnio. It’s up to us what we think is high-risk, surely?”¹⁵⁹

These findings are consistent with Asbury, who observed that parents’ understanding of risk is often “less than fully informed”, due to the ambiguity of ‘high-risk’ and the differing perspectives of risk in this regard.¹⁶⁰ Indeed, Ohman *et al.* also explain that the parents’ perception of high-risk will vary, depending on a range of factors which interplay: personal circumstance; values and morals; and perception of the condition and disability.¹⁶¹ These are factors which are often not reflected in the objectively determined, traditional grouping system.

5.6.5 Risk of Invasive Testing

Parents explained that they were unable to cope with their uncertain risk status following screening, revealing that undertaking invasive testing was the only way to relieve the anxiety

¹⁵⁸ Lean Beulen and others, ‘Women’s and healthcare professionals’ preferences for prenatal testing: a discrete choice experiment’, (2015) 35 *Prenat Diagn*, 549.

¹⁵⁹ low-chance, baby with ES, England.

¹⁶⁰ Bret Asbury, (n34), 306.

¹⁶¹ Susanne Georgsson Ohman and others (n154), 264.

and pressure. They also underlined the detrimental impact this uncertain 'risk/chance status' had on their mental health, wellbeing, and the bond between mother and baby:

*"I understood that I did not have to have the amnio, but it's not that simple. We had to think about if the risk of miscarriage was worth knowing if (names baby) had Edwards' (1:251 risk score). We would not have been able to support a baby with complex needs. But you are essentially playing dice with your baby's life – it is so damaging. We had to have the amnio in the end because we could not cope with the uncertainty."*¹⁶²

Chervenak *et al.* and Lou *et al.* warned that parents often feel compelled by anxiety to undertake invasive testing, as they are unable to cope with their uncertain 'risk' status.¹⁶³ Lou *et al.* reported that parents experience high levels of anxiety following a screen positive result, and feel that the only means of relieving anxiety is through undertaking invasive testing, in hope for a "normal" invasive testing result.¹⁶⁴ This finding is significant as it has been proven that parents may treat the pregnancy tentatively, or delay emotional attachment to the baby, until they are reassured, either antenatally (by invasive testing) or postnatally, that their baby does not have a trisomy.¹⁶⁵

5.6.6 'Low' Risk of Miscarriage

A strong theme, among those parents who opted to undertake invasive testing, was that the 'low' risk of invasive testing is overemphasised in the paper-based information materials, and during counselling. Participants explained that this influenced decision-making, and did not reflect the severity of consequence if the risk materialised:

"I think it's important to address the language that healthcare professionals use around the risk of amnio as well: they say its low-risk. They put a strong emphasis on this word 'low'. We wanted statistics. The midwife said, 'it's only around half a percent'. We ask if there is any

¹⁶² high-chance, baby with trisomy (ES), England.

¹⁶³ Frank Chervenak and others, 'Enhancing patient autonomy with risk assessment and invasive diagnosis: an ethical solution to a clinical challenge', (2008) 199 *Am J Obstet Gynecol*, 19 and Stina Lou and others, 'Does screening for Down's syndrome cause anxiety in pregnant women? A systematic review', (2015) 94 *Acta Obstet Gynecol Scand*, 15.

¹⁶⁴ Stina Lou and others, 'Does screening for Down's syndrome cause anxiety in pregnant women? A systematic review', (2015) 94 *Acta Obstet Gynecol Scand*, 15.

¹⁶⁵ Rowe H, Fisher J and Quinlivan, J, 'Women who are well informed about prenatal genetic screening delay emotional attachment to their fetus', (2009) 30 *Journal of psychosomatic obstetrics and gynaecology*, 34, and Stina Lou and others, "Does screening for Down's syndrome cause anxiety in pregnant women? A systematic review", (2015) 94 *Acta Obstet Gynecol Scand*, 15.

*evidence to support that and she crumbled. I know it probably sounds like we were being pedantic, but we weren't happy with just a throw away comment, 'it's around half a percent'; this is my child's life."*¹⁶⁶

This finding may explain the initial theme in the quantitative study, that the majority of all parent groups – particularly those who were high-risk and/or had a baby with a trisomy – did not believe that equal importance was placed on supporting their understanding of both the advantages and disadvantages of the chosen methods of screening and/or testing. This finding is also supported by Cederholm *et al.* and Malkiel *et al.*, who found that the risk of procedural miscarriage is commonly trivialised during pre-testing consultations.¹⁶⁷ These academics explain that overemphasising the 'low' risk of miscarriage may subtly indicate to parents that this is the recommended treatment option.¹⁶⁸ It may also influence parents decision-making in this regard, conveying the impression that refusing testing would be contrary to medical expertise.

5.6.7 Inconsistent Statistics on Risk of Miscarriage

Parents stressed that the statistical risk of invasive testing varied between sources. Participants observed inconsistencies between the information on procedural risk in the materials/packs, and between HCPs during counselling. Parents stressed the importance of maintaining consistency on the risk, considering its severity of outcome:

*"We asked about the risk of miscarriage because the statistics were so inconsistent. They (HCPs) did give answers which could probably be described as satisfactory, but I would say they were still a little bit vague. It changed from one (HCP) to the next. The first midwife we saw said it was 0.3-0.5%, and the doctor said it was 1-2%. The literature fluctuates between a 0.5-2% risk when it comes to miscarriage, so the literature needs to be more consistent."*¹⁶⁹

This is consistent with Durand *et al.*, who found that parents' understanding of risk, in the context of invasive testing, is "particularly misunderstood", often deciding to consent to the

¹⁶⁶ high-chance, baby with trisomy (ES), England.

¹⁶⁷ Cederholm M, Axelsson O, and Sjoden PO, 'Women's knowledge, concerns and psychological reactions before undergoing an invasive procedure for prenatal karyotyping', (1999) 14 *Ultrasound in Obstetrics and Gynaecology*, 267; and Malkiel A and others, 'Can we improve the content and quality of information delivered prior to amniocentesis', (2008) 147 *Harefuah*, 16.

¹⁶⁸ *Ibid.*

¹⁶⁹ high-chance, baby with trisomy (ES), England.

procedure without appreciating the consequences or being able to assimilate complex probabilistic information.¹⁷⁰ These findings may be explained by the work of John *et al.*, who observed that descriptions of invasive testing and the risk of procedural miscarriage was raised in only thirty-three percent of the booking appointments, with inconsistent or incorrect statistics being referenced in relation to the risk.¹⁷¹

¹⁷⁰ Marie-Anne Durand and others, *Information and decision support needs of parents considering amniocentesis: interviews with pregnant women and health professionals*, (2010) 13 *Health Expectations*, 125.

¹⁷¹ Sophie John and others, (n2), 5.

5.7 Non-Invasive Prenatal Testing (NIPT) as an ‘Alternative’ Testing Option

5.7.1 Delivery of Information on NIPT

Of those parents who undertook NIPT screening, many revealed that, following a screen-positive result, the information packs on NIPT were disclosed *on the same day* as consenting to have the test. Participants explained that this did not provide sufficient time and space for them to reflect on their decision, before delivering consent:

“So I’d never heard of NIPT before I got my high-risk. I saw the consultant and the screening midwife and they both went through the information with me, but there was a lot to take in. I had to make a decision in that appointment”.¹⁷²

This finding is consistent with Metcalfe and van Schendel *et al.*, who found that parents consented to having NIPT on the *same day* as being provided with the information on this test.¹⁷³ This finding is concerning as *Thefaut* emphasised that patients must be given adequate time and space to make healthcare choices.¹⁷⁴ Hartwig *et al.* emphasised that the *timing* of counselling on NIPT is crucial to informed decision-making, stipulating that counselling should take place on an occasion prior to obtaining parents’ consent.¹⁷⁵

5.7.2 NIPT’s Purpose

Parents were very unclear as to the purpose of NIPT. Participants explained that NIPT was proffered as an ‘alternative’ option to invasive testing following their higher-risk screening result. As such, parents confirmed that they interpreted NIPT as being able to provide a definitive diagnosis, equivalent to the role of amniocentesis or CVS.¹⁷⁶ During the research interviews, many parents, in this category, explained they felt irked upon realising that NIPT

¹⁷² high-chance, baby with trisomy (DS), Wales.

¹⁷³ Sylvia A Metcalfe, ‘Genetic counselling, patient education, and informed decision-making in the genomic era’, (2018) 23 *Seminars in Fetal & Neonatal Medicine*, 142 and, Rachel V van Schendel and others, ‘What Do Parents of Children with Down Syndrome Think about Non-Invasive Prenatal Testing (NIPT)?’, (2017) 26 *J Genet Couns*, 552.

¹⁷⁴ *Thefaut v Johnson* (2017) EWHC 497 (QB).

¹⁷⁵ Tanja Schlaikjaer Hartwig and others, ‘High risk – What’s next? A survey study on decisional conflict, regret, and satisfaction among high-risk pregnant women making choices about further prenatal testing for aneuploidy’, (2019) 39 *Prenat Diagn*, 635.

¹⁷⁶ Alexandra Cernat and others, ‘Facilitating informed choice about non-invasive prenatal testing (NIPT): a systematic review and qualitative meta-synthesis of women’s experiences’, (2019) 19 *BMC Pregnancy Childbirth*, 27; see also, Elizabeth Alexander, Susan Kelly and Lauren Kerzin-Storror, ‘Non-invasive prenatal testing: UK genetic counselors’ experiences and perspectives’, (2015) 24 *Journal of Genetic Counseling*, 311.

was still only a method of *screening*. They explained that they would have opted to undertake invasive testing earlier in the pregnancy, as a means to prepare for potentially risky/ complex birth, or to make decisions on whether to continue with the pregnancy, had they known NIPT was not diagnostic:

*“I was under the impression that it was the same thing as the amnio, but just safer. It was explained as an ‘alternative’ to the amnio, so clearly this will lead to other mums believing that it works in the same way. When I found out later that I would still need the amnio anyway, I just thought, ‘what was the point?’”*¹⁷⁷

This finding is consistent with the initial theme identified from the quantitative data, that the majority of parents – particularly those who were high-risk and/or had a baby with a trisomy – felt that the information received by HCPs did not support their understanding of the methods of screening and testing. This finding is also supported by Cernat *et al.* and Laberge *et al.*, who observed that parents were dissatisfied with the quality and quantity of information received on NIPT.¹⁷⁸ Without a clear understanding of NIPT’s purpose, this may significantly impede parents’ ability to make an informed decision.

5.7.3 Balancing the Advantages and Disadvantages of NIPT

Parents felt that the procedural safety and ‘high accuracy’ of NIPT were prioritised and overstated in the paper-based/online materials, and during counselling. Consequently, parents explained that they overlooked NIPT’s disadvantages: that it is unable to provide a definitive result, nor does it replace invasive testing. Parents also felt, at the time, the decision *not* to undergo NIPT screening would be perceived as illogical by HCPs, and going against ‘sensible’ medical advice, due to its purported advantages:

*“Well, it is very difficult to think of reasons why not to have NIPT if the consultant is telling you that it is very safe and it is like another simple blood test.”*¹⁷⁹

This finding is supported by the initial themes identified from the quantitative data, that the majority of all parent groups – particularly those who were high-risk and/or had a baby with

¹⁷⁷ high-chance, baby with trisomy (DS), Wales.

¹⁷⁸ Alexandra Cernat, (n176), 27; and Anne-Marie Laberge and others, ‘Canadian Pregnant Women’s Preferences Regarding NIPT for Down Syndrome: The Information They Want, How They Want to Get It, and With Whom They Want to Discuss It’, (2019) 41 J Obstet Gynaecol Can, 782.

¹⁷⁹ high-chance, baby with trisomy (ES), England.

a trisomy – felt that the HCP(s) did not place equal importance on supporting their understanding of the advantages and disadvantages, of the chosen methods of screening and testing. This is also consistent with the work by Steen et al., which points to the consequent risk of influencing parents’ decision to consent to NIPT, by overstating the test’s advantages in this regard.¹⁸⁰ Garcia *et al.* found that there is an increasing pressure on parents to engage with NIPT, as it circumvents historical moral barriers associated with trisomy screening: the risk of invasive testing, and the ambiguous probabilistic nature of combined screening.¹⁸¹ Lo *et al.* found that NIPT’s safety and increased ‘accuracy’ are commonly overstated, and is used as a “selling point” by HCPs during pre-screening consultations, potentially undermining parent choice and decision-making.¹⁸²

5.7.4 Understanding NIPT’s Detection Rate

During the research interviews, parents explained that the phrase ‘99% accurate’ was commonly used by HCPs to describe NIPT’s detection rate. While parents did not consider this to be an issue at the time – believing this to be true – they later discovered that this is not an accurate description of the test’s performance. Indeed, some parents reported that HCPs – primarily midwives – attempted to reassure them by stating that the possibility of discordant results are very rare, and should be treated as being “like the amnio(centesis)”:

*“I was told that it was 99% accurate by both my midwives. The possibility of it being wrong was said to be really rare and very unlikely; how wrong they were with me (laughing)”.*¹⁸³

This finding is supported by several recent studies on NIPT, all underlining concern surrounding HCPs’ descriptions of NIPT’s detection rate.¹⁸⁴ These studies also revealed that phrases such as ‘99% accurate’, ‘more or less diagnostic’, or ‘similar to the amniocentesis’, were commonly used by HCPs when counselling parents on NIPT.¹⁸⁵ Dane *et al.* found that

¹⁸⁰ Sanne L. van der Steen and others, ‘Offering a choice between NIPT and invasive PND in prenatal genetic counselling: the impact of clinician characteristics on patients’ test uptake’, (2019) 27 European Journal of Human Genetics, 235.

¹⁸¹ Elisa Garcia and others, ‘Non-Invasive Prenatal Testing (NIPT) and Pregnant Women’s Views on Good Motherhood: A Qualitative Study’, (2021) European Journal of Human Genetics, 23.

¹⁸² Tsz-Kin Lo and others, “Decision outcomes in women offered noninvasive prenatal test (NIPT) for positive Down screening results” (2017) 32 The Journal of Maternal-Fetal & Neonatal Medicine, 348.

¹⁸³ low-chance, baby with trisomy (DS), England.

¹⁸⁴ Fergus Perry Scott and others, ‘Factors affecting cell-free DNA fetal fraction and the consequences for test accuracy’, (2018) 31 J Matern Fetal Neonatal Med, 1865.

¹⁸⁵ *Ibid.*

parents viewed the information on accuracy as the *most* important consideration before consenting to NIPT.¹⁸⁶ It has been suggested that HCPs commonly refer to NIPT's detection rate in this manner as this was once the belief, based on initial clinical research.¹⁸⁷ *Contemporary* research recommends that counselling on the detection rate of NIPT must include information on specificity, sensitivity, positive and negative predicted values, which are all components of an individualised and true detection rate for the pregnant person.¹⁸⁸

Subsequently, parents explained that were unaware of the possibility of receiving discordant NIPT results, due to the current narrative surrounding its effectiveness at detecting the common trisomies:¹⁸⁹

*"We did not ask about false negatives or anything because we thought that this 'wonder test' would not have false results. I did not question this because I did not know any better at the time. We were not offered the invasive testing because I was low NIPT; we thought we were safe he would not have it (Down's Syndrome), being supposedly 1 in 1,000,000 (laughing)."*¹⁹⁰

These findings supporting the initial theme from the quantitative data, that parents – particularly those who were high-risk and/or had a baby with a trisomy – felt they did not understand what high-risk and/or low-risk meant in relation to their trisomy screening result. The finding clearly indicate the importance of communicating the risk of receiving a false-positive or negative NIPT result.

5.7.5 NIPT's Potential for Increasing Terminations

Parents, who had a baby with a trisomy, stressed that the implementation of NIPT will be highly damaging to the trisomy community, as it will increase the number of terminations

¹⁸⁶ Dane, A. C., Peterson, M., & Miller, Y. D, 'Talking points: Women's information needs for informed decision-making about noninvasive prenatal testing for Down syndrome', (2018) 27 Journal of Genetic Counseling, 1258.

¹⁸⁷ Alexandra Cernat and others, 'Facilitating informed choice about non-invasive prenatal testing (NIPT): a systematic review and qualitative meta-synthesis of women's experiences', (2019) 19 BMC Pregnancy Childbirth, 27.

¹⁸⁸ Fergus Perry Scott and others, "Factors affecting cell-free DNA fetal fraction and the consequences for test accuracy", (2018) 31 J Matern Fetal Neonatal Med, 1865.

¹⁸⁹ Tanja Schlaikjaer Hartwig and others, 'High risk – What's next? A survey study on decisional conflict, regret, and satisfaction among high-risk pregnant women making choices about further prenatal testing for aneuploidy', (2019) 39 Prenat Diagn, 635; see also, Gregory Kellogg and others, 'Attitudes of Mothers of Children with Down Syndrome Towards Noninvasive Prenatal Testing', (2014) 23 Journal of Genetic Counselling, 1.

¹⁹⁰ low-chance, baby with trisomy (DS), England.

being performed following a higher-risk result. They explained that its ability to detect a trisomy earlier in gestation will significantly increase the number of terminations in the future:

*“It’s just a matter of time before we have no DS community. More and more parents will have NIPT because it’s safer than the amniocentesis, so they can just abort earlier in the pregnancy. I think it will be extremely damaging for our community, and for people looking to make informed decisions”.*¹⁹¹

This finding is significant, as it points to feelings of marginalisation and discrimination experienced by the trisomy community, following NIPT’s introduction. However, Leonard opines that parents are confused as to NIPT’s true purpose: as NIPT is deemed more a ‘accurate’ method of screening than the traditional combined screening, its introduction intended to reduce the number of amniocentesis being performed, and thus reducing the number of associated procedural miscarriages, rather than increasing the number of terminations.¹⁹²

5.7.6 Impact of NIPT’s Private Market on NHS

Parents, in this study, drew the researcher’s attention to the potential influence and impact the *private* NIPT screening market could have on the provision of NHS maternity services. Participants explained that many private clinics and pharmaceutical companies advertise NIPT screening online, which entice parents with their discounts, home testing kits, packages, ‘gender reveals’, and other marketing techniques. However, parents revealed that the quality of counselling for NIPT, on the private market, varied significantly, and many found they had to rely on their NHS midwife for further support in this regard. Indeed, parents who undertook NIPT under private care, and subsequently received a higher-risk result, *all* relied primarily on NHS HCPs for support on decision-making. More concerningly, parents also revealed they undertook screening under private care for ‘very rare’ conditions, which they did not recognise, or fully understood:

“We heard about ‘whole panel screening’ from our community midwife: it was really up and coming apparently, all over the internet, too. When we got there (private clinic), they offered

¹⁹¹ high-chance, baby with trisomy, England.

¹⁹² Online resource tool for NHS workers by Emyr Wile, Dr Bryan Beattie and Dr Samantha Leonard.

*us screening for the most obscure sounding conditions. We did screen for them but we had absolutely no idea what they were. I was high-risk after the harmony (NIPT) for Down's Syndrome, but I relied mainly on my midwife for advice after this because the support was non-existent from the (private) clinic."*¹⁹³

This finding is consistent with the predications of the Nuffield Council report, in 2017, highlighting the additional pressure private care could place on the provision of NHS maternity services. The private market offers NIPT screening for the common trisomies – DS, ES and PS – in addition to other genetic conditions, such as DiGeorge Syndrome and Cri-du-chat Syndrome, to name a few. Thus, a risk exists that parents will seek support from NHS HCPs following a high-risk result for a non-trisomy condition, despite not having the requisite training to support parents decision-making in this regard.¹⁹⁴ It may now be incumbent on NHS HCPs to support parents' decision-making for NIPT, not only as a secondary screening test – traditionally for the common trisomies, tested for on the NHS – but also as a primary test, which includes a much broader panel of conditions available under private care.¹⁹⁵ This finding could point to the allure of undertaking NIPT under private care, due to attractive discounts, offers and care packages, and its potential to undermine parents' ability to make informed decisions on trisomy screening.

The allure of the private market was an evident theme among parents in the private sector study. Parents felt that the screening deals were cheap and were readily accessible to them. They also pointed to the fact that they use their NHS midwife as their first port of call:

"For us, it was more about the fact we could screen for everything in a quicky and easy way; and it was cheap considering the offers they have; all in, it must have worked out at around £500 to screen for everything. We obviously use our NHS midwife as our contact point, but the offers were just there to be taken". ¹⁹⁶

¹⁹³ high-chance, baby with trisomy, England.

¹⁹⁴ Nuffield Council 2017.

¹⁹⁵ Dane, A. C., Peterson, M., & Miller, Y. D, 'Talking points: Women's information needs for informed decision-making about noninvasive prenatal testing for Down syndrome', (2018) 27 Journal of Genetic Counseling, 1258.

¹⁹⁶ Low-chance, baby without trisomy, Wales (private clinic study).

5.8 Concluding Observations and Comments from the Parent Qualitative Findings

The aim of this qualitative stage, under this explanatory research design, was to further explore the themes identified from the quantitative data, and also uncover other themes relating to the experience of parents, and the key areas of interest.

In relation to the key areas of interest, the qualitative data either confirmed or ostensibly rebutted the initial themes uncovered in the quantitative dataset, providing the researcher with a rich contextual understanding of the provenance of the themes at each stage of the trisomy pathway.

The qualitative findings also revealed a range of sub-themes; this allows for a further contextual insight into why the initial patterns of behaviour emerged in the quantitative data. It also provides the researcher with a foundation for further discussion of the identified themes and sub-themes in chapter 7, pertaining to parent experience of decision-making and consent, and whether it could be further supported to effectively cater to the interests of parents along the trisomy pathway.

Chapter 6 – HCP Groups Qualitative Findings

6.1 Justifying the Use of Qualitative Interviews and Surveys To Frame an Empirical Response

The purpose of this chapter is to present the findings from the HCPs qualitative study, as a second phase to the explanatory research paradigm. Following the initial themes identified from the HCP quantitative survey study (see chapter 4 ‘concluding comments on HCP study’), it was necessary for the researcher to further explore the experiences of HCPs operating under current systems of consent for trisomy screening. Similarly to the parent study, this qualitative stage was significant to compare and/or integrate the data collected, confirming or refuting the initial quantitative observations.

This chapter will map the themes identified from the HCP open-survey and interviews, expanding on the initial themes identified from the quantitative dataset. As with the parent qualitative findings, themes overlapped, demonstrating interesting relationships and phenomenological interactions between themes and key areas of interest. However, the researcher sought to distinguish (as far as possible) between the themes, and sub-themes, to provide clarity for the reader.

While the researcher obtained an abundance of rich qualitative data, quotations were carefully selected to best represent key considerations and areas of interest, within each identified theme. The findings present HCPs’ account of securing informed consent for screening, in clinical practice. In light of the considerations identified from *Montgomery* and *Mordel*, compounded by the initial quantitative themes, the primary focus of this chapter was on the system(s) of consent, and the steps HCPs are required to take to secure it. The aim was also to uncover any additional themes or areas of interest, providing the researcher with a rich contextual understanding and insight in this regard.

6.2 Securing Consent for Trisomy Screening

6.2.1 Ambiguity Between Professional Roles to Secure Consent

Following the decision in *Mordel*, midwives questioned their role to secure consent for trisomy screening at the booking appointment. They expressed that if sonographers are required to counsel parents and secure their consent on the day of the scan – consistent with the expectation in *Mordel* – their role becomes ambiguous in this regard. Midwives underlined that, in their opinion, the midwife and sonographer, in *Mordel*, “did nothing out of the ordinary”. Thus, they felt that they are currently scapegoats, or villainised, for broader systemic failings:

“We had a training day on the Mordel case and, to us (midwives), it seems that our role is really unclear for getting the mum’s consent, because this was always our job. Counselling mums for them to only be re-counselled by the sonographer, when they get their NT done, does not make sense. From our perspective, we are just scapegoats for broader problems, and it is actually quite insulting.”¹

Sonographers also demonstrated concern towards their duty to obtain parent consent. They explained that the extension of their duty, in *Mordel*, to play a leading role in securing consent, is not established practice. Sonographers expressed their fear of complaints and litigation in this regard:

“We (sonographers) are very concerned about how we formally get the mum’s consent. It’s the midwife’s job to get consent at the booking appointment, then we make a note of it on our records before the NT scan. This is very worrying and has left me feeling pretty uneasy: it is almost like they’re waiting for us to trip up”.²

This finding is consistent with the quantitative study, that all HCP groups – particularly sonographers and midwives – did not believe appropriate systems are in place to secure parents’ consent, nor they feel that they have received up to date training on securing consent for trisomy screening, since its implementation. This finding is also significant as it highlights

¹ Midwife, Wales – open survey response.

² Sonographer, England – interview response.

the ambiguity surrounding HCP duty to secure parent consent, supporting the quantitative data, that HCP groups – particularly sonographers and midwives – were not clear on their roles for securing parent consent. Midwives’ feelings of being villainised or scapegoated is also concerning, creating significant friction between the law and medical profession.

6.2.2 Overwhelmed by Clinical Guidelines on Consent

Midwives and sonographers both drew attention to the overwhelming amount of clinical guidelines on consent for trisomy screening. They both identified a lack of cohesion and consistency between the guidelines. Both professions feel that guidelines need to be concentrated into a single document for clarity and simplicity:

“There are so many different guidelines we need to follow, it is ridiculous: FASP, NICE, NMC, GMC; the list goes on. We need to have just one set of guidelines to enable us to follow them easily, especially now we are seeing more and more studies saying that consent is not being properly provided”.

This finding is consistent with Kater-Kuipers *et al.*, who outlined the lack of consistency between existing guidelines on trisomy screening, and has been “notoriously poor”.³ In light of the decision in *Mordel*, it may be that guidelines for both midwives and sonographers are aligned to ensure consistency of provision in this regard.

6.2.3 Adapting Systems for Securing Consent During COVID

Midwives explained that, during the pandemic, the established systems for securing consent had to be drastically modified. Booking appointments were taking place on the telephone, and many parents were unable to attend their 12-week appointments due to lockdown. Midwives outlined that the system for securing consent had to be constructed via telecommunication and online platforms. Despite being stressful for both HCPs and parents, midwives drew attention to the benefits of adapting established systems for securing consent to telecommunication/online means:

“It was really stressful for everyone; mum’s could not attend their 12-week appointments and we had to hold booking appointments on the phone. But COVID did show us how quickly we

³ Adriana Kater-Kuipers and others, ‘Rethinking counselling in prenatal screening: an ethical analysis of informed consent in the context of non-invasive prenatal testing (NIPT)’, (2020) 34 *Bioethics*, 671-678.

can adapt the way we manage care, and how we can still work efficiently remotely. Consent for the screening was a clear worry for us; but in many ways, it was much more effective, because it encouraged us to work closely with sonographers.”⁴

Sonographers also outlined the benefits of using telecommunication methods for securing consent. They found that interprofessional relationships were enhanced as a response to maintaining professional standards during the pandemic, and to effectively perform a complete overhaul of established systems for securing parent consent:

“We can all take away the positives from COVID and how we managed to adapt so quickly. I think it is also testament to how closely we worked together, to make sure that mum’s were making the right decisions for them, and that the decisions were properly recorded”.⁵

This finding underlines the capability of maternity units to adapt established systems for consent, if required. Consistent with Power *et al.*, it also reveals how the pandemic encouraged the promotion of interprofessional practice to ensure that consent was sufficiently obtained for trisomy screening.⁶

⁴ Midwife, Wales – interview response.

⁵ Sonographer, Wales – open survey response.

⁶ Alison Power and others, ‘Academics’ experiences of online interprofessional education in response to COVID-19’, (2021) 30 British Journal of Midwifery 1, 6.

6.3 Provision of Information

6.3.1 Ambiguity Between Professional Roles on the Provision of Information

All sonographers stressed that their duty does not extend to providing parents with information, as this is executed by the midwife at the booking appointment/first visit. Thus, they were unable to expand on the research questions in this regard, due to their unfamiliarity with the information materials on trisomy screening, provided to parents:

“That’s the midwife’s job, not our job: we have not got time for that. We do not give them any new information that has not already been covered with their midwife. We just confirm one way or the other if they still want to go ahead with the scan”.⁷

Thus, it is unsurprising that sonographers conveyed concern toward the requirement, in *Mordel*, to re-counsel parents on the information during the examination appointment. Sonographers explained that they simply do not have sufficient time during the examination appointment to fulfil this duty. They also opined that this requirement underlined the court’s lack of understanding and naivety of the demands of clinical practice:

“We have not been expected to counsel the parents on the information before now; that was the midwife’s job. I struggle to see how we can spend the time needed to go back over the information their midwife covered with them (parents), and have the time to perform the scan. We typically have twenty-minutes to perform the scan, so that is not a lot of time anyway. It seems courts don’t have a clue how things actually work.”⁸

Sonographers expressed that it is rare for parents to ask for more information during the examination appointment. However, they drew attention to the unfeasibility of the requirement, in *Mordel*, to send the parents back to the midwife, preferably on the same day, if they do want more information. Sonographers also identified that the functionality of this system is dependent on shift patterns:

⁷ Sonographer, England – open survey response.

⁸ Sonographer, Wales – interview response.

*“I do not really get asked for more information at the NT appointment, but that case said to send them back to the midwife if they want more information. How are we expected to send them away, wait for them to talk to the midwife – if they’re lucky enough to grab hold of one during their shift – and then perform the scan on the same day? The reality is the patient would miss their scan and would need to reschedule it for a different day and time. They could miss their chance to have the combined screening completely depending on their gestation”.*⁹

These findings support the initial theme from the quantitative study, that the majority of HCPs – particularly midwives and sonographers – were not confident supporting parents’ informational needs on trisomy screening. These findings also raise concerns surrounding the ambiguity between professional roles on the provision of information for trisomy screening. It also identifies possible frailties under existing systems for the provision of information. This begs the question whether the training provided to sonographers should be brought in-line to those of midwives, as a means to achieve the desired consistency of provision.

6.3.2 Encouraging Parents’ to Read the Information Packs

A very strong theme among midwives was that parents frequently attend the booking appointment without having read the information booklets and packs. A plethora of reasons were reported for why the information had not been read: parents commonly do not realise they were supposed to read the information; they throw the information away; they do not feel the need to read the information (usually parents with previous experience of screening); they demonstrate a lack of awareness for the information; they prefer face-to-face delivery, or in some cases, they do not receive the information:

*“This study needs to highlight the issue of getting parents to read the packs, because it is part of my job that I find extremely exhausting. Informed consent is all about being informed before they make a decision. If they have not read the packs, they have not got enough time to be ‘informed’ at the appointment, because we have too much to cover.”*¹⁰

⁹ Sonographer, England – open survey response.

¹⁰ Midwife, England – open survey response.

This is consistent with the survey performed by Sadler, finding that encouraging parents to read information packs is usually futile, and that reverting to a generalised approach to information provision is often necessary.¹¹

6.3.3 Concern for Use of Online Resources

Midwives, in this research, were unclear of how to discharge their duty in situations where the relevant information has been provided, but the parents prefer to access online sources of information, of varying quality and reliability. Midwives found that a growing number of parents admittedly supplement the NHS materials – particularly during the pandemic – for pregnancy applications, chat rooms, websites and social media platforms, to inform themselves on trisomy screening:

“A huge challenge for us is getting them (parents) to understand that not everything on the internet is true. I have had all sorts of bizarre questions about trisomy testing which were located on various websites and apps, more so with the pandemic. I find parents much rather use apps and other internet sources to get information than the materials we give them.”¹²

This is consistent with the facts of *Mordel*, with the Claimant preferring to watch ‘Youtube’ videos, rather than reading the information packs the NHS provided, before attending the clinic.¹³ While the popularity of online sources understandably increased during the pandemic, the quality and validity of such sources must be brought into question, as this may interfere with parent decision-making.

6.3.4 Provision of Information on the Trisomies

The challenges supporting parents’ informational needs on the trisomies, emerged as a prominent theme among midwives, particularly in relation to ES and PS. They stressed that the focus of parents’ attention typically falls to DS, as this is a ‘familiar’ condition. Thus, midwives outlined that, due to the recent introduction of ES and PS, additional steps must be taken during the early stages of gestation, to highlight the information on ES and PS:

¹¹ Michael Sadler, ‘Serum screening for Down’s syndrome: how much do health professionals know?’ (1997) 104 *An International Journal of Obstetrics & Gynaecology*, 176.

¹² Midwife, England – open survey response.

¹³ *Mordel v Royal Berkshire NHS Trust* (2019) EWHC 2591 (QB), 36.

“I can see why parents are not aware of Edwards’ and Patau’s Syndrome. They have been recently introduced to our screening services and they are really rare; so not something that would have crossed parents’ minds. But more could be done with the information to raise awareness for them (ES and PS) before coming to us in clinic.”¹⁴

These findings are consistent with the parent findings, highlighting that the information on ES and PS are commonly overlooked. This finding begs the question whether additional measures could be implemented to draw parents’ attention to the recent modifications to the trisomy pathway, as a means increase parents’ awareness in this regard. Highlighting the difference between the traditional ‘DS pathway’ and the trisomy pathway, could provide the desired clarity.

Midwives conveyed they could confidently deliver information on DS, but lacked confidence delivering information on ES and PS. This reported lack of confidence was due to the recent introduction of ES and PS to the pathway, compounded by their inexperience of personally caring for babies with these conditions. Suggestions were made by midwives that resources, including information packs, videos or aids, could be generated by those families *with* experience. They explained that this would not only support the parents’ information needs, but also that of the profession:

“Most healthcare staff will be very familiar with Down’s and could give a really coherent account of it. Edwards’ and Patau’s are very different because they are so rare, and very few healthcare staff would have experience caring for babies with them. I think it would be great if we could have more booklets made by those with experience of having an Edwards’ or Patau’s pregnancy; I have never had formal training on them” .¹⁵

This finding may explain the initial theme from the quantitative data, that the majority of HCP groups – particularly sonographers and midwives – did not feel confident supporting parents’ informational needs on trisomy screening, nor felt confident supporting their understanding of trisomy screening. This theme may also be explained by the quantitative data, which revealed that the majority of all HCP groups – particularly sonographers and midwives – did

¹⁴ Midwife, England – interview response.

¹⁵ Midwife, Wales – interview response.

not feel they received up to date training on supporting parent understanding of the trisomies, since the implementation of the pathway.

These findings support the suggestions made by those parents – particularly those who had a baby with a trisomy – to utilise their own personal experience of having a baby with ES, PS or DS to inform practice. Indeed, the Nuffield Council report found that “healthcare professionals involved in screening can have difficulty in communicating information about Down’s, Edwards’ and Patau’s Syndromes”.¹⁶ A survey-study by Janvier *et al.* concluded that those HCPs who were in most direct contact with families living with children who have ES and PS, acquired the most in-depth, comprehensive knowledge of the trisomies.¹⁷ Thus, it may be necessary to facilitate patient evaluation groups or workshops, as part of midwives’ CPD, to gather contemporary information about the trisomies to inform practice.¹⁸

6.3.5 Process of Tailoring/Individualising the Information

Tailoring the information, subject to the parent’s needs, emerged as a prominent challenge among midwives. They explained that pressing time constraints, and the demand to review an increasing amount of information, means that building rapport with parents is becoming increasingly challenging. Midwives stressed that, while clinical guidelines have been modified subject to the inclusion of ES/PS and NIPT to the pathway, they have not been altered in terms of how midwives review this information within the allocated clinical appointments, and tailor said information to the needs of parents:

“We are expected to make a good assessment of their (parents) needs to individualise the information, but we have not got any guidelines on how to do this. We only have forty-minutes with each mum; that is not enough time, so we need guidance on how to meet this expectation under our time limits. Time is only getting tighter, too.”¹⁹

¹⁶ Nuffield Council Report (2017), 44.

¹⁷ Annie Janvier and others, ‘Health care professionals’ attitudes about pregnancy termination for different fetal anomalies’, (2012) 17 Paediatrics & Child Health, 86, 88.

¹⁸ Celine Lewis, Melissa Hill and Lyn S. Chitty, ‘A qualitative study looking at informed choice in the context of non-invasive prenatal testing for aneuploidy’, (2016) 36 Prenatal Diagnosis, 875 and, Skirton H, O’Connor A and Humphreys A, ‘Nurses’ competence in genetics: A mixed method systematic review’, (2012) 68 Journal of Advanced Nursing, 2387. SOFT families have been heavily involved in various ways to help train NHS staff and raise their awareness of Edwards’ and Patau’s Syndrome.

¹⁹ Midwife, Wales – open survey response.

This finding is supported by Tschudin *et al.* and Munthe, who observed that time constraints hinder HCPs' ability to effectively tailor the information to the needs of parents, as they are unable to build rapport or a meaningful dialogue in this regard.²⁰ Jackson illustrates this point by referring to the GMC survey of HCPs, concluding that HCPs felt personalising information and individualising conversation requires NHS time, which they simply do not have.²¹

6.3.6 Suggestion of Defensive Practices

Midwives acknowledged that there is an “unrealistic” amount of information parents are required to explore and reflect upon, before providing their consent for screening. However, they expressed that the pressures from professional regulation bodies, and the fear of litigation, results to parents being overwhelmed with information. Midwives asserted that overwhelming parents with information is “collateral damage” to broader systemic failings:

“It is unreasonable to expect women to take in all that information. But if I think the mum is really struggling with all the information she is getting, and I leave out bits of information for later on, I could get into a lot of trouble. I do know that mums get overwhelmed with all the information, but we need guidance here on how to manage this.”²²

Sonographers also questioned how far they could rely upon the information provided to parents by midwives at earlier appointments, in terms of discharging their duty in this regard. Sonographers expressed that there is a foreseeable risk of presenting the same information to parents as the midwife had from earlier appointments, which would be illogical under growing time constraints. They suggested that clinical guidelines must outline their approach to counselling – whether a review the information from the booking appointment selectively or in its entirety is required – to avoid repetitiveness, timewasting and overloading the parents with information:

“Without telling us what we need to cover, we will just go back over the same information that the midwife covered with them (parents). With the amount of time pressures we are

²⁰ Tschudin S and others, ‘Pregnant women’s assessment and level of knowledge of prenatal counselling’, (2009) 30 *Ultraschall Med* 157; and Christian Munthe, ‘A new ethical landscape of prenatal testing: individualizing choice to serve autonomy and promote public health: a radical proposal’, (2015) 29 *Bioethics*, 36.

²¹ Emily Jackson, ‘Challenging the Comparison in Montgomery Between Patients and ‘Consumers Exercising Choices’, (2021) 00 *Medical Law Review*, 6.

²² Midwife, Wales – open survey response.

*under, this is definitely very stressful and extremely timewasting. We need to be told what aspects of counselling we need to cover, to avoid repeating what the midwife has already said, and avoid running the risk of overloading them with information".*²³

This finding is supported by Hertig *et al.* and Robertson *et al.*, who revealed that while HCPs acknowledge there is currently too much information for parents to reasonably be expected to absorb, they are not confident omitting or summarising the information, subject to the increasing scrutiny from professional bodies and fear of clinical negligence claims.²⁴ Consistent with Mattei *et al.*, these findings may suggest that HCPs adopt defensive practices to care to protect their own interests.²⁵

²³ Sonographer, Wales – interview response.

²⁴ Solene Gouihers Hertig and others, 'Doctor, what would you do in my position?' Health professionals and the decision-making process in pregnancy monitoring', (2014) 40 *J Med Ethics*, 310; and Judith Robertson and Ann M. Thomson, 'A phenomenological study of the effects of clinical negligence litigation on midwives in England: The personal perspective', (2014) 30 *Midwifery*, 121.

²⁵ Valentina Di Mattei, Federica Ferrari and others, 'Decision-making factors in prenatal testing: A systematic review', (2021) 2 *Health Psychology Open*, 1, 20.

6.4 Supporting Parent Understanding

6.4.1 Ambiguity Between Professional Roles on Supporting Parent Understanding

Sonographers expressed that their duty does not extend to ensuring that parents understand the information provided by the midwife. They explained that they have not been trained to counsel parents, and that they are under considerable time pressures, which restrict them from exploring parent understanding. Sonographers explained that, traditionally, their duty does not extend to confirming whether the parents understood the information, but rather their decision to accept or refuse trisomy screening, from the booking appointment, is forthcoming on the day of the ultrasound examination:

“We are technicians by definition; that is our role. We have to check that they still wanted their NT measurement taken, but that is the extent of our ‘counselling’; it does not go to checking understanding. I understand this is now different, but we feel at a loss because we have not received the proper training on counselling.”²⁶

Sonographers, in this study, also sought clarification on the requisite steps they must take if they discover the parents require further counselling to support their understanding, before obtaining consent. Sonographers asserted that many maternity units are stretched for time and resources; thus the provision of further counselling, on the day of the examination appointment, is unrealistic:

“I am unclear what we need to do if it is clear they (parents) need more support. This probably means the midwife needs to be in the room with us when we do the scan, just in case. If we followed what they said in that recent case (Mordel), everything would come to a grinding halt in these units.”²⁷

These findings speak to a number of the initial themes identified in the quantitative study: that the majority of HCP groups – particularly sonographers and midwives – do not have a clear understanding of their role; they are not confident supporting parents informational needs on trisomy screening; and they are not confident supporting parent understanding of

²⁶ Sonographer, England – open survey response.

²⁷ Sonographer, England – open survey response.

trisomy screening. This finding may also speak to the initial theme that the majority of HCPs – particularly sonographers and midwives – do not feel they have received up to date training on the trisomies since the implementation of the pathway. This interaction between themes is significant, as it demonstrates a relationship between the key areas of consideration, pertaining to HCP interests for securing consent.

These findings may also suggest that the law is out of touch with the realities of clinical practice within maternity units. Again, we return to the question of whether it is necessary to turn to *Bolam* – with the gloss of *Bolitho* – to provide the judiciary with the expertise necessary to conclude on matters of clinical practice.

6.4.2 Importance of Experiential Knowledge to Support Parent Understanding of the Trisomies

Midwives demonstrated concern towards the “imbalanced and inaccurate” information used to support parent understanding of the health implications and prognosis of the trisomies. They explained that a lack of professional experiential knowledge of the conditions, compounded with parents’ preference for online information, hinders parent understanding, particularly for ES and PS. Midwives explained that e-learning modules and training days (usually provided by SOFT) are currently available; however, completion of these modules is *not* compulsory:

“I am not confident that we are giving women an accurate and balanced account of what it would be like to have a baby with the trisomies. I went to the training day SOFT put on which got mothers in to talk about their experience of having a baby with them (ES and PS), and how we should approach it as midwives, which was really eye-opening and amazing.”²⁸

This finding is consistent with the initial theme from the HCP quantitative data, that the majority of all HCP groups do not feel that they have received up to date training on supporting parent understanding of the trisomies, since the implementation of the pathway. This finding is significant, as it underlines the risk unbalanced and inaccurate information could have on parent understanding. It may also speak to the risk of information bias. This

²⁸ Midwife, England – open survey response.

underlines the importance of CPD to support the HCPs understanding of the conditions and available care pathways for parents.²⁹

6.4.3 'Trisomy' Model and Parent Misunderstandings

Midwives expressed that a predominant challenge, in practice, is ensuring that parents understand that while 'trisomy' screening will screen for DS, as well as ES and PS, these conditions are *not* the same in terms of health characteristics and prognosis. Midwives identified that presenting the programme as 'trisomy' screening currently creates this impression, and it risks misleading parents in this regard. They also explained that it is commonplace for parents to feel disgruntled and "taken by surprise" upon discovering their risk-score for ES and PS, as they were *unaware* these conditions were included in the programme:

"The challenge here is getting parents to understand that the screening will also look for trisomy 18 and trisomy 13, and these are not the same thing as Down's Syndrome; they (ES and PS) are deadly. I can see why parents think they are all the same because the name 'trisomy' screening will give off the impression that the conditions are all the same when they are very different from each other".³⁰

This finding could explain the initial theme from the quantitative data, that the majority of HCPs revealed they did not feel confident supporting parents' choices along the trisomy pathway. This finding is also consistent with the parents' findings, which identified that presentation of the 'trisomy' programme risks misleading parents, in this regard. It may be that a fragmentation of the pathway is warranted, to enable bespoke consent to be delivered by parents on *all three* trisomies.

6.4.4 Parents' Focus on Invasive Testing

Supporting parents' understanding of the difference between combined screening and invasive testing, emerged as a prominent challenge, among midwives. They expressed that it

²⁹ Celine Lewis, Melissa Hill and Lyn S. Chitty, 'A qualitative study looking at informed choice in the context of non-invasive prenatal testing for aneuploidy', (2016) 36 *Prenatal Diagnosis*, 875, and Skirton H, O'Connor A and Humphreys A, 'Nurses' competence in genetics: A mixed method systematic review', (2012) 68 *Journal of Advanced Nursing*, 2387.

³⁰ Midwife, England – open survey response.

is common for parents to focus on the invasive methods of testing, as these carry a physical risk of miscarriage. Thus, parents enter the pathway “entirely focussed” on the invasive methods of testing, which are only used if the parents are indeed higher-risk:

*“It is really challenging for us to get parents to understand that combined screening is just a scan and a blood test. We always get mums coming in terrified of the amnio, and think that they are going to have that, rather than the combined test. I constantly remind my girls that we only use the amnio if they are high-risk from the combined”.*³¹

This is consistent with the parent findings, underlining that many parents are fearful of the invasive methods of testing, and may overlook the combined screening as a result.

6.4.5 Parent Naivety of Screening’s Consequences

In this research, sonographers highlighted the challenges they experience supporting parents’ understanding of the consequences of combined screening. Sonographers explained that parents often do not consider the reality of scanning, and the potential consequences following the examination, treating the ultrasound examination appointment “like a day out”, or an opportunity to find out the sex:

*“I try my best to enter into a conversation with the parents during their scan. They are usually really excited to see the baby properly for the first time and it is all one big rush. I feel I have to enter into that conversation, because they tend to forget the seriousness of the scan; all they want to do is see the baby and know if it is a boy or a girl.”*³²

These findings are supported by Sholapurkar *et al.*’s study of sonographers in antenatal care.³³ Sonek and Oztekin underline that sonographers must seek to maintain *consistency* of approach and performance quality, in the context of first-trimester ultrasound screening, to ensure that parents understand the implications of the ultrasound scan for the purposes of combined screening, before securing consent.³⁴

³¹ Midwife, Wales – interview response.

³² Sonographer, England – open survey response.

³³ Shashikant L Sholapurkar, Stephen O’Brien and Joanne Ficquet, ‘Use of ultrasound in the antenatal space’, (2021) 29 British Journal of Midwifery, 6.

³⁴ Jiri Sonek, ‘First trimester ultrasonography in screening and detection of fetal anomalies’, (2007) 145 Am J Med Genet C Semin Med Genet, 45, 61 and Ozgur Oztekin, ‘First trimester ultrasound: current approaches and practical pitfalls’, (2009) 36 J Med Ultrason, 161.

6.4.6 Conflating Combined Screening and Invasive Testing

From the experience of sonographers, many parents enter the examination appointment unaware of the difference between screening (combined) and invasive testing (amniocentesis/CVS). Sonographers explained parents frequently attend the ultrasound examination appointment under the assumption of having invasive testing, specifically amniocentesis. They stressed that this is concerning, as parents have consented to screening with a fundamental misunderstanding of its practicalities:

*“My concern is the amount of mums who come to see me thinking they are about to have the amnio. I just wonder how have mums have reached this stage without being corrected of this mistake. This is a big concern for sonographers”.*³⁵

This finding is consistent with the expert witness statement in *Mordel*, which underlined this is a common misunderstanding in clinical practice. The finding is concerning as the practicalities and function of screening and testing are entirely different, rendering consent obsolete in such circumstances.

6.4.7 Use of Medical Jargon

Midwives recognised that medical jargon could act as a significant barrier to communication. They explained that medical jargon is often used in the paper-based materials, but they attempt to clarify said terminology when counselling parents at the various appointments. Midwives revealed that, on occasion, it is “second nature” or a “force of habit” to use medical terminology with parents:

*“Using medical terms is only natural in this job. But it becomes an issue when the woman does not understand what you are explaining. We need to remember that terms we are familiar with, the average person on the street wouldn’t have a clue.”*³⁶

This finding is consistent with John *et al.* who found that use of medical jargon could significantly impede understanding and communication. Use of complex medical terminology

³⁵ Sonographer, England – open survey response.

³⁶ Midwife, England – interview response.

- that impedes understanding - is commonplace along screening pathways, according to Winter.³⁷

6.5 Parent Decision-Making and Choice

6.5.1 Ambiguity Between Professional Roles on Supporting Parent Decision-Making and Choice

Sonographers questioned their role in terms of whether they are expected to revisit the parent's choice to screen and, if so, how is this to be exercised. They explained their duty does not extend to "double-checking" the parent's choice, as this may undermine their decision following the midwife's booking appointment. Sonographers also confirmed that their role currently requires them to check that the parent's decision from the booking appointment is forthcoming, rather than to provide further counselling on the matter:

"It is not for us (sonographer) to go into why they have made a choice. They give consent with the midwife and we have to record it when they come to us before their NT scan. We have not been trained to go into why they have made a decision: that was always the midwife's job, until now".³⁸

In this study, sonographers were concerned about the execution of their duty in circumstances where parents exhibit a change of mind between the booking appointment, with the midwife, and the day of the examination. Sonographers explained that a change of mind is very common in practice; however, this instils a significant amount of fear, as they feel vulnerable and exposed to complaints, or even litigation:

"It is really common for mums to change their minds from when they see the midwife, to when they get to us. But that case (Mordel) really showed how careful we need to be documenting

³⁷ George F Winter, 'Semantics', (2021) 29 British Journal of Midwifery, 1, 3.

³⁸ Sonographer, England – interview response.

*this change. Clearly it is not enough to just take their word for it anymore, we need to make the right checks – but what are those checks?”*³⁹

These findings may explain the initial theme from the quantitative data, that HCPs – particularly sonographers and midwives – felt they were not clear on their role for securing parents’ consent for trisomy screening. These findings are also supported by Hartwig *et al.*, who confirmed that change of mind is common for screening, as the circumstances of the parents change.⁴⁰ Indeed, this is also consistent with the opinion of the expert witness for the Defendant in *Mordel*.⁴¹ However, the decision, in *Mordel*, failed to construct a logical solution, and indeed highlighted sonographers’ vulnerability in this regard.

Sonographers opined that, to ensure that they are protected in circumstances of change of mind, a more robust method of documentation is necessary. They expressed that their current method – to select the relevant tick-box on the computer system and pregnancy notes – is insufficient, in light of legal expectation:

*“It will get to the stage where we will need to voice record all the appointments with tapes to protect ourselves if they (parents) do change their mind. I cannot see any other way of proving this from the case with the Polish couple. The sonographer recorded the decision, but that was not enough. We need to know what is enough.”*⁴²

These findings support to two key themes from the quantitative data: that the majority of HCPs – particularly midwives and sonographers – do not believe that there are appropriate systems in place to secure parent consent, and that they are not confident supporting parent choice along the trisomy pathway.

These findings also speak to the need to establish a robust method of documenting pre-and post-screening consultation to evidence discussions. Suggestions were raised by Moyo *et al.* and Di Mattei *et al.* to record discussions from the appointment as evidence of the change of

³⁹ Sonographer, England – interview response.

⁴⁰ Tanja Schlaikjaer Hartwig and others, ‘High risk – What’s next? A survey study on decisional conflict, regret, and satisfaction among high-risk pregnant women making choices about further prenatal testing for fetal aneuploidy’, (2019) 39 *Prenat Diagn*, 635.

⁴¹ *Mordel* (n13), 76.

⁴² Sonographer, England – interview response.

mind.⁴³ However, this method would raise its own consent issues, as the nature of these discussions are highly sensitive and emotive; parents often reveal information about themselves that is highly confidential, and may not be appropriate to audio record.⁴⁴ It may be that establishing consent-based clinics with trained staff, which specifically deal with issues of consent, could appropriately facilitate such a technique.

6.5.2 Risk of Undermining Parent Choice

A strong theme among midwives and sonographers was the premise that exploring the understanding of parents, without appearing to undermine their choice, is often very challenging. Midwives and sonographers explained that their education and training places emphasis on *discouraging* HCPs from probing into the parents' understanding, as it appears infantilising and patronising, and could undermine parent choice:

“Checking patients’ understanding is really tricky because we do not want to come across as patronising or to undermine their decisions. In our (midwife) training, we are reminded not to undermine them (parents), but this is difficult if we suspect they do not understand.”⁴⁵

This finding is supported by the theme identified from the quantitative data, that HCPs – particularly sonographers and midwives – do not feel confident supporting parents' choices along the pathway. A possible solution to this has emerged in the form of the “teach me/back” method. This tool has recently been dubbed as an effective technique to explore parents' understanding without appearing interrogative, undermining or patronising.⁴⁶ A study by Yen *et al.*, in 2019, suggests that use of the “teach-back” method pose little, if any, significant risks to the autonomy of patients and effectively fortifies parent choice.⁴⁷ This technique requires parents to teach or repeat the information they received at their earlier appointments to the HCPs, as a method of exploring whether they have understood the information, before

⁴³ Mpatisi Moyo and others, ‘Healthcare practitioners’ personal and professional values’, (2016) 21 *Advances in Health Sciences Education*, 257; and Valentina Di Mattei, Federica Ferrari and others, ‘*Decision-making factors in prenatal testing: A systematic review*’, (2021) 23 *Health Psychology Open*, 1, 20.

⁴⁴ Pregnant person is asked about issues of domestic abuse, drug use, medical history etc.

⁴⁵ Midwife, Wales – open survey response.

⁴⁶ Sophie John and others, ‘*Influence of midwife communication on women’s understanding of Down Syndrome screening information*’, (2019) 27 *British Journal of Midwifery*, 1, 12.

⁴⁷ Peggy Yen and Renee Leasure, ‘Use and Effectiveness of the Teach-Back Method in Patient Education and Health Outcomes’, (2019) 36 *Federal Practitioner*, 284.

consenting to screening/testing.⁴⁸ However, this technique may lack the desired efficiency in light of the current time constraints experienced by NHS staff.

6.5.3 Concerns for Predetermined Decision-Making

Midwives disclosed that many parents enter the booking appointment having made a predetermined decision on screening, outside the clinical setting. They highlighted the challenge this poses, as parents feel they need not to consider the information materials on screening once they have made their ultimate decision. Thus, midwives fear that parents are not fully informed before making a decision to screen, and underline a lack of clinical guidelines to effectively navigate their duty in such circumstances:

“I ask my women why they want screening, and it is usually a decision they have made with their boyfriends or husbands before coming to see us. That is when difficulties start because they then do not feel the need to read the booklets or listen to the information in the appointment.”⁴⁹

This finding may explain the initial theme from the quantitative data set, that the majority of HCPs – particularly midwives and sonographers – are not confident supporting parent choices along the trisomy pathway. This is also consistent with the parents’ findings that revealed many parents will make the decision to screen, or not to screen, before engaging with the HCPs. This provides additional challenges for HCPs in terms of effective counselling.

Midwives demonstrated concern, in this study, to current practice which mandates that, if the parents are undecided at the booking appointment, they are to be booked to have the NT measurement with the 12-week dating scan, reserving the option *on the day* of the scan to decline screening. The reason for this is that additional time is required for the sonographer to take an NT measurement, as the baby must be in a specific position, and bloods will also need taking. However, midwives questioned whether parents feel pressured to have their NT measurement taken, once they had been booked to have it:

“Our biggest concern is that if they (parents) are unsure whether they want their NT scan or not, we book them in to have it because the sonographer needs more time to do that part of

⁴⁸ Ibid.

⁴⁹ Midwife, England – interview response.

the scan. If we do not book them in for the longer slot, and they decide they want it, they would need to re-arrange the appointment for a later time that week. I can see a problem with this because if they (parents) have been booked in to have the longer slot, how likely is it they will refuse screening on the day”.⁵⁰

This finding speaks to the provision of balanced and supportive care pathways. While it is protocol to book undecided parents to have the NT measurement performed, it is paramount that parents are reminded that they are able to decline screening on the day of the appointment, and they will be *supported* in doing so.

6.5.4 ‘Routine’ Pregnancy Care

Midwives are worried that the option to *refuse* screening is not being emphasised during pre-screening counselling. They explained that the option to refuse screening must be discussed with parents before consenting to screening, as parents often believe screening is a component of their ‘routine’ or ‘standard’ care. Remarkably, a small minority of midwives believed trisomy screening was ‘routine’, and not optional:

“What concerns us (midwives) most, is the fact that parents are usually not aware that screening is a choice, and a choice they can turn down. I do not think it is being emphasised enough at the booking appointments, from my experience.”⁵¹

This finding is consistent with the theme identified from the quantitative data, that the majority of parent groups were unaware that they were able to refuse trisomy screening and/or testing at any time. This finding also echoes the fears underlined in the parent findings on the risks of screening’s routinisation. Indeed, several studies that found parents perceived screening as ‘routine’ care. Thus, it may be that midwives ensure that the option to refuse screening is discussed in sufficient depth before obtaining the parent’s consent.

However, midwives expanded on this point, explaining that it is not within their duty to challenge the decision of parents once they have made a decision. They highlighted that an exploration into the parent’s decision-making may create the sense that their decision has

⁵⁰ Midwife, England – open survey response.

⁵¹ Midwife, England – open survey response.

not been respected, or that it was the ‘wrong’ decision. Midwives identified that this could pressure parents to change their mind:

*“We are not supposed to check and check the mum’s decision. If we did this, it could risk her (mum) to change her mind. It could also give off the impression that her decision is not being properly respected by us”.*⁵²

This finding is echoed by the fetal-maternal expert in *Mordel*, who affirmed that the midwife’s duty could not extend to ‘ask further questions’ on the point of parent choice, as revisiting the issue runs the risk of conveying the impression that the mother has made the wrong decision, and could ultimately change her mind.⁵³

6.5.5 Unintentional Decision to Screen for ES and PS

Sonographers reported that following brief discussions at the beginning of the examination, parents are frequently under the impression that they are *only* screening for DS, unaware of the fact they have also chosen to screen for ES and PS:

*“On the computer system and the notes we get, I can see if they want their NT measured or not for the trisomies. I ask if they still want the NT measured for the combined, but my heart sinks I ask them to confirm the go-ahead for Edwards and Patau’s and they ask, ‘what are they?’”*⁵⁴

This is consistent with the parent findings, explaining that they were only aware of DS, and subsequently overlooked that ES and PS were also being screened for. This finding alludes to the fact that consent may not be bespoke to each trisomy under the current screening model/programme.

6.5.6 Incidental Findings

Sonographers also outlined the ongoing issue surrounding their duty to report an increased NT measurement to the fetal medicine unit, despite the parents’ decision to decline having their baby’s NT measurement taken. They agreed that this duty should be better highlighted in the paper-based information or when they receive counselling from midwife. From the

⁵² Midwife, England – open survey response.

⁵³ Mordel (n13), 129.

⁵⁴ Sonographer, Wales – interview response.

sonographers experience, their overriding duty to report an increased NT measurement is often met with frustration, confusion or distress by parents:

“Our most challenging issue on withholding information is our duty to report a large NT fold - or other soft markers - to fetal medicine even if the parents have consented to not have it done. Understandably, mums do not realise that we have to report a bigger nuchal fold to the fetal medicine centre. Mums are given the option to have their bloods done to make up the other part of the test if it (NT) is increased, but they always say, ‘yes’, because it is human nature to want to know.”⁵⁵

This finding could explain the initial theme from the quantitative data, that HCPs – particularly sonographers and midwives – do not feel confident supporting parents’ choices along the trisomy pathway.

6.5.7 Managing Directiveness

The complete removal of bias, when imparting information on screening, is entirely idealistic, according to midwives. Midwives explained that parents will “see bias” in the information, as they will approach screening with preconceived ideas, and may interpret the information subject to their expectation of screening:

“It is not possible to impart non-bias information. Every human will naturally emit a particular bias, but the question is how we manage the bias. Parents will see a bias in whatever you say to them because they have preconceived ideas of screening, and what they expect from it”.⁵⁶

This finding is consistent with Ahmed *et al.*, who found that parents interpret information subject to their experiences, values and beliefs.⁵⁷ However, Steen *et al.* explained that HCPs are typically unaware of their directiveness, and may even unintentionally orchestrate the parents care management plan.⁵⁸ HCPs must be attentive to the existence of bias in

⁵⁵ Sonographer, England – interview response.

⁵⁶ Midwife, Wales – open survey response.

⁵⁷ Ahmed S, Bryant L and Hewison J, ‘Balance is in the eye of the beholder: providing information to support informed choices in antenatal screening via antenatal screening web resource’, (2010) 10 Health Expectations 309, 320.

⁵⁸ Sanne L. van der Steen and others, ‘Offering a choice between NIPT and invasive PND in prenatal genetic counselling: the impact of clinician characteristics on patients’ test uptake’, (2019) 27 European Journal of Human Genetics, 235.

communicating information on screening, to avoid influencing parent choice.⁵⁹ Indeed, the Nuffield Council emphasise that, “being skilled in delivering information in a non-directive way ... is essential for healthcare professionals who are supporting women to make informed choices”.⁶⁰

6.5.8 Routinisation of Terminations

Midwives expressed concern toward the possible routinisation of terminations. They asserted that if the information is overly pessimistic, and presents termination as the first choice following a positive diagnosis of a trisomy, this risks undermining parent choice. Midwives underlined the importance of access to specialist support units and organisations in this regard, particularly in the context of ES and PS:

“There is a well-known concern that terminations could become routine if practice is not regulated and reviewed consistently. Some of the information parents get convey a very negative portrayal of Down’s, and could risk influencing their choice.”⁶¹

This finding is consistent with the parent findings on ending pregnancies following a positive diagnosis of a trisomy. Guedj and Bianchi underlined the risks posed to parents’ autonomy from the routinisation of terminations.⁶²

Midwives – both screening and bereavement – explained that the provision of information and care for continuing with a pregnancy, post-diagnosis, requires a balanced and patient-centric approach. They explained that the risk of undermining parents’ autonomy is present if they do not receive the correct support for decision-making on ending or continuing with the pregnancy:

“It is so important that mums receive the right support at the right time before making decisions on aborting the pregnancy. There are specialist counsellors available, and it is so

⁵⁹ L. Bangsgaard and A. Tabor, ‘Do pregnant women and their partners make an informed choice about first trimester risk assessment for Down syndrome, and are they satisfied with the choice?’ (2013) 33 Prenatal Diagnosis, 146, 152; and Anne Moller and others, ‘Danish Sonographers’ Experience of the Introduction of “Moderate Risk” in Prenatal Screening for Down Syndrome’, (2018) 2 Journal of Pregnancy, 7.

⁶⁰ Nuffield Council Report (n16), 45.

⁶¹ Screening midwife, Wales – open survey response.

⁶² Guedj F and Bianchi D.W, ‘Noninvasive prenatal testing creates an opportunity for antenatal treatment of Down syndrome’ (2013) 33 Prenatal Diagnosis, 614.

*important that parents are referred to them when making these choices. It is our job to make sure they are given the proper support whatever choice they make”.*⁶³

This finding is consistent with Cope *et al.*, who found that the provision of specialist support, post-diagnosis of a potentially life-limiting condition, reduces the risk of parents suffering from negative physical and psychological outcomes.⁶⁴ Of those parents who decide to continue with the pregnancy, the provision of specialist support may also improve health outcomes for the baby, with early access to resources and neonatal care.⁶⁵

6.5.9 Partners’ Inclusion in Decision-Making

Midwives felt that partners’ inclusion in the decision-making process could be better encouraged at consultations and appointments. They felt that partners often submit to the needs of the pregnant person, and rarely become involved in the decision-making process for trisomy screening. Midwives felt that they could also do more to encourage inclusion, however there is a lack of training available to effectively execute this objective:

*“From my experience, there is no solution to this, but I think training could help us out with this issue. I think it is important to get the father’s involved, but how do we do this without interrogating them for questions, either; this could make them really uncomfortable and then they will avoid going to follow-up appointments. More training is probably the answer.”*⁶⁶

This finding could explain the initial theme from the quantitative data, that HCPs – particularly sonographers and midwives – did not feel they have received up to date training on securing consent since the implementation of the trisomy pathway. This finding is supported by Watterbjork *et al.* who found that partners typically lack confidence, and feel that the pregnant person should take the focus of the discussion at consultations.⁶⁷ The focus of these findings must not be placed on appropriately dividing the share of the HCP’s attention during counselling, but rather the importance of empowering partners, by creating an inclusive

⁶³ Midwife, England – open survey response.

⁶⁴ Cope H and others, ‘Pregnancy continuation and organisational religious activity following prenatal diagnosis of a lethal fetal defect are associated with improved psychological outcome’, (2015) 35 Prenatal Diagnosis, 761.

⁶⁵ Ibid.

⁶⁶ Midwife, Wales – open survey response.

⁶⁷ Wätterbjörk and others, ‘Decision-making process of prenatal screening described by pregnant women and their partners’, (2015) 18 Health Expectations: An International Journal of Public Participation in Health Care & Health Policy, 1585.

environment to encourage engagement with the decision-making process; this is particularly pertinent to those parents who face an unanticipated outcome from screening and testing.⁶⁸ These findings may also speak to the possible creation of a ‘partner-specific clinic’, which could provide the desired balance for partners.

6.6 Communication and Relationship Between Professional and Parent

6.6.1 Nature of the Parent-Professional Relationship

Midwives explained that a common concern among the profession is parents’ perception of their role as being synonymous with ‘Google’. Thus, they stressed that building rapport is increasingly difficult, and impacts their ability to uphold *Montgomery’s* shared decision-making values:

*“Lots of parents treat us like machines: they walk in, want all the information like we are ‘Google’, have the screening, and then carry on. Guidelines on consent tell us we need to maintain a relationship and a dialogue, but it works both ways”.*⁶⁹

Midwives also revealed that parents seek advice on their choices along the trisomy pathway, particularly during hardship. They reported that parents frequently ask for recommendations and suggestions at appointments on their approach to care management. Midwives felt that this factor is challenging, as they do not want to appear to be evading responsibility; however, they are prohibited from orchestrating parents’ care. They were also concerned that parents are willing to sacrifice their own autonomy by placing sole discretion over their choices into the hands of the profession:

*“We always get, ‘what would you do if you were me?’. It is really worrying that parents are willing to just say, ‘you make the choices for me’. We have to get the message across that we are well versed in promoting shared decision-making, but our role is to support their best interests, not for them to delegate decision-making to us – that is not our role here.”*⁷⁰

⁶⁸ Stina Lou and others, ‘Coping with worry while waiting for diagnostic results: a qualitative study of the experiences of pregnant couples following a high-risk prenatal screening result’, (2016) 16 BMC Pregnancy Childbirth, 321.

⁶⁹ Midwife, England – interview response.

⁷⁰ Midwife, Wales – interview response.

This finding is consistent with the study by Hertig *et al.*, who found that parents often rely on the HCPs when confronted with uncertainty, originating from anxiety and stress.⁷¹ A fine line between supporting parents' care and directing it, requires HCPs to engage into a continuous process of reflectivity throughout the parent's care management.⁷²

6.6.2 Protecting Professional Interests

As the law and practice guidelines have developed, midwives explained that the "safe option" is to adopt a neutral information provider approach to care management. They explained that this method is currently being adopted as a means to protect their professional interests, and to avoid "potential complaints" for directiveness. They revealed that complaints are becoming increasingly common, and therefore worry that supporting decision-making could be interpreted as favouring particular decisions:

*"It has got to the stage that being a neutral information provider is our only viable option to make sure we do not get complaints. Obviously we want to support the parents, but the risk is that parents will think we are favouring choices or promoting choices; it is a minefield."*⁷³

This finding could explain the initial theme identified from the quantitative data, that HCPs – particularly sonographers and midwives – do not feel confident supporting parents' choices across the pathway. This finding is concerning, as it points to the profession's fear of litigation. As a consequence, it appears that a model of care based on consumerism, devoid of the GMC's desired 'partnership', is preferred by the profession.

6.6.3 Building and Maintaining Rapport

Midwives explained that time limitations pose a significant hinderance to building and maintaining rapport across the pathway. They expressed that the ever-increasing amount of information they must review, within the allocated booking appointment, is impeding interactivity with parents during counselling:

"We only get forty-five minutes with our girls at the booking appointment, and that's including information on all the other parts of their pregnancy. I spend no more than a couple of minutes

⁷¹ Solene Gouihers Hertig and others, 'Doctor, what would you do in my position?' Health professionals and the decision-making process in pregnancy monitoring' (2014) 40 J Med Ethics, 310.

⁷² Ibid.

⁷³ Midwife, Wales – interview response.

talking about trisomy screening because we simply don't have the time. We have more and more things we need to cover every year but we do not have longer appointments."⁷⁴

This finding is supported by John *et al.*, who found that midwives are experiencing more pressure than ever before to counsel parents on trisomy screening, due to time limitations.⁷⁵ Indeed, these academics expressed that time pressures are having a significant impact on the ability of HCPs to build rapport with parents, necessary to effectively support their decision-making.

Sonographers found that the most prominent barrier to communication was the time limitations they experience in clinical practice. Sonographers felt that the decision in *Mordel* will only exacerbate this issue, as sonographers are required to counsel parents before performing the scan at the examination appointment. They conveyed that it is wholly unrealistic to expect sonographers to effectively communicate and counsel parents before the scan, without allowing more time during clinical appointments:

*"The biggest barrier is definitely time. This is only going to get worse in the wake of that case (Mordel). I think that it is completely unrealistic to properly communicate with parents – and build the desired rapport – without extending the time we have with mums for the ultrasound appointment. The time we have right now means that we could only counsel or perform the scan – not both."*⁷⁶

This finding speaks to the urgent need for programme co-ordinators to review current clinical guidelines for sonographers in this regard. Indeed, *Mordel's* requirements are not in keeping with the reality of clinical practice. It would be unreasonable – under current time constraints – to expect sonographers to counsel parents before securing their consent, and also perform the scan. This requirement is placing an unjust amount of pressure on sonographers to effectively execute their duty of care to support decision-making.

6.6.4 Intervention

Supporting parent choice and decision-making on continuing or ending the pregnancy, emerged as a theme among midwives and consultants. Consultants explained that the most

⁷⁴ Midwife, England – interview response.

⁷⁵ Sophie John (n13), 22.

⁷⁶ Sonographer, experience of both NHS and private, England – interview response.

challenging aspect of this stage in the pathway was consolidating parents' understanding of the positive and negative outcomes associated with the trisomies. They underlined that it is vital parents understand the implications, on both mother and baby, of having a baby with a trisomy. Consultants recognised that this is typically the source of dissent, and often friction, between parents and the profession:

*"Another issue this study needs to reflect is preparing patients for the outcome of an amniocentesis, including receiving a diagnosis. This is very challenging because women, understandably, do not want to accept there is anything wrong with the baby, but this is not always the case."*⁷⁷

This finding resonates with the views of some parents, in this study, who had a baby with a trisomy: they focussed on the negative information delivered by the consultants, and interpreted the discussion as demonstrating little, if any, value towards their unborn baby. One must question whether a parent would ever be prepared for such outcomes; however, it certainly underlines the value of the provision of specialist services (SOFT, DSA, etc).

Consultants also stated that the provision of specialist care, particularly for ES and PS, is subject to resource availability. They explained that it is often the case that there is limited access to specialist care, due to NHS resource limitations; an issue that is only worsening. Consultants stressed that parents do not have unmitigated access to the specialist care their child may need, which is often a source of controversy during counselling:

*"I completely get that expectant parents will be very stressed if their child gets diagnosed with T13 or T18. What we have to do is discuss the likelihood of the child being severely disabled, and weigh-in the fact that we are also restricted by resources, particularly in our Trust. The baby will need to be ventilated, twenty-four hour care and extensive surgery; we simply may not have the resources for this type of care. So, we have to discuss whether termination is the right option for parents, after careful consideration of these factors."*⁷⁸

This finding is significant, as it points to the broader systemic constraints HCPs must consider when counselling parents on decision-making for intervention. It may also speak to the

⁷⁷ Fetal medicine consultant, England – open survey response.

⁷⁸ Consultant, England – interview response.

difficulty HCPs experience communicating these factors to parents, without hindering rapport and tarnishing the relationship with the profession.

6.7 Counselling Parents on 'Risk'

6.7.1 HCPs' Role for Counselling on 'Risk'

Sonographers questioned their role to counsel parents on matters of 'risk'. They explained that, traditionally, their role did not extend to supporting parents' *understanding* of risk, rather they "briefly" cover it at the beginning of the appointment. Sonographer reiterated that they have not got the time during the examination appointments to support parents understanding of risk, as it is multifaceted and complex:

*"We do not usually go into detail on risk and what it means; this is usually done by the midwife. But since that case (Mordel), we are expected to go into detail with risk to make sure they understand all aspects of the procedure. We need to have the proper training on this because we just have not got the time to have a conversation about risk; it takes a lot of time because it is complicated".*⁷⁹

This finding is supported by the initial theme identified from the quantitative data, that HCPs – particularly sonographers and midwives – did not feel confident supporting parent understanding of 'risk'.

6.7.2 Rarity of ES and PS

Midwives also asserted that a risk exists of parents overlooking the possibility of obtaining a higher-risk result for ES or PS, following first-line screening. Consistent with the parent's findings, they underlined that, due to the recent introduction of the conditions to the pathway and their perceived rarity, parents who obtain a higher-risk result may be unprepared for this

⁷⁹ Sonographer, England – open survey response.

outcome. Midwives suggest that pre-screening consultations must pay equal attention to all trisomies, while avoiding overemphasising ES' and PS' rarity:

“A big concern is when mums get a high-risk result for Patau and Edwards but have no idea that screening was going to pull up a risk for these; their focus is usually on Down’s. The syndromes are very rare and unlikely, but this is obviously really upsetting for them, and they usually rely on us to support them with their choices at this time with other specialists.”⁸⁰

This finding supports the initial theme identified in the quantitative data, that the majority of HCPs – particularly midwives and sonographers – did not feel confident supporting parent understanding of ‘risk’. These qualitative findings also contextualise the quantitative theme, placing particular focus on the difficulties experienced supporting parent understanding of ‘risk’, in the context of ES and PS. Arthur and Gupta argue that HCPs are not sufficiently trained to provide risk factors on ES or PS, as they routinely fail to accurately disclose this information during pre-screening consultations.⁸¹ Under the *Montgomery* assessment of materiality, parents must be provided with accurate and contemporary information on the risk/prevalence of the conditions before providing their consent.

6.7.3 Counselling on Population and Individual Risk

Midwives expressed that a significant challenge, in practice, is to support parents’ understanding of the difference between population and individual risk. Consistent with the parent findings, midwives also found that many parents who would be categorised as lower-risk pre-screening – primarily due to age and medical history – are subsequently unprepared for a higher-risk result following primary screening:

“There is a lot of confusion with parents between what the overall population risk is for each trisomy – Down’s Syndrome being the highest out of all three – and their own individual risk after they have screening done. Obviously there is a substantial difference, but this needs to be communicated properly when we see them in clinic. I really have to drive that point home with younger couples”⁸².

⁸⁰ Midwife, Wales – open survey response.

⁸¹ Joshua D Arthur and Divya Gupta, ‘You Can Carry the Torch Now: A Qualitative Analysis of Parents’ Experience Caring or a Child with Trisomy 13 or 18’, (2017) 29 HEC Forum, 223.

⁸² Midwife, England – interview response.

Asbury underlined that HCPs typically spend far less time counselling younger parents on the risk of having a baby with a trisomy, who would be categorised as lower-risk based on population probability, compared to mature parents.⁸³ However, pre-screening counselling must support parents' understanding of the difference between population and individual risk in this regard.

6.7.4 Challenges Communicating Risk

Midwives explained that communicating the concept of risk to parents is challenging. They explained that inconsistency between the use of statistics and the risk categories are common in practice, as HCPs will prefer a particular mode of communicating this concept. Midwives strongly emphasised that they wish only to communicate a purely statistical interpretation of parents' risk, as opposed to using the policy determined high-and low-risk categories, due to the varying interpretations of these categories between Trusts:

*"I can see it must be confusing for them; I often get myself confused. I try to be as consistent as I possibly can when explaining what risk means, but I do see inconsistencies between the booklets and what is said at the clinic. I think the high-risk and low-risk categories are not effective for their understanding: I try to only use the statistics and explain what they mean."*⁸⁴

This finding supports the qualitative data, that the majority of midwives and sonographers did not feel confident supporting parents' understanding of 'risk' in the context of their screening results. This is consistent with John *et al.*, who outlined that midwives exhibited difficulty imparting information on risk/chance, and also maintaining the required consistency.⁸⁵ John *et al.*, reinforced that in only sixty percent of the appointments the risk statistics were disclosed, with some midwives *incorrectly* quoting the statistical interpretation or "cut-off" of the categories.⁸⁶

6.7.5 Parents' Interpretation of Screening Results

Consistent with the parent findings, midwives asserted that many parents translate a high-risk screening result as being diagnostic, and a lower-risk result as meaning the baby does not

⁸³ Bret Asbury, (n34), 301.

⁸⁴ Midwife, England – interview response.

⁸⁵ Sophie John and others (n13), 12.

⁸⁶ *Ibid.*

have a trisomy. They underlined the difficulties counselling parents in this regard, and encouraging them to appreciate that screening results are not determinative of the babies health outcome. Midwives affirmed that communicating the concept of false-positive and negative screening results is paramount before securing parents' consent:

*"Mums see 'high-risk' and instantly panic. All parents think that their child has the condition from the risk result. The difficulty comes getting them to appreciate that because they are high or low-risk, that does not mean the baby has – or does not have – the syndrome. It is a 'risk'."*⁸⁷

This finding is supported by many studies which underline parents' misunderstanding in this regard.⁸⁸ Dahl *et al.* revealed that up to sixty-five percent of the participants, in their study, were not aware of the possibility of false-positive and negative results.⁸⁹ This speaks to managing the parent's misconceptions of the purpose of screening results; that is, it is a probability score, rather than being indicative of *actual* risk.

⁸⁷ Midwife, Wale – open survey response.

⁸⁸ Susanne Georgsson Ohman, Charlotta Grunewald and Ulla Waldenstrom, 'Perception of risk in relation to ultrasound screening for Down's syndrome during pregnancy', (2009) 25 Midwifery, 264.

⁸⁹ Katja Dahl, Ulrik Kesmodel, Lone Hvidman and Frede Olesen, 'Informed consent: attitudes, knowledge and information concerning prenatal examinations', (2006) 85 Acta Obstet Gynecol Scand, 1414.

6.8 Alternative Methods of Testing

6.8.1 Training on Non-Invasive Prenatal Testing (NIPT)

The provision of training on NIPT for sonographers, emerged as a prominent theme, in this study. Sonographers feel that the *same* CPD and training, that midwives receive for counselling parents on NIPT, should also be made available to them. They explained that questions from parents on NIPT screening, at the examination appointment, is becoming increasingly common; however, they feel unequipped to answer questions, due to a lack of professional training:

“I’m getting more questions on NIPT. But if the woman asks about NIPT and how it works, I usually refer them to the booklets or tell them that this will be covered by their midwife if they are high-risk. But I think, if we are supposed to get mums’ consent, surely this means we are supposed to dedicate some time covering NIPT ourselves. If that is the case, then we need to be trained to go over this information properly, otherwise the information we give will be inaccurate”.⁹⁰

This finding supports the initial theme from the quantitative data, that the majority of HCP groups – particularly sonographers and midwives – felt that they had not received up to date training on NIPT, for the purpose of supporting parent decision-making in this regard. Moller *et al.*’s study exposed sonographers’ lack of understanding of the practicalities and implications of NIPT, as they had not received the requisite training since its introduction to public health care services.⁹¹ While ASW confirmed that training resources are currently being

⁹⁰ Sonographer, experience in both NHS and private sector, England – interview response.

⁹¹ Anne Moller and others, ‘Danish Sonographers’ Experience of the Introduction of “Moderate Risk” in Prenatal Screening for Down Syndrome’, (2018) 3 Journal of Pregnancy 1, 7.

designed for sonographers on counselling parents for NIPT, these findings signify the importance of CPD and mandatory training resources (e-learning) to support sonographers in effectively counselling parents on the practicalities of NIPT, including the implications of the test.⁹²

6.8.2 Quality of Professional Training on NIPT

Midwives demonstrated concern toward the quality and delivery of professional training on NIPT. Training varied significantly among this group: ‘training’ was referred to as either attending workshops, ‘cascade’ training, e-learning resources, or receiving information documents from screening leads. Midwives, primarily those based in English NHS Trusts, explained that while e-learning is their current source of training, this had not been modified since the anticipated roll-out of NIPT in 2016. All midwives stressed that with the implementation of NIPT, they must have access to updated training based on the most recent research studies:

*“We do have the training available on the Public Health England e-learning module, but it is not compulsory, and no one checks if it had been completed. We are supposed to do it every 18 months, but nothing flags up that you need to do it. I always have to check myself on e-learning whether I need to do a course or not”.*⁹³

This finding supports the initial theme from the quantitative data, that the majority of HCP groups – particularly sonographers and midwives – felt that they had not received up to date training on NIPT. This finding is also supported by the Nuffield Council report, which stated that training is available on NIPT via e-learning modules provided by Public Health England; however, uptake of these modules is not compulsory.⁹⁴ These findings are consistent with Ekelin and Crang-Svalenius, who found that midwives routinely failed to receive adequate training following the introduction of a new method of screening.⁹⁵ Geranmayeh *et al.* drew attention to the quality of e-learning in prenatal care, and found that the quality of training varies significantly. It may be that enhancing interactivity on e-learning modules could

⁹² Ida Charlotte Bay Lund and others, ‘Preferences for prenatal testing among pregnant women, partners and health professionals’, (2018) 65 Dan Med J, 5486.

⁹³ Midwife, England – interview response.

⁹⁴ Nuffield Council report (n16), 47.

⁹⁵ Ekelin M and Crang-Svalenius E, ‘Midwives’ attitudes to and knowledge about a newly introduced foetal screening method’, (2004) 18 Scandinavian Journal of Caring Sciences, 287.

encourage HCPs to engage with the training, and subsequently acquire better knowledge on recent developments in prenatal care.⁹⁶ This finding also calls for the unification of training standards provided to all Trusts, across England and Wales, to ensure consistency and quality among health boards.

6.8.3 NIPT as an ‘Alternative’ to Invasive Testing

Midwives framed NIPT as an ‘alternative’ method of testing to amniocentesis and/or CVS. They explained that NIPT is offered to those parents who do not want to undergo ‘risky’ invasive methods of testing, following a screen-positive result. Midwives, in England, reported that they are obligated to make parents aware of the availability of NIPT as an alternative on the private market, if their Trust did not offer NIPT on the NHS:

“We offer NIPT instead of the amnio if the mum is high-risk. NIPT is safer and less risky than the amnio, so it is the attractive option for mums.”⁹⁷

This finding is significant and could explain why parents currently confuse NIPT as being an ‘alternative’ to invasive testing. This finding may be explained by the initial theme identified from the quantitative data, that all HCP groups – particularly sonographers and midwives – felt that they had not received up to date training on NIPT, for the purpose of supporting decision-making. Offering NIPT ‘instead of the amnio’ could convey the impression that it *replaces* invasive testing, which it does not.

6.8.4 ‘99% Accurate’ Narrative

Following a high-risk, or screen-positive, result, parents are typically referred to obstetricians and/or fetal medicine/neonatal consultants for further counselling. During consultations, counselling is provided on NIPT as an option, *alongside* amniocentesis/ CVS, to establish a care plan. Consultants explained that use of the phrase ‘99% accurate’, to describe NIPT’s detection rate, irked them. They explained that this is simply not true, and emphasised that this phrase does not correctly describe NIPT’s detection rate. Consultants confirmed that employing terms such as positive and negative predicted values, sensitivity and specificity, to

⁹⁶ Mehrnaz Geranmayeh and others, ‘Interactive and non-interactive e-learning in prenatal care’, (2021) 29 1, 9.

⁹⁷ Midwife, Wales – open survey response.

accurately describe the individual patient's chance of having a baby with a trisomy, is mandatory for parent understanding:

*"It really annoys me when I see that phrase '99% accurate'; it is just simply not true. I think we need to start making sure that healthcare staff, who offer this screening, need to be reminded to use the terms PPV, NPV, sensitivity and specificity. Understanding these terms can be complicated, but that is the only way to accurately describe what that patient's likelihood is."*⁹⁸

This is consistent with the Nuffield Council report, which emphasised that conveying the positive and negative predicted values of NIPT is crucial to support parents' understanding of the test's accuracy.⁹⁹ This report explains that it is not sufficient for HCPs to describe NIPT's accuracy under a catch-all '99%', as the detection rate will vary significantly between populations, and the conditions tested.¹⁰⁰

6.8.5 Concerns for NIPT's Routinisation

Midwives expressed concern towards the potential routinisation of NIPT, primarily due to its procedural simplicity and safety. They underlined the risk that NIPT may be perceived as 'just another routine blood test', not only by parents, but also HCPs. As a result, midwives fear that without the provision of specialist counselling, parent decision-making and consent may become undermined. They also explained that, from their experience, parents often undertake NIPT screening to seek reassurance, and expect the result to be negative in this regard:

"There is that risk that mums will view NIPT as 'just another one of those blood test' they have during pregnancy, if they are not being properly counselled on it, and might see NIPT as something that they have routinely throughout the pregnancy for other things anyway. It's about making sure that mums understand that this is a very important decision to make, with potentially lifechanging consequences".¹⁰¹

⁹⁸ Fetal Medicine Consultant, Wales – open survey response.

⁹⁹ Nuffield Council Report (n16), 45.

¹⁰⁰ Ibid.

¹⁰¹ Midwife, Wales – interview response.

This finding is consistent with Heuvel *et al.*, who underlined the risks to NIPT's routinisation, in light of the current narrative surrounding its procedural safety and accuracy.¹⁰² This finding is concerning, as it may be that parents receive a positive NIPT result, without adequate support or preparation for the options that will follow.¹⁰³

This finding is further compounded by the introduction of NIPT as an *automatic* contingency test, under some NHS Trusts in England, following a higher-risk (1:800 or more) combined result. Following the booking appointment, the pregnant person consents to have two vials of blood drawn at their 12-week appointments: one for the purposes of the combined test and the other for NIPT, in the event of a screen-positive combined result:

*"The way they do things here, is that if my girls (pregnant persons) score high-risk from the combined screening, the second sample of blood the hospital has is automatically send for NIPTY purposes; that is all done automatically, and the first mums will know of the higher-risk, from the combined, is when they get that letter through the door for the NIPTY results."*¹⁰⁴

This finding is consistent with the Nuffield Council report.¹⁰⁵ This begs the question whether this automatic process deprives parents of the opportunity to receive specialised counselling on NIPT, and the time required to reflect on their decisions and choices. Indeed, the Nuffield Council working group opined that this process risks entirely compromising parents' ability to deliver informed consent for NIPT.¹⁰⁶

6.8.6 Incidental or Unanticipated Findings from NIPT

The risk of incidental or unanticipated findings to maternal health, following NIPT's introduction to the pathway, was a prominent concern among consultants. They explained that incidental/unanticipated findings from NIPT include unforeseen information on the pregnant person's genetics, and maternal cancers/malignancies. Consultants explained that, despite laboratories having the ability to conceal the presence of such findings and only

¹⁰² van den Heuvel A and others, 'Will the introduction of non-invasive prenatal diagnostic testing erode informed choice? An experimental study of health care professionals,' (2008) 78 Patient Education and Counselling, 24.

¹⁰³ Ibid.

¹⁰⁴ Midwife who has a baby with trisomy (DS), England – interview response.

¹⁰⁵ Nuffield Council Report (n16), 98.

¹⁰⁶ Ibid.

disclose the information requested by the parents, incidental findings raise may significant legal and ethical implications, requiring a review of practice guidelines in this regard:

“The great concern coming from medical research is secondary findings from NIPT, and how staff are expected to deal with these. Some labs we send the bloods to are capable of using technology to mask secondary findings, but others do not. Once NIPT becomes standardised across all Trusts, the method of how we deal with such findings, if they do arise, must be reviewed to avoid harm to our patients.”¹⁰⁷

This finding is consistent with Bianchi *et al.*, who concluded that provision of specialist training, education and support must be available, within healthcare systems, to inform practice for incidental findings, following NIPT screening.¹⁰⁸ The Nuffield Council conclude that more research on patients’ experiences must be undertaken to further inform practice in this regard.¹⁰⁹

6.8.7 Impact of the Private Market

Midwives demonstrated concern towards the growing popularity of NIPT under the private market. They explained the impact of this is being felt across many NHS maternity units, with parents often seeking to rely upon the midwife’s advice and support after receiving a screen-positive result from private NIPT screening. Midwives expressed that the level of knowledge required to support parents’ decision-making mandates urgent specialist training, as there are a growing number of rare conditions being introduced under private screening panels:

*“The main concern for me is that mums will come to us who have been told their NIPT result is high, but for very rare conditions. How are we supposed to support their choices, when we do not understand what these conditions mean for the parents ourselves? It has got to the stage where we need access to training on this”.*¹¹⁰

This finding is consistent with the concerns of the Nuffield Council, who reported that midwives are unlikely, or will be “ill-equipped”, to counsel parents on the rarer conditions

¹⁰⁷ Fetal Medicine Consultant, England – interview response.

¹⁰⁸ Diana Bianchi and others, ‘Noninvasive Prenatal Testing and Incidental Detection of Occult Maternal Malignancies’, (2015) 314 *Jama*, 162.

¹⁰⁹ Nuffield Council Report (n16), 66.

¹¹⁰ Midwife, England – interview response.

available on the private market, due to a lack of training and knowledge in this regard.¹¹¹ These findings may provide an early indicator and warning to screening co-ordinators, to implement support and training programmes for midwives in this regard, given the increasing popularity of private NIPT screening.

Consultants drew attention to the provision of information and support provided under the private market, and the risk it poses to parent decision-making and consent. Consultants note that there has been a considerable increase in parents relying on the NHS for advice and support following private NIPT screening. They explained that the allure of private care is greater, due to the ever-growing panel of conditions available for screening, and marketing tactics (i.e discounts, care packages, one-time deals etc). However, consultants conveyed that the provision of information and counselling, provided under the private sector, varied significantly in quality, commonly being described as misleading, inaccurate, or aspires to reassure parents without sufficiently outlining the consequences of the test. They expressed that this has significant implications for NHS staff on their ability to support parents' decision-making following a higher-risk result:

*“My greatest concern is mums getting NIPT done privately. There is a substantial disparity in the quality of counselling and information provided by private care practitioners on NIPT. More and more mums come to us (NHS) shocked that they have had a high-risk result for a particular condition, and then we have to assist them to make choices. I can only see this issue getting worse, too”.*¹¹²

This finding is consistent with the Nuffield Council's concerns in this regard.¹¹³ The Council explained other risks raised by these findings such as, “increased shock, distress and confusion upon receipt of a higher-risk result, or even the termination of an unaffected fetus if the chance of a false positive is not clearly communicated”.

6.9 Concluding Observations and Comments from the HCP Qualitative Findings

The aim of this qualitative phase in the explanatory research paradigm was to further explore the initial themes of identified from the quantitative data, but also uncover any sub-themes

¹¹¹ Nuffield Council Report (n16) 101.

¹¹² Neonatal Consultant, England – open survey response.

¹¹³ Nuffield Council Report (n16), 98.

relating to the experience of HCPs operating under current systems of consent for trisomy screening. This phase of the research allowed the researcher to confirm or refute the initial themes identified from the quantitative dataset, and to provide a contextual insight into the initial patterns of behaviour.

The qualitative data has also uncovered significant relationships between the quantitative and quantitative dataset, providing the researcher with a rich understanding and contextual insight into the intersection between current systemic considerations and the interests of HCPs when securing parent consent for trisomy screening and/or testing. It has also revealed a range of sub-themes, which allowed the researcher to gain a clearer understanding of the individual roles HCPs play along the pathway, and how current systems of consent may not be conducive to legal expectation (namely *Montgomery* and *Mordel*) in this regard.

Chapter 7: Discussion

This chapter will draw on the empirical data, from the HCP and parent quantitative and qualitative studies, as a means to explore the parent and professional interests for delivering and securing consent for trisomy screening. Subject to the themes initially identified in the quantitative data, and subsequently explored in the qualitative data, this chapter will be divided into seven sections for clarity and cohesion: securing consent; provision of information, supporting parent understanding; supporting parent choice; delineating communication between the HCP and parents; supporting parent understanding of risk; and consideration of alternative methods of testing.

7.1 Securing Parent Consent: Addressing Systemic Inconsistencies and Failings

7.1.1 Delineating HCP and Systemic Duty

The decision in *Montgomery* has been widely cited as being beneficial to enhance the patient's ability to exercise autonomy and self-determination, in the context of medical treatment and care. Analysis and assessment of discharging the HCP's duty for information disclosure is pervasive throughout academic commentary following *Montgomery*, often reiterating the notion that patients are the bearers of rights, and that information must be provided on the benefits, risks and alternatives of proposed methods of medical treatment and procedures. Indeed, 'materiality' of said information is assessed subject to whether a reasonable person in the patient's position would likely to attach significance to the risk, or the HCPs is, or should reasonably be aware, that the particular patient would likely attach significance to it.¹

However, this is only the first dimension in terms of assessing whether consent has been obtained. To treat consent as a 'one-off event', confined solely to pre-treatment consultations, is becoming increasingly outdated.² An accurate interpretation of the process should be in line with that of a continued and dynamic dialogue, fortifying the shared decision-making values of clinical guidelines (namely the GMC), and contemporary common law decisions on consent. Thus, it seems that the remaining portion of the tale should rest on the assessment of provision and reasonableness in *securing* parent consent.

This research has found that consent, in the context of trisomy screening, operates as a multistage process.³ Securing consent for trisomy screening requires the input of midwives (booking appointment) and sonographers (ultrasound examination appointment), with consultants becoming increasingly involved following the pathway's inclusion of ES and PS.⁴ Thus, the court's approach to establishing whether consent is 'valid', may require

¹ *Montgomery v Lanarkshire Health Board* (2015) UKSC 111, 87.

² Graeme Laurie, Shawn Harmon and Edward Dove, *Mason and McCall Smith's Law and Medical Ethics* (11th edition, 2019), pp.98.

³ HCP Qualitative Findings, Section 6.2.

⁴ HCP Qualitative Findings, Section 6.2.3.

consideration of this reality, broadening its scope to involve an assessment of the *systems* HCPs are required to operate, during the acquisition of parent consent.⁵

The notion of a ‘systemic duty’ was considered by Brooke LJ in *Robertson v Nottingham*.⁶ This case assessed the operation of communication systems between units and HCPs, during the course of the Claimant’s treatment. It was held that the hospital had breached its duty of care to establish a safe system of operation during the Claimant’s care management.⁷ Brooke LJ surmised that:

*‘it is customary to say that a health authority is vicariously liable for breach of duty if its responsible servants or agents fail to set up a safe system of operation ... this formulation may cloud the fact that in any event it has a non-delegable duty to establish proper system of care ...’*⁸

More specifically to the issue of obtaining consent, systemic failings – during the course of securing consent – were outlined in *ARB v IVF Hammersmith*.⁹ The court, in this ‘wrongful birth’ case, had to assess whether the clinic’s failure to obtain ARB’s written informed consent before implanting an embryo – containing ARB gametes – into R, amounted to a breach of its duty to take reasonable care to obtain the Claimant’s consent. On the Claimant’s second ground of appeal – relating to the process of securing consent – Lady Justice Davies held that the clinic’s system of obtaining consent was *‘not only illogical, it makes a mockery of the process’*, despite expert witness supporting the established system.¹⁰ The process was said to represent *‘... an abrogation by the clinic of its duty to obtain consent’*.¹¹ This case reinforced the significance of establishing reasonable and logical systems of obtaining parent consent, which is the cornerstone of the shared decision-making values of the NHS, and the principles of law from *Montgomery*.¹²

⁵ Rob Heywood, ‘Systemic Negligence and NHS Hospitals: An Underutilised Argument’, (2021) 32 King’s Law Journal, 437.

⁶ *Robertson v Nottingham Health Authority* (1997) 8 Med LR 1.

⁷ *Ibid*, 13.

⁸ *Ibid*, 13.

⁹ *ARB v IVF Hammersmith* (2018) EWCA Civ 2803.

¹⁰ *Ibid*, 59.

¹¹ *Ibid*.

¹² *Ibid*.

This study suggests that systemic failings may be hindering the HCP's ability to sufficiently secure parent consent for trisomy screening. The research has affirmed that the duty on HCPs to take *reasonable* steps to secure parent consent currently is unclear; neither sonographers nor midwives have a clear understanding of where this duty truly rests, and feel they are "scapegoats" for existing systemic deficiencies.¹³

The decision in *Mordel v Royal Berkshire Trust* specifically alluded to broader systemic fragilities when securing consent for DS screening.¹⁴ To the question of whether the midwife or sonographer secured parent consent, the expert witnesses and counsel, for both the Claimant and Defendant, conflicted on the established systems under which HCPs must operate. Indeed, the divergent expert witness statements were symbolic of the current inadequacies and ambiguity surrounding established systems.

The expert witness for the Defendant, in *Mordel*, stated that: '*the midwife at the booking appointment goes through the issues in significant detail, and effectively obtains consent at that moment in time ... the sonographer's role is to confirm that consent is forthcoming on the day of the (combined) test*'.¹⁵ This interpretation is also consistent with this study's findings.¹⁶ Despite this, Jay J disagreed with this interpretation of the sonographer's role, and asserted that, '*I do not accept that the sonographer's role is limited to taking the patient's decision on way or another ... informed consent to the procedure in question still had to be provided*', and subsequently obtained by the sonographer.¹⁷ Jay J expressed that: '*... it is the sonographer's duty to satisfy herself that the patient is consenting to the procedure, either with or without the NT, before it is undertaken on the basis of proper information; and that her consent is informed*'.¹⁸ The decision introduced the notion that the midwife has merely provided an informed 'offer' at the booking appointment, rather than secured consent; however, the reasoning for this remains unclear.

While *Mordel* concluded that it is the responsibility of the sonographer to secure consent under the proposed system, this research suggests that convention has long established that

¹³ HCP Quantitative Study, Section 4.4 and Qualitative HCP Findings, Section 6.2.1.

¹⁴ *Mordel v Royal Berkshire NHS Trust* (2019) EWHC 2591.

¹⁵ *Mordel* (n14), 62.

¹⁶ HCP Qualitative Findings, Section 6.2.1.

¹⁷ *Mordel* (n14), 89.

¹⁸ *Ibid*.

consent is secured by the midwife at the booking appointment, and it is the midwife who plays a leading role in this regard.¹⁹ Indeed, it seems that the operative portion of Jay J's judgment, in *Mordel*, ran contrary to established system for securing consent; however the question remains as to why.

The application of *Bolitho* in *Mordel* to the assessment of a 'reasonable' system retained the desired judicial autonomy to the question of breach: careful consideration was taken of the risks and benefits of the working system to secure consent, and the logic in which it was founded. Despite correctly stating that the NHS could not operate if 'fail-safes' were implemented, Jay J's assessment of 'reasonableness' highlighted the difficulties the courts may encounter when applying the question of reasonableness to *systemic* failings, particularly when practice extends across two different fields: midwifery and ultrasonography. The decision created significant ambiguity and uncertainty as to whom truly obtains parent consent, due to the lack of a coherent or synthesised approach.²⁰

While Jay J should be commended for broadening and reshaping his assessment of reasonableness from the individual HCPs, which is traditionally the focus of actions in tort, to the system in which they operate, he failed to exercise a rounded and holistic evaluation of interdisciplinary practice. An isolated and coherent assessment of the various nuances and challenges of working within busy, time-pressured NHS maternity units, should also have been executed by Jay J: an argument identified from this research, and put forth by the Defendant's expert witnesses.²¹ As it stands, Jay J's interpretation of what is 'reasonable' would warrant an overhaul of established practice, requiring significant amendments to existing systems for securing parent consent, and subsequently training standards.

The Society of Radiographers (SoR) have recently updated their clinical guidelines by directing sonographers to the latest FASP clinical guidance on securing consent for trisomy screening. The SoR noted "some extremely important additions to the (FASP) document relating to parent-centred care and choice ... in light of *Mordel* ...".²² In reference to the FASP guidelines, the SoR underline that "it clarifies the role of the midwife and sonographer in the consent

¹⁹ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.2.1.

²⁰ Ibid.

²¹ Rachael Baillie, "A square peg in a round hole: Reshaping the approach to systemic negligence in modern public service", 2014, Auckland University Law Review, 20, 39-66.

²² Society of Radiographers Guidelines on Consent (2021), section 7.

process, while also highlighting that consent is not a static decision, it can change throughout the screening pathway”.²³ While this is certainly a promising development, FASP have arguably left the ‘process’ ill-defined. In line with *Mordel*, section 3 on ‘personal choice’ states that, “midwives are responsible for explaining national information on screening at first contact ... ultrasound practitioners are responsible for checking the woman’s understanding”; however, there is no mention of whether the sonographer or midwife secures consent, or how this should be executed.

Modifications and adaptations to existing systems for securing parent consent may not be beyond the capacity of the maternity units, if deemed necessary. The outbreak of COVID-19 demonstrated that significant overhauls and remoulding of established systems can be executed efficiently and (presumably) effectively within maternity units. As the findings illustrate, the established system for securing parent consent was abandoned following the pandemic, with appointments and the recording of consent moving entirely to online and telecommunication systems.²⁴ While the effectiveness and robustness of the virtual methods of securing consent for trisomy screening are yet to be validated, the pandemic underlined that maternity units are able to swiftly adapt and modify established systems when required.²⁵

Despite this, the findings of this research beg the question whether the judiciary are best placed to decide on the reasonableness of a system, particularly where expert witnesses are undecided on such specialist, interprofessional practice. Ultimately, the decision may bring into question the court’s ability to assess the reasonableness of a system – to secure parent consent – in areas where practice extends across two separate disciplines: here across the fields of midwifery and ultrasonography. This may allude the bluntness of the conventional tort principles as a tool for deciding on matters of systemic negligence, in the context of interprofessional and specialist practice. It may also point to the underutilisation of the doctrine *res ipsa loquitur*, as a medium to capture the nature of existing systemic failings, rather than interfering with matters for medical judgment.²⁶

²³ Ibid.

²⁴ HCP Qualitative Findings, Section 6.2.3.

²⁵ Ibid

²⁶ Rob Heywood, (n5), 446.

7.1.2 Enhancing Interprofessional Partnerships

What is clear from *Robertson, ARB* and *Mordel*, is the need to enhance interprofessional partnerships, for the purpose of effectively implementing a 'reasonable' system for HCPs in which to operate. On the issue of obtaining consent, the 'drop-down' tick-box system of recording consent the HCPs used in *Mordel*, was subject to intense scrutiny and criticism by the judiciary, labelled as simplistic and lacking the desired systemic robustness. However, the court failed to provide a *reasonable* alternative during its assessment of the systemic failing. The findings of this research also underline the fragility of existing 'appropriate' systems for securing the parent's consent between units, and highlights the vulnerability of midwives and sonographers to complaints or litigation in this regard.

Mordel provided a timely demonstration of the frailty of established methods for recording parent consent, under a time-pressured NHS maternity unit. A narrative that emerged from the Defendants' counsel on the breakdown in communication between the sonographer and midwife, pointed to the time-pressures and an innate lack of direction on how to reasonably record and secure consent, under such pressured environments. Sonographers are particularly concerned with the disconnect between ultrasonography and midwifery units, and its impact on effectively obtaining consent.²⁷

This study underlines the importance of establishing interprofessional partnerships to secure consent for trisomy screening. The area of maternity care and obstetrics are fielded by specialist and highly skilled professionals, such as midwives and sonographers. However, it became abundantly clear from the expert witness statements and subsequent decision in *Mordel*, that a lack of cohesion and synergy exists between the professional roles. As Newsam underlines, her experience of working within obstetrics and maternity care reinforced a lack of collaboration between professions, often exhibiting an 'us and them' culture.²⁸ This is also consistent with this study's findings, with midwives and sonographers often stating, "that's their job, not mine".²⁹

²⁷ HCP Qualitative Findings, Section 6.2.1.

²⁸ Rebecca Newsam, 'Operating department practitioners and midwives: The undervalued obstetric care collaboration', (2018) 26 *British Journal of Midwifery* 1, 8.

²⁹ HCP Qualitative Finding, Section 6.2.1.

It is plausible that this culture of segregation between professional roles may be responsible for the systemic disconnect, demonstrated in *Mordel*. This culture may be exacerbated not only by “the lack of understanding of different professional roles”, but “a deficit in training and education in obstetrics for different professional groups”.³⁰ The issue of inadequate training was particularly pertinent to the question of breach in *Gottstein v Maguire and Walsh*.³¹ While this case concerned the hospital’s failure to provide adequately trained staff for the purpose of specialist care management within an intensive care unit, it underlined the importance of the provision of continued professional development and training, to ensure appropriate provision of care management and systems of communication between units.

Arguably, it is desirable to establish and promote interprofessional partnerships, particularly in the context of securing consent, to enhance the quality and consistency of best practice.³² Bridging the gaps in knowledge and understanding between the professions will not only enhance the quality and consistency of clinical practice, but could also provide a nuanced and holistic insight for the courts to effectively decide on such specialist matters. Thus, as Newsam suggests, it may be that “incorporating interprofessional training programmes into undergraduate healthcare degrees is ... vital to encourage a culture of synthesised, integrated, holistic and safer care for the future”.³³

It may be that the training provided to midwives on counselling and securing parent consent be brought into line with training offered to sonographers. This would provide the desired “consistency in provision of counselling and assurance of individuality for the purposes of securing consent”.³⁴ To avoid exacerbating existing fragilities in the system, rather than run contrary to established practice, as Jay J did in the decision, a reasonable system could also be to extend the role of the midwife to address issues of consent at the 12-week examination appointment *with* the sonographer.³⁵ Another possibility is to employ midwives with a specific purpose of supporting sonographers to secure consent at the 12-week ultrasound appointment. However, while the creation of a role to specifically deal with matters of

³⁰ Rebecca Newsam, (n28), 1.

³¹ *Gottstein v Maguire and Walsh* (2007) 4 IR 435.

³² Rebecca Newsam, (n28), 1.

³³ *Ibid*, 7.

³⁴ *Ibid*.

³⁵ Emyr Owain Wile and Alys Einion-Waller, ‘*Understanding Mordel: obtaining informed consent for trisomy screening*’, (2021) 29 *British Journal of Midwifery*, 111.

consent is arguably required, this may be idealistic, considering the widespread shortage of trained NHS staff and resources.³⁶

The creation and distribution of interprofessional educational programmes on consent for trisomy screening may be effectively implemented online. As Power *et al.* underline, the response to the COVID-19 pandemic meant that the fortification of interprofessional relationships – primarily to protect the operation of existing systems in clinical practice – had to be executed online.³⁷ These academics note that while the shift to online interprofessional education posed unprecedented technological challenges, many benefits emerged in terms of accessibility and synthesis of professional programmes.³⁸ Thus, it may be that existing programmes for securing consent, in the fields of midwifery and ultrasonography, could be implemented and synthesised using online methods.³⁹

7.1.3 Sonographer-Midwife Consent Clinic

With the above in mind, a ‘reasonable’ system may be to establish an interprofessional, sonographer-midwife-led consent clinics. Under *Bolitho’s* assessment of risk against benefit to the question of a reasonable system, it would be logical to synthesise the process of securing parent consent through the creation of interprofessional consent clinics, with an objective to address any ambiguity and lacunae in the process. Newsam explains that “bringing different professional groups together to learn as a team can give each group new insights and ideas, synthesising their differences and individual skill sets”.⁴⁰

Sparks and Nixon note the benefits of establishing interprofessional clinics to enhance the fluidity of systems and practice in maternity care, specifically third trimester ultrasound screening.⁴¹ Indeed, this proposed model of care could also be effectively transferred and utilised for the purposes of establishing a synchronised approach to secure consent for trisomy screening.

³⁶ See *Bull v Devon Area Health Authority* (1993) 4 Med LR 117 on the issue of assessing systemic negligence subject to resource implications.

³⁷ Alison Power and others, ‘Academics’ experiences of online interprofessional education in response to COVID-19’, (2022) 30 *British Journal of Midwifery*, 1.

³⁸ *Ibid.*

³⁹ HCP Qualitative Findings, Section 6.2.3.

⁴⁰ Rebecca Newsam, (n28), 6.

⁴¹ Pippa Sparks and Vicky Nixon, ‘Midwife scan clinic: response to increased demand for third trimester ultrasound’, (2022) 30 *British Journal of Midwifery* 1,7.

While resources, financial constraints, and available time predictably give rise to challenges for the creation of such clinics, this proposition could be seamlessly implemented as a derivative to the booking appointment. Rather than counselling parents twice on issues of consent, as *Mordel* recommends – first by the midwife’s ‘informed offer’ at the booking appointment, and subsequently the sonographer’s duty to secure consent at the examination appointment – logic would dictate that both HCPs could work in tandem to establish a robust method for documenting and securing consent, within the *same* clinical appointment. This suggestion may also alleviate the pressures sonographers’ experience to counsel parents on matters of consent, and to effectively perform the ultrasound during the same appointment.

7.1.4 Divergence or Convergence – Systemic Considerations under Devolution on Trisomy Screening

As discussed in chapter 2, health is a devolved matter. Indeed, divergency is evident in NHS and institutional structuring, public health policy, education and training, and continued professional development, across England and Wales.⁴² As the national settings were identified as a variable in the quantitative and qualitative research, it is necessary to analyse the significance of divergency regarding the implementation and execution of trisomy screening pathway. It is also necessary to consider whether divergency is significant in the context of delivering and securing consent, subject to stakeholder interests and values.

As illustrated by the first phase quantitative analysis, all parent groups, across England and Wales, expressed dissatisfaction for all key areas of consideration, pertaining to their experiences of decision-making and consent along the trisomy pathway.⁴³ Similarly, all HCP groups, across England and Wales, expressed dissatisfaction for all key areas of consideration, pertaining to systemic considerations and individual professional roles for securing consent, for trisomy screening.⁴⁴

⁴² Scott L. Greer and Alan Trench, ‘Intergovernmental relations and health in Great Britain after devolution’, (2010) 38 Policy and Politics 509, at 515.

⁴³ Parent Quantitative Findings, Section 4.3. While they all expressed dissatisfaction for all identified areas of consideration, there was a significant difference, between parents from England and Wales, on the provision of information and communication with HCPs; this may be related to trisomy screening’s staggered implementation.

⁴⁴ HCP Quantitative Findings, Section 4.4. While all HCP groups expressed dissatisfaction for all key areas of consideration, there was a significant difference between the responses of midwives, across England and Wales, on confidence supporting parents’ choices and informational needs, and on the training received on trisomy screening.

Relatedly, there were no identifiable distinctions or differences, following Thematic Analysis of the dataset for the parent qualitative study, regarding parents' experience of consent and decision-making for trisomy screening, across England and Wales. While the year of the trisomy pathway's implementation differed across England and Wales, in 2016 and 2018 respectively, this study suggests that its execution has been significantly similar, from the perspective of parent groups.

A study by Peckham *et al.* into devolution and patient choice policy, revealed that "while at the national policy level there appeared to be a substantial difference between countries, at an operational level and in the way choices were experienced by patients there was much less difference".⁴⁵ This was suggested to be a result of the ingrained systems NHS were required to operate, across the four nations.⁴⁶

Peckham *et al.* also found that while policy differences were generally explained in terms of maintaining distinctiveness, being articulated by the devolved governments as both spatially and politically, their findings suggest such differences do not necessarily materialise in practice.⁴⁷ They explain that this distinction between stated policy and practice has been noted in previous studies concerning policy implementation; however, the current study supported the premise that "implementation within a specific service may lead to similar practices between countries with different national policy frames, providing an insight into potential impacts of devolution within the UK".⁴⁸

Smith and Hellowell also argue that, despite early rhetoric pointing towards diverging health care systems between the four nations post-devolution, public health policy across the UK demonstrates a "degree of consistency".⁴⁹ In their study, these academics found that, drawing on analysis of textual sources (namely policy documents and literature for health care), approaches to healthcare provision and addressing public health problems were

⁴⁵ Stephen Peckham, Nicholas Mays, David Hughes, Marie Sanderson, Pauline Allen, Lindsay Prior, Vikki Entwistle, Andrew Thompson and Huw Davies, 'Devolution and Patient Choice: Policy Rhetoric versus Experience in Practice', (2012) 46 *Social Policy and Administration* 199, at 211.

⁴⁶ *Ibid*, at 214.

⁴⁷ *Ibid*, at 213.

⁴⁸ *Ibid*, at 213.

⁴⁹ Katherine Smith and Mark Hellowell, 'Beyond Rhetorical Differences: A Cohesive Account of Post-devolution Developments in the UK Health Policy', (2012) 46 *Social Policy and Administration* 178, 179.

“remarkably similar” across the UK.⁵⁰ UK-wide professional bodies will also exemplify the similarities between health systems, according to Peckham *et al.*⁵¹

However, analysis of the qualitative findings from the HCP study, revealed the potential impact of divergency on the provision of professional education and training, including continued professional development.⁵² Based on the findings of the qualitative study, it was clear that access to education and training on Patau’s and Edwards’ Syndromes, and on NIPT’s purpose for trisomy screening, was left wanting across English health boards.⁵³ This research also revealed that English HCPs felt they had not received adequate training on securing consent since the introduction of ES and PS, and NIPT technology.⁵⁴ Indeed, English Trusts had not updated their training since 2016, before the implementation of the trisomy pathway.⁵⁵

While all English and Welsh HCP groups, in the quantitative study, expressed their dissatisfaction for all key areas of consideration and interest, the impact of divergent approaches to education and training may be evident in light of the significant difference, between the responses of English and Welsh midwives, on confidence supporting parents’ informational needs and choices, and on the training received on consent and trisomy screening, since its implementation.⁵⁶ The statistical difference, in this study, may be explained by the divergent and staggered approach to the implementation of trisomy screening, across NHS Trusts in England and Wales. It may also be a result of divergent approaches and *responses* to education and training.⁵⁷ Indeed, these issues did not emerge as a prominent theme for professional staff in Wales, which may be indicative of a prompt response to education and training across Welsh health boards.

In their research into patient choice and devolution, Peckham *et al.* state that divergence is clear in both health policy, not only in relation to public health regulation and outcomes, but also professional training standards.⁵⁸ While this is a rather unsurprising finding, its

⁵⁰ *Ibid*, 179-180.

⁵¹ Peckham *et al.* (n45), 200-201.

⁵² HCP Qualitative Findings, Sections 6.2.1, 6.4.2, 6.8.1, and 6.8.2.

⁵³ HCP Qualitative Findings, Sections 6.4.2 and 6.8.2.

⁵⁴ *Ibid*.

⁵⁵ *Ibid*.

⁵⁶ HCP Quantitative Findings, Section 4.4.

⁵⁷ HCP Qualitative Findings, Section 6.4.1, 6.8.1 and 6.8.2.

⁵⁸ Peckham *et al.* (n45), at 201.

significance is potentially wide-reaching, particularly in relation to consistency and quality of education and training between nations.⁵⁹

The findings for the parent qualitative study did not allude to the significance of this divergence; however, in the parent quantitative study, while they all expressed dissatisfaction for all identified key areas of consideration, there was a significant difference, between parents from England and Wales, regarding their experiences of the provision of information and communication with HCPs.⁶⁰ While it is not possible to draw any definitive or conclusive remarks in this regard, as concluded above, it may be related to trisomy screening's staggered implementation, and divergent approaches to professional training and CPD.

Indeed, O'Dowd reflects on the risks of educational divergence, underlining that divergency in education and training may encourage discrepancies in national standards and quality assurance, across the NHS.⁶¹ This was reflected in the findings of this research, with midwives and sonographers demonstrating concern toward the quality of e-learning and training standards on NIPT, and on the introduction of Edwards' and Patau's Syndromes.⁶²

This issue is particularly significant, as the 'cross-border health arrangements between England and Wales report conveyed that the implementation and execution of healthcare policy "on one side of the border can have an impact on patients on the other".⁶³ The report recommended "better co-ordination and information-sharing between the healthcare services of the two countries, to ensure patients received the same quality of healthcare wherever they lived", and was deemed "especially important" considering policy divergence since devolution.⁶⁴

Greer and Trench explain that "it is not simple" to combine UK-wide professional regulation, with devolved quality control and clinical governance.⁶⁵ As quality control and clinical governance reportedly rely on professionalism, and professional regulation typically involves

⁵⁹ HCP Qualitative Findings, Sections 6.4.2 and 6.8.2.

⁶⁰ Parent Quantitative Findings, Section 4.3.

⁶¹ Adrian O'Dowd, 'Devolution in health policy is threatening unity of the NHS', (2008) 336 *British Medical Journal* 241.

⁶² HCP Quantitative Findings, Section 4.4, and HCP Qualitative Findings, Sections 6.3.4, 6.4.2, and 6.8.2.

⁶³ Cross-border health arrangements between England and Wales, House of Commons Welsh Affairs Committee, Third Report of Session 2014-15, 1-44, at 29.

⁶⁴ *Ibid*, at 29.

⁶⁵ Scott L. Greer and Alan Trench, 'Intergovernmental relations and health in Great Britain after devolution', (2010) 38 *Policy & Politics* 509, at 515.

ensuring professional conformity to quality standards, these factors mean that a process of convergence becomes logistically complicated in this regard.⁶⁶

However, health administrators and programme co-ordinators prefer a unified approach to education and training standards, ensuring that professionals, across England and Wales, are able to effectively navigate the quality standards set by the UK NSC on trisomy screening, and requisite legal and ethical standards, to secure consent.⁶⁷ Indeed, Jervis and Plowden underline that the Royal Colleges, and other professional organisations, historically valued their unified all-UK networks, and conveyed concern toward a possible fragmentation under devolution.⁶⁸ The Nuffield Trust Report on the ‘Impact of Devolution on the UK’s Health Services’, has long outlined the preference of professional bodies for conformity, in the flow of information and ideas, standards of clinical practice, training and education, and conditions of service.⁶⁹ These bodies were viewed as being a force for policy stability, commonality and consistency, pointing to the benefits of these characteristics on the provision of professional training, education and standards.⁷⁰ Indeed, convergence may be necessary to ensure that quality standards set by the UK NSC on trisomy screening, in conjunction with the considerations identified from *Montgomery* and specifically *Mordel*, are consistently upheld across England and Wales.

If convergence is necessary in this regard, as Katikireddi *et al.* note that, “public health professionals should be at the forefront in developing the health-focussed evidence-base for these areas, ready to advocate for the health needs of their populations when opportunities present themselves”.⁷¹

Peckham *et al.* also suggests that attention needs to be focussed on commonalities pertaining to how the NHS overcome issues and manage the roles of institutions, in the implementation of health policy.⁷² Greer and Rowland conclude that “what matters ... is that the health

⁶⁶ Ibid, at 515.

⁶⁷ Researcher attended meetings with FASP and ASW screening leads and programme co-ordinators on the matter of implementation and CPD for trisomy screening programmes, across England and Wales.

⁶⁸ Paul Jervis and William Plowden, ‘The impact of political devolution on the UK’s health services’, (2003) The Nuffield Trust Report of Project to Monitor Impact of Devolution on the UK’s Health Services, 1-91, at 59.

⁶⁹ Ibid, 59-60.

⁷⁰ Ibid, 60.

⁷¹ Srinivasa Vittal Katikireddi, Katherine E. Smith, David Stuckler, Martin McKee, ‘Devolution of power, revolution in public health?’, (2016) 39 Journal of Public Health 241, at 245.

⁷² Peckham *et al.* (n45), at 215.

systems and the values contained within survive and thrive under devolution ... it is the shared values and divergent ones be best pursued and promoted".⁷³

⁷³ Scott L. Greer and David Rowland, *'Devolving Policy, Diverging Values? The Values of the United Kingdom's National Health Services'*, (2007) The Nuffield Trust, 1-104, at 94.

7.2.0 Provision of Information on Trisomy Screening: Situating Professional Duty

Sonographers are unclear as to whether their duty of care extends to the provision of information on trisomy screening.⁷⁴ Jay J, in *Mordel*, stated that: *'I appreciate that sonographers are busy, are working under time-pressures and that their lists are full, but I am driven to conclude she (sonographer) should have done more to lay the ground properly'*.⁷⁵ However, Jay J did not clearly outline what 'laying the ground properly' meant, or how this expectation could be *reasonably* implemented into existing systems for securing consent.

Inevitably, the opinion of the expert witnesses conflicted significantly and were diametrically opposed in this regard. The Defendant's expert witness outlined that, *'it was not the sonographer's role to re-counsel the patient or to provide further information'*, consistent with this study's findings.⁷⁶ On the other hand, the Claimant's expert witnesses stated that the sonographer should have asked a series of questions before performing the scan, pertaining primarily to information on the scan's purpose: how the information from the scan, together with a blood test, will provide a risk for DS.⁷⁷ Jay J preferred the latter's interpretation of the sonographer's duty, however this does not provide a clear explanation of the sonographer's duty, in terms of the information they are required to review with parents before obtaining consent, and whether this could be reasonably executed under established interdisciplinary systems.

As it stands, it is impractical for sonographers to review all the information with the parents at the examination appointment.⁷⁸ An opportunity arose here to isolate the issue of how the role and individual duty of the HCP could reasonably fit within the broader system and framework for securing parent consent, taking into account the unique challenges those working in the field of ultrasonography frequently encounter: primarily time pressures to perform the ultrasound within the allocated appointment. Furthermore, by delineating the individual HCP's role within the system, the court would have identified a troubling contradiction: introducing information to parents for the first time on the day of the scan

⁷⁴ HCP Quantitative Findings, Section 4.4, HCP Qualitative Findings, Section 6.3.1.

⁷⁵ *Mordel*, (n14), 92.

⁷⁶ *Ibid*, 70.

⁷⁷ *Ibid*, 93.

⁷⁸ Pilnick A.M, Fraser D.M, and James D.K, 'Presenting and discussing nuchal translucency screening for fetal abnormality in the UK', (2004) 20 *Midwifery*, 82.

would not adhere to its *own* recommendation, and current practice guidelines, to allow at least *twenty-four hours* to lapse for parents to reflect on the information, before providing their consent.⁷⁹

While Jay J favoured the Claimant's expert witnesses' interpretation of the sonographer's duty, he understood that this "modicum of exploration" – that he held the sonographer was duty-bound to perform – could lead to a series of questions being asked by the parents: something he agreed sonographer could be unqualified to answer.⁸⁰ Jay J stated that, "*a sonographer is not a midwife, and is not trained to provide advice*", however suggested that the 'obvious' solution would be to "*refer the patient for further consultation with the midwife, preferable on the same day*".⁸¹ Despite this being an 'obvious' solution to Jay J, this study identified a series of potential difficulties sonographer's may experience in terms of effectively discharging their duty in this regard.⁸²

Parents' perception of the HCP's role impacts their readiness and preparedness to ask questions.⁸³ As sonographers are not perceived, by parents, as being ready to answer their questions, this could hinder parents from asking questions at the examination appointment.⁸⁴ Furthermore, sonographers do not receive questions from parents, as the provision of information is performed and reviewed by the midwife.⁸⁵ Thus, this raises the question whether the parent's perception of the HCP's role is currently depriving them of the opportunity to obtain further information.⁸⁶ It also points to the dysfunctionality of Jay J's proposed system in this regard.

This decision also suggests that the standard for information disclosure and functionality of the system to obtain full information, in part, rests on the parent's asking questions. *Montgomery* endorsed the position in *Wyatt*, that concluded it is illogical and unreasonable to invoke the HCP's duty to disclose information, on the basis that the patient asks specific

⁷⁹ Mordel (n14), 88.

⁸⁰ Ibid, 94.

⁸¹ Ibid.

⁸² HCP Qualitative Findings, Section 6.3.

⁸³ Parent Qualitative Findings, Section 5.2.9.

⁸⁴ Parent Qualitative Findings, Section 5.2.9.

⁸⁵ HCP Qualitative Findings, Section 6.3.1.

⁸⁶ McCourt C, '*Supporting choice and control? Communication and interaction between midwives and women at the antenatal booking visit*', (2006) 62 Social Science & Medicine, 986.

questions, “... *there is something unreal about placing the onus of asking upon a patient who may not know there is anything to ask about*”.⁸⁷

The functionality of the proposed system is also dependent on parents’ confidence.⁸⁸ Indeed, parents often lack the confidence required to ask questions at clinical appointments.⁸⁹ *Montgomery* found that:

*“an approach which requires the patient to question the doctor disregards the social and psychological realities of the relationship between a patient and her doctor, whether in the time-pressured setting of a GP’s surgery, or in the setting of a hospital ... few patients do not feel intimidated or inhibited to some degree.”*⁹⁰

To send the parent away for further counselling with the midwife – on the same day as the examination appointment – would not be systemically reasonable or logical, as this would run the risk of missing the opportunity to perform the ultrasound examination. While many would argue that the parent can merely reschedule the scan for a later date, the next opportunity would be at 16+ weeks using quadruple screening, which provides a less accurate screening result.

It is also important to note that Jay J’s proposed system works on the assumption that both midwives and sonographers have similar, or the same, shift patterns. Jay J’s system would not be reasonable or logical, considering that midwives and sonographers will typically have differing and unsynchronised shift patterns.⁹¹ This was a factor considered in *Campbell v Border Health Board* on the functionality of care management systems.⁹² Thus, this again raises the question whether the judiciary are best placed to decide on matters of multidisciplinary practice, alluding to *Bolam*’s usefulness in this regard.

⁸⁷ *Wyatt v Curtis* (2003) EWCA Civ 1779, 19 and *Montgomery*, 58.

⁸⁸ Wätterbjörk, I., Blomberg, K., Nilsson, K., & Sahlberg-Blom, E, “Decision-making process of prenatal screening described by pregnant women and their partners”, *Health Expectations: An International Journal of Public Participation in Health Care & Health Policy*, 2015, 18(5), 1582–1592.

⁸⁹ Parent Qualitative Findings, Section 5.2.9.

⁹⁰ *Montgomery* (n1), 58.

⁹¹ HCP Qualitative Findings, Section 6.3.1.

⁹² *Campbell v Border Health Board* (2011) CSOH 73.

7.2 Provision of Information for Trisomy Screening: Key Considerations

7.2.1 Parents' Failure to Read Information

Transparency and openness, between parents and professionals, are key aspirations from *Montgomery* and clinical guidelines to empower the desired shared decision-making model of consent. However, shared decision-making may become impaired by parents falsely confirming that they have read the information materials when, in fact, they have not.⁹³ The significance of parents' failure to read the NHS materials was also highlighted in *Mordel*; however, this issue was not effectively addressed by Jay J, in terms of its impact on the HCP's ability to secure consent.

Mordel identified that "checking" the parents have read the information provides the foundation for informed decision-making. However, parents' preference for face-to-face counselling on screening correlates to an innate reluctance to read the information packs, provided by the midwife, at the booking appointment.⁹⁴ With parents exhibiting a reluctance to read the information, preferring to receive the information at the consultations, this places substantial pressure on HCPs to review 'sufficient information' on trisomy screening at the scheduled appointments.⁹⁵

However, Jackson reminds us that information materials and booklets, on their own, are insufficient, and good practice – under the GMC guidance – mandates that this information is accompanied by a supported dialogue.⁹⁶ Indeed, *Thefaut* endorsed the position in *Montgomery*, that the *modus operandi* of communication, following the delivery of paper-based materials, is supporting dialogue.⁹⁷ Nevertheless, it is apparent from the findings that parents could be over reliant on the HCPs 'supporting dialogue' as a means to provide *all* the information required at the appointments, posing challenges for HCPs to discharge their duty in this regard.⁹⁸

⁹³ Parent Qualitative Findings, Section 5.2.1.

⁹⁴ Parent Qualitative Findings, Sections 5.2.1 and 5.2.2.

⁹⁵ Parent Qualitative Findings, Sections 5.2.1 and 5.2.2.

⁹⁶ Emily Jackson, "Challenging the Comparison in Montgomery Between Patients and 'Consumers Exercising Choices', (2021) 00 Medical Law Review, 6.

⁹⁷ *Thefaut v Johnson* (2017) EWHC 497 (QB), 58.

⁹⁸ Parent Qualitative Findings, Section 5.2.2.

HCPs are unable to force parents to read information. *Montgomery* underlined that, “A person can of course decide that she does not wish to be informed of risks of injury (just as a person may choose to ignore the information leaflet enclosed with her medicine) ...”.⁹⁹ Despite referring to the therapeutic exception here, this consideration is significant, as it may indicate that the court acknowledges not only a right not-to-know, but accepts that patients may choose not to read the information materials, associated with a proposed course of treatment or intervention. This contrasts with the perspective of Jay J in *Mordel*, who opined that reasonable and responsible practice would require HCPs to check that the parents have received and *read* the information booklets, before engaging with screening. This begs the question whether HCPs would be in breach of this duty if parents reveal they have not read the information, but proceed with screening. It is safe to conclude that common law has left HCPs in a catch-22, as the judgment in *Mordel* effectively contradicts that in *Montgomery*.

The findings of this study also throw into question the requisite steps HCPs must take where parents have intentionally misled HCPs. Indeed, both sonographers and midwives may be caught between a rock and a hard place, as parent dishonesty will foreseeably place HCPs in a position where they are not reasonably able to effectively discharge their duty. In *Mordel*, despite the Claimant admitting to not having read the information packs before attending the booking appointment, Jay J suggested that there is *still* a remaining expectation to ‘lay the ground properly’ before securing parents’ consent.¹⁰⁰ Indeed, this abstract concept of ‘laying the ground’ remains a source of speculation, and current guidelines also do not provide a steer on issues of dishonesty, or the challenge this may pose to securing parent consent.

7.2.2 Individualising Information on Trisomy Screening

The need for transparency is also pertinent to the HCP’s ability to tailor information to the particular patient’s needs, consistent with the expectation in *Montgomery* and clinical guidelines.¹⁰¹ Indeed, a growing body of medical research, across many areas of practice, champion the implementation of systems, whereby HCPs are to individualise information effectively and efficiently to the particular patient, before securing consent to treatment.

⁹⁹ *Montgomery*, (n1), 85.

¹⁰⁰ *Mordel*, (n12), 92.

¹⁰¹ Parent Qualitative Findings, Section 5.2.3; see also *Al Hamwi v Johnson* (2005) EWHC 206 (QB).

While *Montgomery* and the GMC demonstrate their distaste for the delivery of generalised information on proposed methods of treatment, openness and honesty are both core elements to effectively achieve this.¹⁰² However, parent dishonesty and a lack of transparency may interfere with the HCP's ability to engage into the process of individualisation of information.¹⁰³ A consequence of parents' failure to review the information before counselling, means that both HCPs and parents are unable to determine what information they consider relevant or material, to exercise an informed choice.¹⁰⁴ As a result, HCPs may feel obliged to revert to presenting a generalised version of the information and, as a consequence, risk overwhelming parents with the information at consultations.¹⁰⁵

Considering maternity units' time-pressured and resource-limited systems in mind, constructing a predetermined set of questions may ostensibly provide solution; however, this method must be approached cautiously. This suggestion is a common and plausible technique to tailor the informational subject to parents' needs: a method that is currently being trailed in maternity units across England and Wales. Nevertheless, ultimately, adoption of this method risks a process of prioritisation or filtration of information, subject to what HCPs consider relevant to the particular patient.

Indeed, one could conclude that misuse or ineffective execution of this method, may be akin to a *Bolam* standard of care, as this standard would ultimately become self-regulated. As it stands, the questions are constructed by HCPs, for the use of HCPs. Thus, a risk exists of falling into a mechanical 'tick-box' exercise, which would not be sufficiently bespoke or conducive to the patient-centric model of care, endorsed in *Montgomery*'.¹⁰⁶

The decision in *Mordel* also begs the question whether *both* the midwife and sonographer are required to tailor the information, subject to the particular patient, and if so, how this could

¹⁰² Parent Qualitative Findings, Section 5.2.3.

¹⁰³ HCP Qualitative Findings, Section 6.3.2 and Parent Qualitative Findings

¹⁰⁴ Parent Qualitative Findings, Section 5.2.3 and HCP Qualitative Findings, Section 6.3.2 and Parent Qualitative Findings; see also, F. E. Carroll, A. Owen-Smith, A. Shaw, and A. A. Montgomery, 'A qualitative investigation of the decision-making process of couples considering prenatal screening for Down syndrome,' (2012) 32 *Prenatal Diagnosis*, 57.

¹⁰⁵ Parent Quantitative Findings, Section 4.3 and Parent Qualitative Findings, Section 5.2.3, and HCP Qualitative Findings, Section 6.3.2; and Tanja Schlaikjaer Hartwig and others, 'High risk – What's next? A survey study on decisional conflict, regret, and satisfaction among high-risk pregnant women making choices about further prenatal testing for fetal aneuploidy', (2019) 39 *Prenat Diagn*, 635.

¹⁰⁶ Judy M Laing, 'Delivering informed consent post-Montgomery: Implications for medical practice and professionalism', (2017) 33 *Journal of Professional Negligence*, 128.

be reasonably achieved in clinical practice. Reasonable practice, in this context, must require consideration of the broader systems in place on the provision of information between the relevant HCPs, to ensure appropriateness of approach and consistency of execution. The design of this method may also benefit from facilitating NHS patient evaluation group discussions on consent for trisomy screening; a growing trend among many research and development units in the health service, as a means of quality assurance.

The COVID pandemic saw a surge in popularity for the NHS-created pregnancy online applications as a means to obtain information during lockdown.¹⁰⁷ This study, along with many others, revealed that parents are typically reluctant to read lengthy paper-based materials, preferring to access summarised and concise information packages, as they are convenient and user-friendly.¹⁰⁸ With the surge in popularity for applications on handheld electronic devices, it may be that the NHS could capitalise on this trend by delivering the information in this format.¹⁰⁹ Online portals and user accounts could also provide a means for HCPs to verify whether the information has been accessed by the parents before attending clinic.¹¹⁰ This method is currently being trialled under Trusts in England and Scotland, in the neonatal services; however, its popularity is extending to maternity services.¹¹¹ The RCOG have stated that they aim to replace all paper-based materials with online information, within the next five years; however, a move to online information – and the use of applications - may give rise to unprecedented challenges surrounding the personalisation and individualisation of information.¹¹² Indeed, this bears the risk of providing generic information to parents, in an attempt to cater to the ‘majority’. To avoid this, the recommendation could be presented to

¹⁰⁷ Parent Qualitative Findings, Section 5.2.4 and HCP Qualitative Findings, Section 6.3.3.

¹⁰⁸ Parent Qualitative Findings, Section 5.2.4; see also, Owen Barr and Heather Skirton, ‘*Informed decision making regarding antenatal screening for fetal abnormality in the United Kingdom: A qualitative study of parents and professionals*’, (2013) 15 *Nursing and Health Sciences*, 318, 325.

¹⁰⁹ Parents Qualitative Findings, Section 5.2.4.

¹¹⁰ This was also an idea presented by Antenatal Screening Wales in a meeting the researcher attended in June 2021.

¹¹¹ BadgerNet is the Maternity Information System (MIS) trialled by some Trusts across England and Scotland, and it is used as a clinical tool. This online app allows midwives to record and monitor the pregnancy, share information on screening, share screening results, and provides a communication tool for parents in a purely electronic form. Its recent aim is to replace paper-based information, meaning that parents (who are under a Trust that have opted in to use it) have the option to receive all their information electronically. It is currently being widely used in *neonatal* services across England, Wales and Scotland. However, it is now being used amongst *maternity* services in Scotland, and *some* Trusts in England.

¹¹² A L Kratovil and W A Julion, “*Health-care provider communication with expectant parents during a prenatal diagnosis: an integrative review*”, (2017) 37 *Journal of Perinatology*, 2.

patient review groups, before it is implemented into clinical practice, for constructive feedback.

7.2.3 Quality and Validity of Online Information

While COVID brought to bear the benefits of using online information and pregnancy applications, their quality and validity must be questioned. Many parents use online applications and websites as *primary* sources of information on trisomy screening.¹¹³ Indeed, the growing popularity of pregnancy applications, online support groups and pregnancy-dedicated websites, primarily due to factors of convenience and time, has been noted in many previous studies.¹¹⁴ However, a significant number of online sources that provide pregnancy information, particularly on trisomy screening, are unvalidated, unreliable, or misleading.

This study revealed a concerning preference by parents to obtain information from online resources, which varied significantly in reliability and validity, rather than verified NHS booklets and materials.¹¹⁵ *Montgomery* forewarned of this societal change, stating that, “*it has become far easier, and far more common, for members of the public to obtain information about symptoms, investigations, treatment options, risks and side-effects via such media as the internet (where, although the information available is of variable quality, reliable sources of information can readily be found) ...*”¹¹⁶ Indeed, Mrs Mordel openly admitted, in her statement, that she preferred to refer to ‘Youtube’ as a source of information, rather than utilising the NHS verified materials.

Despite having obvious implications on Mrs Mordel’s ability to inform her decision, Jay J failed to address this in isolation, and its potential challenges for HCPs to secure consent. In light of this, clinical guidelines may be required to outline HCPs’ duty in circumstances where parents supplement NHS materials with online sources, and the risk this poses on parents’ ability to exercise an informed choice.¹¹⁷ While providing parents with a list of verified online sources may ostensibly be the clear, albeit prosaic solution, this could contribute to the infantilisation

¹¹³ Parent Qualitative Findings, Section 5.2.4.

¹¹⁴ Parent Qualitative Findings, Section 5.2.4; see also, A L Kratovil and W A Julion (n112), 2.

¹¹⁵ HCP Qualitative Findings, section 6.3.3 and Parent Qualitative Findings, Section 5.2.4.

¹¹⁶ *Montgomery*, (n1), 76.

¹¹⁷ Mercer M.B, Agatista P.K, Farrell R.M, ‘What patients are reading about non-invasive prenatal testing: an evaluation of Internet content and implications for patient-centred care’, (2014) 34 *Prenatal Diagnosis*, 986; see also, Lagan B.M, Sinclair M, Kernohan W.G, ‘A web-based survey of midwives’ perceptions of women using the Internet in pregnancy: A global phenomenon’, (2011) 27, *Midwifery*, 273.

of care. What is 'reasonable' may require HCPs to explain, at first contact or the booking appointment, the risks of supplementing the verified NHS materials for online sources on parents' ability to make informed decisions.

7.2.4 Appetite for Information

Another factor HCPs should also take into consideration, during the process of individualising information, are parents' differing levels of experience of trisomy screening.¹¹⁸ Parents with past experience of screening feel more confident with the process, and their expectation of the requisite care pathway.¹¹⁹ While this conclusion is relatively unsurprising, confidence may lead experienced parents to omit consideration of the information packs or materials, as they have been through the process on previous occasions.¹²⁰ Indeed, experienced parents do not feel the need to read the information booklets before attending counselling, as they have previous experience of screening.¹²¹ This revelation may pose significant hinderances to their ability to make informed choices, under the *amended* trisomy pathway.

While parents will become accustomed to the amendments over time, at present, the introduction of new methods of screening and additional trisomies, mean that parents risk overlooking imperative considerations, subsequently interfering with their ability to exercise an informed decision. There has been a paradigm shift in terms of the implications of screening since the introduction of ES and PS, and therefore the HCP's duty may now include notifying parents of the recent amendments to the pathway. Acknowledging the varying appetites for information is crucial, possibly requiring adaptations to the information packs to include prominent warnings, highlighting any changes to the pathway since their previous experience.

7.2.5 Fragmentation of the Trisomy Pathway

Before parents are able to make an informed decision on screening for the common trisomies, FASP and ASW outline that information must be provided to parents on DS, ES and PS. While trying not to overcomplicate matters, it may be necessary for programme directors to

¹¹⁸ Parent Qualitative Findings, Section 5.2.5.

¹¹⁹ Parent Qualitative Findings, Section 5.2.5.

¹²⁰ Parent Qualitative Findings, Section 5.2.5.

¹²¹ Parent Qualitative Findings, Section 5.2.5.

fragment the current 'trisomy' system/model, subject to the individual phenotype: DS, ES and PS. Under a generalised 'trisomy' model of screening, a risk exists of underappreciating the uniqueness of each condition, in terms of their health characteristics and subsequent decision-making for parents, in the event of a high-risk or positive diagnosis.¹²²

A fragmentation of the trisomy pathway is justified on the basis of the parents' apparent lack of awareness for ES and PS, before undertaking trisomy screening.¹²³ The information is material for the purposes of discharging the HCP's duty for information disclosure; therefore, it would harm parent decision-making and reproductive autonomy if they were to overlook, or fail to be sufficiently directed to, the information on ES and PS. With all parent groups reporting their lack of awareness toward the information on these conditions, the cause must be urgently considered.¹²⁴

Parents' lack of awareness could rest on their failure to read NHS materials, and/or preferring to access information online, leading to a lack of awareness of ES and PS. Considering the recent introduction of ES and PS onto the screening pathway, many of the non-NHS verified websites – commonly referred to during the research interviews – may not demonstrate this change.¹²⁵ A quick search on a very popular pregnancy application used by parents, across England and Wales, revealed that the 'trisomy pathway' is still being called 'Down's Screening', completely omitting information on ES and PS. With parents revealing that they had little, if any, awareness of ES and PS before and *after* leaving the pathway, having consented to screen for these conditions, highlights an urgent need for review.¹²⁶

Another probable cause may extend from the fact that midwives typically refer to the trisomy pathway as 'the down's screening', at clinical appointments.¹²⁷ While this issue may be a matter of semantics – and a consequence of language, which they have become accustomed to following years of education and practical experience – HCPs must be mindful to the importance of using accurate terminology.¹²⁸ Indeed, while acknowledging the position in *Wyatt*, which correctly addressed that the duty for information disclosure should not rest on

¹²² HCP Qualitative Findings, Section 6.3.4 and Parent Qualitative Findings, Section 5.2.6.

¹²³ HCP Qualitative Findings, Section 6.3.4 and Parent Qualitative Findings, Section 5.2.6.

¹²⁴ Parent Qualitative Findings, Section 5.2.6.

¹²⁵ HCP Qualitative Findings, Section 6.3.3.

¹²⁶ Parent Qualitative Findings, Section 5.2.6.

¹²⁷ HCP Qualitative Findings, Section 6.3.4.

¹²⁸ Anna Marsh, 'The importance of language in maternity services', (2019) 27 *British Journal of Midwifery*, 1.

the patient's desire to ask questions, utilising the term 'trisomy screening' is more likely to elicit questions from parents, due to their unfamiliarity with this term.

The fragmentation of the trisomy pathway may benefit both HCPs and parents in this regard. As ES and PS are ultimately 'new' conditions, in the context of screening, a fragmentation could operate as a means to isolate the information for parents, requiring them to dedicate time and focus upon the unique implications and choices they could be presented with, in the event of a high-risk or positive diagnosis.¹²⁹ Furthermore, with HCPs revealing that they are less confident supporting parents' informational needs on ES and PS compared to DS, due to their unfamiliarity with the conditions and a lack of training, a fragmentation could encourage the introduction of bespoke professional training programmes, tailored to the condition's unique characteristics and implications on parent decision-making.¹³⁰ While this overhaul would inevitably warrant an entire restructure of the trisomy pathway, this would arguably raise the awareness of parents to ES and PS, and may increase the confidence of HCPs to disclose accurate information on their unique health characteristics, aetiology and prognosis.¹³¹

7.2.6 Supporting Provision of Information on Practicalities of Screening and Testing

Under both the FASP and ASW screening programme policy guidelines, HCPs are required to review the information on *all* current routine methods of screening and testing, before obtaining parents' consent. However, the reluctance of parents to read the information provided on the methods of screening and testing, is inevitably hindering their ability to distinguish between the practicalities of *screening* and invasive testing.¹³² As screening is often a novel and unfamiliar experience for parents, it may be that parents fail to read the information on the methods of testing, as a means to minimise stress and anxiety.¹³³

This may also correlate to the considerable levels of trust parents exhibited towards HCPs.¹³⁴ Indeed, Lords Kerr and Reed, in *Montgomery*, suggested, "... *that some patients would rather*

¹²⁹ Parent Qualitative Findings, Section 5.2.6.

¹³⁰ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.3.4.

¹³¹ Parent Qualitative Findings, Section 5.2.6.

¹³² Parent Quantitative Findings, Section 4.3 and Parent Qualitative Findings, Section 5.2.7.

¹³³ Parent Qualitative Findings, Section 5.2.7; see also, Samantha Thomas, Kate O'Loughlin and Jillian Clarke, 'Sonographers' communication in obstetrics: Challenges to their professional role and practice in Australia' (2020) 23 Australasian Journal of Ultrasound in Medicine, 129.

¹³⁴ Parent Qualitative Findings, Section 5.2.7.

trust their doctor than be informed of all the ways in which their treatment might go wrong ...".¹³⁵ While a relationship of trust and confidence is required under the desired GMC HCP-patient partnership model of care, a risk exists of over-reliance and dependency on the HCPs to manage parents' care, which could encourage the existence of infantilisation and paternalism.

As will be discussed later in the chapter, it is human nature to deflect potentially stress-inducing information, and this is no different in pregnancy care.¹³⁶ It may be that discussion of the information on receiving a higher-risk result, and having to consider invasive testing that carries a risk of procedural miscarriage, induces said stress and anxiety, and that it is merely a coping mechanism to put this information to one side. Medical studies, in other areas of medicine, have long since identified this risk of "switching off" or "panicking" in the face of potentially distressing information, and suggest that more than one person attends clinical appointments to ensure that information is not overlooked.¹³⁷

Understanding that parents share responsibility, and must read the information on the practicalities of screening and testing, is paramount.¹³⁸ Indeed, the inability of parents to distinguish between the practicalities of screening and testing, may be due to the failure of HCPs to provide parents with the more detailed information on invasive testing at the first instance, as some medical studies suggest.¹³⁹ However, given the severity of the risk, the gravity of this information on decision-making, and the likelihood of facing this option being one in five parents, this would arguably be a very rare occurrence in clinical practice.

It is fundamental to informed choice that HCPs accurately disclose information on the practicalities of screening and testing.¹⁴⁰ Reasonable practice, following the decision in *Mordel*, mandates that HCPs are required to discuss *all* possible methods of screening and testing, before securing parents' consent.¹⁴¹ ASW and FASP also underline that the consequences of screening and testing – in this context, the risk posed by invasive testing –

¹³⁵ Montgomery (n1), 92.

¹³⁶ Parent Qualitative Findings, Section 5.2.7.

¹³⁷ Parent Qualitative Findings, Section 5.2.7; see also, Anna Marsh, (n128), 1.

¹³⁸ Karuna RM van der Meij and others, 'Routinisation of prenatal screening with the non-invasive prenatal test: pregnant women's perspectives', (2021) 2 European Journal of Human Genetics, 1.

¹³⁹ Parent Qualitative Findings, Section 5.2.7; see also, John et al. (n12), 4.

¹⁴⁰ Ibid.

¹⁴¹ Mordel (n14), 5.

must be clearly and fully discussed before obtaining informed consent, to safeguard reproductive autonomy.

7.2.7 Systemic Disconnect and Information Overload

The examples identified of the challenges HCPs may encounter when discharging their duty for information disclosure, such as overlooking information on ES and PS, and failing to distinguish between screening and testing, may also be exacerbated by ‘information overload’. Parents are being overwhelmed with information early in pregnancy, which may harm their ability to absorb and reflect on their choices before deciding to screen.¹⁴²

The findings suggest that accountability and blame, for being overwhelmed and bombarded with information, commonly rests on the individual HCPs, rather than any systemic accountability, under which HCPs are mandated to practice.¹⁴³ With an ever-increasing amount of information HCPs are duty-bound to review, in an ever-decreasing timeframe, this will undoubtedly pose unprecedented challenges for HCPs in facilitating informed and shared decision-making.¹⁴⁴ Overwhelming parents with information may also be suggestive of defensive practices, in this regard.¹⁴⁵ Indeed, as Steen *et al.* opine, “prenatal counselling has become increasingly demanding due to the more complex information resulting from technological progress”.¹⁴⁶ *Montgomery* established that bombarding the patient with information does not discharge the HCP’s duty for information disclosure¹⁴⁷; however, the decision offered little in terms of explanation as to why a HCP may feel the need to disclose information in this way, and how to effectively mitigate against this risk.

The Nuffield Council report foresaw that the rapidly developing landscape of prenatal screening will inevitably put additional strain onto existing systems, within maternity units, in the context of information disclosure and securing parent consent.¹⁴⁸ Agreeably, the

¹⁴² Parent Qualitative Findings, Section 5.2.8; see also, Beulen L and others, ‘The effect of a decision aid on informed decision-making in the era of non-invasive prenatal testing: a randomised controlled trial’ (2016) 24 *European Journal of Human Genetics*, 1409.

¹⁴³ Parent Qualitative Findings, Section 5.2.8.

¹⁴⁴ HCP Qualitative Findings, Section 6.3.6.

¹⁴⁵ HCP Qualitative Findings, Section 6.3.6.

¹⁴⁶ Sanne L. van der Steen and others, ‘Offering a choice between NIPT and invasive PND in prenatal genetic counselling: the impact of clinician characteristics on patients’ test uptake’, (2019) 27 *European Journal of Human Genetics* 235.

¹⁴⁷ *Montgomery* (n1), 90.

¹⁴⁸ Nuffield Council Report, 23.

expectation on parents to receive, retain and process the information on trisomy screening, alongside their other pregnancy care information, is wholly unrealistic.¹⁴⁹ In addition, retention and processing of this information, in this way, demands increasingly high levels of medical literacy from parents.¹⁵⁰ Even *professionals* with an expert level of medical literacy, including sonographers and midwives, experience some difficulty navigating the information along the trisomy pathway.¹⁵¹

Thus, it is no wonder that parents felt aggrieved and frustrated by the demand to review said information.¹⁵² It is no wonder, either, that parents naturally hold the individual midwife or sonographer accountable for being overwhelmed, at this stage.¹⁵³ However, it is only logical to consider the broader system in which HCPs are expected to operate, and whether this is indeed ‘reasonable’.

This reported experience of being overwhelmed with information, from the perspective of both parents and HCPs, may be indicative of broader systemic disconnect.¹⁵⁴ Indeed, the risk of overwhelming parents could be a consequence of the ambiguity surrounding the assessment of what information HCPs must individually cover, at the various stages of pregnancy.¹⁵⁵ A conflict can be observed from the study’s findings, between midwives and sonographers, on what information they are duty-bound to cover.¹⁵⁶ With both HCPs revealing they work with a looming fear of breaching their duty for information disclosure, primarily due to existing ambiguity, it is plausible that HCPs feel that providing parents with as much information as possible is the only ‘reasonable’ solution, indicative of defence practices.¹⁵⁷ However, defensive practices are deeply problematic, as it undermines patient

¹⁴⁹ Parent Qualitative Findings, Section 5.2.8. Regardless of educational background and perceived ‘intelligence’ levels – thankfully seldom utilised as a factor in medical research to draw damaging and divisive conclusions about the parent’s standard of education and their ability to retain and process information.

¹⁵⁰ Anna Marsh, (n128), 1.

¹⁵¹ HCP Qualitative Findings, Section 6.3.6.

¹⁵² Parent Qualitative Findings, Section 5.2.8.

¹⁵³ Parent Qualitative Findings, Section 5.2.8.

¹⁵⁴ HCP Qualitative Findings, Section 6.3.6.

¹⁵⁵ HCP Qualitative Findings, Section 6.3.1.

¹⁵⁶ HCP Qualitative Findings, Section 6.3.1.

¹⁵⁷ HCP Qualitative Findings, Section 6.3.6.

autonomy, violates fiduciary obligations, exposes patients to harm without the benefit, and may also encourage feelings of distrust toward the profession.¹⁵⁸

Jay J stated that *Montgomery's* relevance to the decision, in *Mordel*, was to outline HCPs' duty to provide "sufficient information to make an informed decision"; however, this definition of 'sufficient' remained unaddressed in the judgment.¹⁵⁹ Although clinical guidelines provide a general steer, this definition of 'sufficient' is subjective, as it stands, leaving this open to broad interpretations by HCPs. Agreeably, NHS could not function if it was required to implement fail-safes; however, it is likely that accountability will fall upon individual HCPs, rather than addressing any broader systemic failings, for overwhelming parents with information.

The implementation of a system, to appropriately delegate or share responsibility between the midwife and sonographer, could mitigate the risk of overwhelming parents with information. This issue was also raised in *Deriche v Ealing Hospital Trust*. In this case, the court threw into question how far the HCPs could rely upon information provided during counselling at earlier appointments.¹⁶⁰ The HCP relied upon the fact the patient received information from previous appointments, as a means to avoid overwhelming them with information. The court's focus rested on the individual HCPs, outlining that they should not simply rely upon the notes of the earlier counselling, but instead take the necessary steps to 'check' the patient is in receipt of the relevant information from previous consultations, before continuing with treatment. However, the decision placed undue focus on the conduct of the HCPs, omitting the necessary consideration of the system they were required to practice, and how to avoid this risk in the future.

A 'reasonable' system could seek to enhance interdisciplinary roles to better navigate the recommended requirements by Jay J, in *Mordel*: that the midwife provides an 'informed offer', and additional counselling pertaining to the information is performed by the sonographer on the day of the scan. This interplay between both sonographers and midwives could also lead to effectively identifying possible systemic fragilities and disconnects, which would allow individual HCPs to assess the risk of overwhelming parents with information, and

¹⁵⁸ Johan Christiaan Bester, 'Defensive practice is indefensible: how defensive medicine runs counter to the ethical and professional obligations of clinicians', (2020) 23 *Medicine, Health Care and Philosophy* 413, 420.

¹⁵⁹ *Mordel* (n14) 18.

¹⁶⁰ *Deriche v Ealing Hospital NHS Trust* (2003) EWHC 3104.

how this could be mitigated under an efficient and synthesised system. Ultimately, a degree of systemic objectivity is required for HCPs to sufficiently navigate this duty. Without clearly assessing what is 'reasonable', discharging the duty to deliver 'sufficient information' remains speculative for both midwives and sonographers.¹⁶¹

¹⁶¹ Sanne L. van der Steen (n146), 243.

7.3.0 Supporting Parent Understanding: Situating Professional Duty

Ambiguity currently surrounds HCPs' duty to support parent understanding for trisomy screening.¹⁶² Sonographers are currently unclear as to whether their duty extends to support parent understanding.¹⁶³ Jay J, in the operative portion of his judgment, emphasised that it was the sonographer's duty to satisfy herself, by 'brief questioning', that the patient understood the essential elements and purposes of screening, at the examination appointment.¹⁶⁴ However, the sonographer's established duty does not extend to ensuring that the patient understands the various components of screening, as they have not received the requisite training, and are restricted by resource and time constraints.¹⁶⁵

In his judgment, Jay J had previously imposed a rather convoluted duty that sonographers are required to perform when exploring parents' level of understanding: "*gentle exploration is required of the patient's state of mind, conducted for the limited and specific purpose of checking that she understands what is entailed*"; however, he failed to define a 'gentle exploration'.¹⁶⁶ Indeed, a lack of guidance is currently available on the most effective method to explore parent understanding without appearing to undermine the parent's decision-making, consistent with the Defendant's argument in *Mordel*.

Sonographers are also unclear of their duty in situations where parents do not understand the information.¹⁶⁷ Referring to *Mordel*, FASP – section 3 on 'personal choice' – states that "... there needs to be a *process* to allow women to have further discussion with a HCP when they attend the ultrasound scan but do not understand its purpose ... this process should not impact on the woman's eligibility for screening"; however, the guidelines left the 'process' ill-defined.¹⁶⁸ While this is inevitably subject to the discretion of local guidelines, consistency is required at national level to effectively situate a 'reasonable' and workable system, for securing parent consent. What is clear, however, is that FASP have not implemented the recommendation of Jay J, to send the parent back to the midwife for further counselling on

¹⁶² HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.4.1.

¹⁶³ HCP Qualitative Findings, Section 6.4.1.

¹⁶⁴ *Mordel* (n14), 89.

¹⁶⁵ HCP Qualitative Findings, Section 6.4.1.

¹⁶⁶ *Mordel* (n14), 85.

¹⁶⁷ HCP Qualitative Findings, Section 6.4.1.

¹⁶⁸ FASP (2021) section 3.

the same day as the examination appointment, possibly demonstrating the impracticality of Jay J's 'reasonable' system.¹⁶⁹

As the law stands, effective execution of this requirement is at the discretion of HCPs.¹⁷⁰ While being able to exercise discretion over this requirement could allow for greater flexibility and service-user autonomy, a foreseeable side-effect to this is the potential for a wide-range of interpretations and outcomes.¹⁷¹ A risk also exists of misinterpretation or improper execution of this requirement, borne from the court's lack of clear guidance in this regard.¹⁷² Thus, the process of a 'gentle exploration', and instruction as how this should be executed – particularly where parents demonstrate a lack of understanding – within existing systems, is left wanting.

An exploration into assessing what a 'reasonable' system may constitute, is required. While Jay J alluded to the time-pressures sonographers' experience in practice, his assessment of a 'reasonable system' demonstrated the court's limitations for deciding on such matters. With the time pressures of the examination appointment, an issue outlined by Jay J in the judgment, the current duty imposed by Jay J on sonographers could only be described as unreasonable and illogical.¹⁷³

What may be 'reasonable', in this context, would be to extend the role of midwife, to include involvement at the 12-week examination appointment.¹⁷⁴ Returning to the need for enhanced interprofessional practice, it may be that midwives can support sonographers in their duty during this period of transition.¹⁷⁵ This suggestion could also provide the desired consistency in provision of counselling and expertise, to support parent understanding.

Logic mandates that if sonographers were required to explore parent understanding before obtaining consent, their training *must* be brought in-line to that midwives receive on counselling. *Gottstein* highlighted that in the event of a breach of duty, due to inadequate

¹⁶⁹ Mordel (n14) 94.

¹⁷⁰ Emyr Owain Wile and Alys Einion-Waller, 'Understanding Mordel: obtaining informed consent for trisomy screening', (2021) 29 British Journal of Midwifery, 106.

¹⁷¹ Mpatisi Moyo and others, 'Healthcare practitioners' personal and professional values', (2016) 21 Advances in Health Sciences Education, 257.

¹⁷² Anne Moller and others, 'Danish Sonographers' Experience of the Introduction of "Moderate Risk" in Prenatal Screening for Down Syndrome', (2018) 2 Journal of Pregnancy, 1.

¹⁷³ Emyr Owain Wile and Alys Einion-Waller, (n170), 106.

¹⁷⁴ Mordel, (n14), 92.

¹⁷⁵ Emyr Owain Wile and Alys Einion-Waller (n170), 110.

¹⁷⁶ Ibid.

training and CPD, the courts may turn to the possibility of systemic negligence, and whether this is causative of the individual error by the HCP.¹⁷⁶ *ARB* reiterated that the process of obtaining patient consent must not be illogical or unreasonable, and thus support is required for sonographers to navigate the modified process.¹⁷⁷

7.3 Supporting Parent Understanding: Key Considerations

7.3.1 Dishonesty

Parents may experience feelings of embarrassment or shame for not understanding the information on trisomy screening.¹⁷⁸ In light of this, there is a substantial risk that parents will confirm to HCPs that they have understood the information when, in fact, they have not; this carries potentially significant repercussions on the ability of HCPs to effectively support parent understanding. *Mordel* underlined that HCPs should endeavour to explore whether the patient has ‘truly and genuinely’ comprehended the information before accepting, or refusing, the offer to screen.¹⁷⁹ However, taking into account the reluctance of parents to admit that they do not fully understand the information, the question remains as to how this duty is to be reasonably discharged.

This begs the question as to what HCPs are expected to do if they suspect parents do not understand the information. Indeed, sonographers are unsure of their duty if they suspect that a parent has misunderstood the information.¹⁸⁰ While issue of patient dishonesty was not specifically addressed by Jay J in *Mordel*, a common narrative emerged that suggested if the sonographer suspects the parents have not understood the information, they are to be sent back to their midwife, preferably on the same day, for re-counselling.¹⁸¹

It is safe to conclude that this is not a ‘reasonable’ or ‘logical’ system for securing parent consent. Taking into account midwives’ and sonographers’ differing shift patterns, in addition to the straightjacket of increasing time constraints and resource-limited NHS maternity units, this suggestion is “at best idealistic, and at worse, entirely impractical and unlikely”.¹⁸² Lords

¹⁷⁶ *Gottstein v Maguire and Walsh* (2007) 4 IR 435.

¹⁷⁷ *ARB v IVF Hammersmith* (2018) EWCA Civ 2803.

¹⁷⁸ Parent Qualitative Findings, Section 5.3.1.

¹⁷⁹ *Mordel* (n14), 89.

¹⁸⁰ HCP Qualitative Findings, Section 6.4.1.

¹⁸¹ *Mordel* (n14), para 88.

¹⁸² *Emyr Owain Wile and Alys Einion-Waller* (n170) 110.

Kerr and Reed, in *Montgomery*, acknowledged that the duty on HCPs, “... who are more hurried, are obliged to pause and engage in the discussion which the law requires ... this may not be welcomed by some healthcare providers”.¹⁸³ As Jackson opines, a dialogue with parents will cost time and money, components of care which many NHS units do not possess under the current climate.¹⁸⁴ In addition, *Thefaut* confirmed that patients must be given adequate time and space to make healthcare choices; therefore, being sent for re-counselling, on the same day as the scan, may not be considered reasonable.¹⁸⁵

With the addition of more information, and an increasing demand for an enhanced understanding of complex medical considerations, it is foreseeable that this issue will only be exacerbated following the recent amendments to the trisomy pathway. This also raises concern surrounding the effective execution of the HCP’s duty to explore parents’ understanding of screening, subject to the expectation in *Mordel*.

7.3.2 Understanding Edwards’ and Patau’s Syndrome

Down’s Syndrome is a very visible condition in British society: we interact with people with DS in our community; we watch people with DS in films and documentaries; we read about DS in books or newspaper articles; and we hear about DS in recent court cases.¹⁸⁶ ES and PS, on the other hand, are much less visible. Parents will typically encounter information on ES and PS for the first time at pre-screening appointments, having never heard about them previously.¹⁸⁷

With this in mind, parents are often introduced to ES and PS upon entry of the screening pathway, without them having any preconceptions or opinions on the conditions.¹⁸⁸ Thus, what they read and hear at pre-screening consultations will ultimately cement their foundational understanding and perception of ES and PS; therefore, as ASW and FASP underline, providing a balanced and accurate account of ES and PS, to support parent understanding, is vital at this stage.

¹⁸³ *Montgomery* (n1), 93.

¹⁸⁴ *Emily Jackson*, (n96), 6.

¹⁸⁵ *Thefaut v Johnson* (2017) EWHC 497 (QB).

¹⁸⁶ *R (Crowter and Ors) v SSHSC* (2021) EWHC 2536. Permission granted for appeal (08/03/2022).

¹⁸⁷ HCP Qualitative Findings, Sections 6.4.2 and 6.4.3, and Parent Qualitative Findings, Section 5.3.2.

¹⁸⁸ Parent Qualitative Findings, Section 5.3.2.

Disclosure of unbalanced or inaccurate information on ES and PS, risks hindering parents' understanding of the conditions.¹⁸⁹ ES and PS are being misunderstood by parents as the *same* or 'worse forms' of DS, demonstrating a fundamental misunderstanding of the conditions, particularly in terms of what the aneuploidy means for maternal and fetal outcome.¹⁹⁰ Indeed, ES and PS both differ significantly from DS, in prognosis and care management. Without a foundational understanding of the trisomies, parents risk providing consent to screen for ES and PS, having misunderstood the consequences obtaining a higher-risk or positive diagnosis, and the impact this may have on their pregnancy care.¹⁹¹

A concerning narrative has emerged that ES and PS are 'lethal' or 'deadly' conditions, that are 'incompatible with life'.¹⁹² While were once generalised thus, this misconception has now been dispelled.¹⁹³ Improvements in medical knowledge and specialised care pathways, mean that some babies with *full* ES and PS, can live into late childhood and early adulthood. Janvier and Watkins conclude that "trisomy 13 and 18 are not lethal conditions ... they are life-limiting conditions".¹⁹⁴ Use of terminology such as 'lethal' and 'deadly' has been widely criticised, as it "is used for a heterogenous group of conditions to imply an ethical conclusion, rather than to present a clear prognosis: it obscures rather than aids communication and counselling".¹⁹⁵ A review of medical literature reveals that there is no agreed 'list' of conditions that fall within this definition.¹⁹⁶ However, as a result of ES and PS being labelled 'lethal', the provision of life-saving treatments are often withheld, and the option to terminate is normalised.¹⁹⁷

Information on ES and PS is often presented as binary: either the baby has ES or PS, or they do not, with "little explanatory nuance(s) as to what exactly a positive diagnosis might mean for the baby's future".¹⁹⁸ This omits consideration of the phenotypic nuances of the

¹⁸⁹ Parent Qualitative Findings, Section 5.3.2.

¹⁹⁰ HCP Qualitative Findings, Section 6.4.3 and Parent Qualitative Findings, Section 5.3.2.

¹⁹¹ Parent Qualitative Findings, Section 5.3.2.

¹⁹² HCP Qualitative Findings, Section 6.4.3 and Parent Qualitative Findings, Section 5.3.2.

¹⁹³ Parent Qualitative Findings, Section 5.3.2.

¹⁹⁴ Annie Janvier and Andrew Watkins, 'Medical interventions for children with trisomy 13 and trisomy 18: what is the value of a short disabled life?' (2013) 102 *Acta Paediatrica*, 1116.

¹⁹⁵ DJD Wilkinson, P Thiele, A Watkins and L De Crespigny, "Fatally flawed? A review and ethical analysis of lethal congenital malformations", (2012) 110 *British Journal of Obstetrics and Gynaecology*, 1032.

¹⁹⁶ Caroline Simons, 'Incompatible with life: does article 40.3.3 permit abortion for 'fatal foetal abnormality', (2015) 21 *Medico-Legal Journal of Ireland*, 13.

¹⁹⁷ Martin McCaffrey, 'Lives Worth Living', (2017) 32 *Issues in Law & Medicine*, 215.

¹⁹⁸ Bret Asbury, 'Fostering Informed Choice: Alleviating the Trauma of Genetic Abortions', (2015) 25 *Cornell Journal of Law and Public Policy*, 301.

conditions, which are determinative of the baby's health characteristics and prognosis.¹⁹⁹ Indeed, babies with mosaic or translocation DS, ES or PS present differently to those with full aneuploidy, often exhibiting fewer dysmorphic features and an increased prognosis.²⁰⁰ The material risk of stillbirth and/or miscarriage is also reduced, if the baby presents as mosaic or translocation.²⁰¹

Thus, the provision of balanced and accurate information is crucial to support parents' understanding of ES and PS, requiring that HCPs effectively manage generalisations and outdated depictions of the conditions.²⁰² Counselling on ES and PS must also include evidence-based survival figures, and should avoid use of language that assumes an outcome, preferably from the perspective of those with lived experience.²⁰³ *Webster (A Child) v Burton Hospital NHS Trust* emphasised the significance of informing patients of emerging and recent evidenced-based research, albeit incomplete.²⁰⁴ This information must also be readily accessible to those *providing care*.²⁰⁵

It is important to contend the growing narrative among academics that generalisations of ES and PS are used by the medical community to *intentionally* harm this population, as suggested by Janvier and Watkins.²⁰⁶ This may be a reflection on the provision of training, or lack thereof. Many midwives were not introduced to ES and PS during their degree programmes/training, as the existing screening programme only included DS.²⁰⁷ Similarly, sonographers have not had *any* bespoke training on ES or PS, since the introduction of the conditions to the pathway.²⁰⁸ While the Nuffield Council report also underlined the urgent need for professional training on ES and PS, the Council underline that, "these training courses are not compulsory and are limited in reach".²⁰⁹ The implementation of *mandatory* training

¹⁹⁹ Chih-Ping Chen, 'Prenatal Diagnosis and Genetic Counselling for Mosaic Trisomy 13', (2010) 49 Taiwan J Obstet Gynecol, 13.

²⁰⁰ Ibid.

²⁰¹ Ibid.

²⁰² Annie Janvier and Andrew Watkins, (n194) 12; see also, Steven R Leuthner and Krishna Acharya, 'Perinatal Counseling Following a Diagnosis of Trisomy 13 or 18: Incorporating the Facts, Parental Values, and Maintaining Choices', (2020) 20 Adv Neonatal Care, 204.

²⁰³ John Carey, '*Perspectives on the care and management of infants with trisomy 18 and trisomy 13: striving for balance*', (2012) 24 Curr Opin Pediatr, 672.

²⁰⁴ *Webster (A Child) v Burton Hospital NHS Foundation Trusts* (2017) EWCA Civ 62, 40.

²⁰⁵ Ibid.

²⁰⁶ Annie Janvier and Andrew Watkins, (n194), 1117.

²⁰⁷ HCP Qualitative Findings, Section 6.4.2.

²⁰⁸ HCP Qualitative Findings, Sections 6.4.1 and 6.4.2.

²⁰⁹ Nuffield Council Report, 47.

programmes for midwives *and* sonographers on the trisomies, in addition to provision of experiential knowledge, would support HCPs' understanding of the trisomies, increasing their confidence to support parent decision-making.²¹⁰

Use of these generalisations and terms, to support parents' understanding of ES and PS, may be borne from the influence of common law on the *modus operandi* of communicating risk, in an increasingly litigious society. With common law reinforcing the legal significance of ensuring effective communication for patient understanding and materiality of risk, as per *Montgomery* and *Mordel*, HCPs may be seeking to ensure that this duty is discharged, possibly at the detriment of providing a balanced account of all cases of ES and PS. It may be that the health risks for baby and mother, associated with ES and PS, are prioritised by HCPs at this stage in the pathway to protect their professional interests, rather than purposely 'harming' the population.

7.3.3 Differentiating Between First Line and Invasive Testing

Prioritisation of information on 'risk' may also lead to misunderstandings by parents between first-line screening and invasive testing.²¹¹ A key determinant for Jay J finding in favour the Claimant in *Mordel*, was the failure of the HCPs to explore the parent's understanding of the practicalities of screening before securing consent.²¹² Consistent with the findings of this research, the expert witness in *Mordel* explained that it is common for parents to misunderstand the purpose and practicalities of the various methods of screening, and underlined how supporting parents' understanding of these methods is crucial to informed decision-making.²¹³

The decision in *Mordel* would imply that *both* the midwife and sonographer are duty-bound to support parent understanding on the difference between the methods of screening and testing, including the essential elements and their purposes, before securing consent.²¹⁴

²¹⁰ E. Carroll and others, 'A qualitative investigation of the decision-making process of couples considering prenatal screening for Down syndrome,' (2012) 32 *Prenatal Diagnosis* 57; and Lou S and others, 'This is the child we were given: A qualitative study of Danish parents' experiences of a prenatal Down syndrome diagnosis and their decision to continue the pregnancy', (2020) 23 *Sex Reprod Healthc*, 1, and Brian Skotko, 'Mothers of children with Down syndrome reflect on their postnatal support', (2005) 115 *Paediatrics*, 64.

²¹¹ HCP Qualitative Findings, Sections 6.4.4 and 6.4.6, and Parent Qualitative Findings, Section 5.3.3.

²¹² *Mordel* (n14), 60

²¹³ HCP Qualitative Findings, Sections 6.4.4 and 6.4.6; and *Mordel* (n14) 63.

²¹⁴ *Mordel*, (n14) 89.

However, traditionally, sonographers are not required to provide further counselling to parents on such matters, as this information had been reviewed by the midwife before attending the appointment.²¹⁵

The reported misunderstanding between the methods of screening and testing, by the research, participants is not entirely surprising, considering that parents typically do not read the information materials before attending the appointments, and are presented with information on a host of other tests they can opt to undertake, as part of their antenatal care.²¹⁶ Indeed, as parents are required to undertake various blood tests and 'routine' ultrasound scans throughout early pregnancy for other purposes, it is foreseeable, in the absence of clear explanatory dialogue, they may become disorientated, losing sight of which blood draw, test and ultrasound examination is being performed for the purposes of trisomy screening.²¹⁷ Furthermore, upon receiving information on invasive testing and the risk of procedural miscarriage, it would be reasonable to presume that this would manifest in the mind of most parents before engaging with the pathway, and their focus would remain on this consideration for the duration of their early pregnancy care.²¹⁸

This misunderstanding may indicate the sparsity of resources and aids necessary to support parent understanding.²¹⁹ Research by Garcia-Retamero and Portocarrero *et al.* conveyed the benefits of employing aids and resources to enhance patient understanding, utilising pictures, flowcharts, and illustrations to support discussion at clinical appointments.²²⁰ The use of aids has also been proven to reduce maternal anxiety, when faced with complex medical information, by promoting and supporting the parent's medical literacy.²²¹ However,

²¹⁵ HCP Qualitative Findings, Section 6.4.1.

²¹⁶ Parent Qualitative Findings, Section 5.3.3; see also, Neeltje M Crombag and others, 'Determinants affecting pregnant women's utilization of prenatal screening for Down syndrome: a review of the literature' (2013) 26 J Matern Fetal Neonatal Med, 1676.

²¹⁷ Parent Qualitative Findings, Section 5.3.3.

²¹⁸ Parent Qualitative Findings, Section 5.3.3.

²¹⁹ Parent Qualitative Findings, Section 5.3.3.

²²⁰ Garcia-Retamero R, Galesic M, 'Who profits from visual aids: Overcoming challenges in people's understanding of risks', (2010) 70 Social Science & Medicine 1019; and Maria Esther Leiva Portocarrero and others, 'Use of a patient decision aid for prenatal screening for Down syndrome: what do pregnant women say?', (2017) 17 BMC Pregnancy Childbirth, 90.

²²¹ Rebecca Say and Stephen Robson, 'Helping Pregnant Women Make Better Decisions: A Systematic Review of the Benefits of Patient Decision Aids in Obstetrics', (2011) BMJ Open, 1:e000261.

midwives seldom use ‘aids’ to support parents’ understanding of screening and testing, due to time, costs, and access to the provision of specialist resources.

7.3.4 Demystifying the Jargon

‘Trisomy’, ‘Nuchal fold’ ‘NT measurement’, ‘crown rump length’, ‘combined screening’, ‘amniocentesis’, are all reported to be commonly used terms in the information packs and during counselling.²²² However, use of medical jargon or complex terminology risks interfering with parents’ understanding of the process.²²³

Smith established that cultivating the patient’s medical literacy requires appropriate support at pre-treatment consultations to enable informed choice.²²⁴ This case suggested that HCPs utilise appropriate and non-technical language when explaining the implications of a particular intervention.²²⁵ Consistent with the GMC guidelines, *Smith* required HCPs to take reasonable steps to ensure the language used is intelligible to the particular patient.²²⁶ *Thefaut* also reinforced that maintaining jargon-free and simplistic language, when imparting medical information, is crucial to supporting understanding: a point raised by the NMC and GMC.²²⁷

While maternity reports and information pertaining to screening understandably include complex terminology, abbreviations and statistics, HCPs must take reasonable steps to interpret and explain these concepts, to render information comprehensible for parents. This may require HCPs to provide a user-friendly index to accompany the maternity report, explaining commonly used terms, abbreviations and statistics.²²⁸

²²² Parent Qualitative Findings, Section 5.3.4.

²²³ HCP Qualitative Findings, Section 6.4.7 and Parent Qualitative Findings, Section 5.3.4.

²²⁴ *Smith v Tunbridge Wells HA* (1994) 5 Med. L.R. 334.

²²⁵ *Ibid.*

²²⁶ *Ibid.*

²²⁷ *Thefaut v Johnson* (2017) EWHC 497 (QB).

²²⁸ Skirton H, O’Connor A and Humphreys A, ‘Nurses’ competence in genetics: A mixed method systematic review’, (2012) 68 *Journal of Advanced Nursing*, 2387.

7.4.0 Supporting Parent Choice: Situating Professional Duty

Midwives and sonographers face a growing number of challenges in the execution of their duty to support parent choice for trisomy screening.²²⁹ Traditionally, midwives would support parent choice for screening, making a record of their initial decision at the booking appointment.²³⁰ The sonographer's role, on the other hand, was limited to confirming that parent consent was forthcoming on the day of the examination appointment. This interpretation is consistent with the expert witness statement for the sonographer in *Mordel*, which outlined that sonographers are not required to explore parent choice, due to the significant risk of undermining the parent's decision, at this stage in the pathway, if their choice was brought into question; this risk is particularly evident if they *decline* screening.²³¹ Indeed, on the issue of supporting parent choice, midwives' and sonographer's professional training actively *discourage* 'exploring' parent choice, particularly if screening has been declined, as this risks undermining their decision.²³²

However, Jay J, in *Mordel*, felt that the sonographer should play a more active role in the process of supporting parent choice before securing consent. He contended that the sonographer had "*overstated the difficulty in exploring the patient's level of understanding without at the same time appearing to undermine her right to choose*".²³³ He followed on from this point by explaining that he rejects the contention that counselling parents on their understanding "*involves in some way prying inappropriately into the patient's reasons or reasoning and undermining her free choice*".²³⁴ Jay J outlined, *obiter dictum*, that the risk of undermining patient choice will only arise in the presence of "*maladroit or insensitive interrogation*", stating that this is "*a risk that NHS professionals are well habituated to avoid*".²³⁵ Jay J concluded that the sonographer could have explored Mrs Mordel's choice on the day of the scan, and not just taken her decision at face value.²³⁶

Following the Claimant's rejection of screening at the ultrasound appointment with the sonographer, Jay J assessed whether the midwife should have further explored this choice at

²²⁹ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.5.1.

²³⁰ HCP Qualitative Findings, Section 6.5.1.

²³¹ *Mordel* (n14), 70.

²³² HCP Qualitative Findings, Section 6.5.2

²³³ *Mordel* (n14) 85.

²³⁴ *Ibid*, 90.

²³⁵ *Ibid*.

²³⁶ *Ibid*, 92.

later consultations.²³⁷ The expert witness for the midwife explained that “*there was no duty on the midwife to ask further questions*”, and “*if you revisit the issue, you run the risk that the mother will feel that she has made the wrong choice; and will therefore be pressurised into changing her decision*”.²³⁸ However, after applying *Bolitho* to the question of reasonableness, Jay J held that taking reasonable steps meant that the midwife, after discovering that Mrs Mordel had originally booked to have screening but refused on the day of the scan, should not have left the matter there.²³⁹ Jay J suggested that “*a simple and straightforward exploration and check that what has occurred, or not has occurred, was and is in accordance with the patient’s wishes continues to place her at the centre of the decision-making process and amounts to that taking of reasonable steps to ensure that everything has gone and is continuing to proceed according to plan*”.²⁴⁰

While identifying individual error and accountability is a natural part of the process for deciding on matters of systemic negligence, it is arguable that Jay J placed undue focus on the midwife’s conduct at this stage, and thus ineffectively addressed the glaringly obvious deficiencies within the broader clinical system for supporting parent choice and consent. The decision in *Mordel* has resulted to midwives feeling that that they are being villainised, scapegoated and “are not doing (their) jobs properly.”²⁴¹

Thus systemic fragilities, when supporting parent choice, may stem from the disconnect between units and roles of the midwife and sonographer. Following Jay J’s unnuanced assessment to the question of a ‘reasonable’ system, it can only be assumed that sonographers are required to appropriate a hybrid and chameleonic role when exploring parent choice.²⁴² As sonographers, under the established system, were only required to confirm whether consent was forthcoming on the day of the scan, the question remains whether exploration into parent choice is required at this stage.²⁴³ If so, the provision of appropriate training must be mandatory to negate the risk of undermining parent choice, particularly if the patient has *declined* screening.

²³⁷ Ibid, 129.

²³⁸ Ibid.

²³⁹ Ibid, 136.

²⁴⁰ Ibid, 136.

²⁴¹ HCP Qualitative Findings, Section 6.2.1.

²⁴² Mordel (n14) 96.

²⁴³ Jay J alludes to this in para 92 but it is not clear.

Constructing a 'reasonable' system becomes increasingly pertinent in situations where parents change their mind on screening, as the pregnancy progresses. A parent's change of mind for screening currently instils a significant amount of anxiety and concern among the profession, as they feel the current system for securing consent does not provide the necessary protection, both legally and ethically. While the expert witness for the Defendant, in *Mordel*, explained that changes of mind were common – often warranting a deeper discussion into the parent's choice to accept/decline screening, in such circumstances – what is clear from the divergent expert witness statements was the lack of a cogent and uniform system to secure consent, if parents change their mind.²⁴⁴

National guidelines, namely FASP and ASW, do not proffer clear instruction for HCPs, either. Taking into account that the process of securing consent now straddles both the midwife and sonographer appointments – which occur on various stages along the pathway – the risk of systemic failings, in the absence of clear instruction on how to evidence change of mind, significantly increases.

Aside from providing sonographers with the same training midwives receive on counselling parents on choice, a broadening of scope to include systemic considerations allows a fair assessment of whether existing systems are able to effectively facilitate Jay J's suggestion. The case of *Robertson* conveyed the importance of establishing lines of communication between units and HCP teams, for the purpose of enhancing systems of clinical practice.²⁴⁵ Drawing from the recommendations of Brooke LJ, in this decision, it may be logical and reasonable to enhance interprofessional practice by constructing clear and effective modes of communication between units. Establishing interprofessional maternity units may be required to encourage a model of consent based on shared and continued communication between the professions and the parents, throughout the screening pathway.

²⁴⁴ Ibid, 76.

²⁴⁵ *Robertson v Nottingham* (1997) 8 Med LR 1.

7.4 Supporting Parent Choice: Key Considerations

7.4.1 Situating Parent Decision-Making and Choice

The HCP's duty, to support parent decision-making, may become further complicated by the reported tendency of parents to make a choice on screening outside of the clinical setting.²⁴⁶ Indeed, Crombag *et al.* opine that decisions on screening typically take place either before, or just after conception.²⁴⁷ Upon review of patient decision-making in other areas of medicine, it is not uncommon for patients to make a decisions on treatment options before engaging with the professionals.²⁴⁸

Predetermined decision-making poses significant challenges for HCPs, in their efforts to discharge their duty of care.²⁴⁹ While it is evidently probable that decisions on pregnancy care, including screening, will be made outside the clinical setting, parents may decide to screen, or indeed not to screen, *before* receiving professional advice.²⁵⁰ As such, parents do not feel the need to engage with the information or advice, as they had made a predetermined decision.²⁵¹ This factor underlines the risk of consent becoming a ritualistic 'tick-box' process, rather than an informed and deliberate decision to engage with the pathway.²⁵²

7.4.2 Opportunism and Uptake

With this in mind, consideration of the potential influences on decision-making and screening uptake is logical. Opportunism operates as a key influence on the uptake of trisomy screening.²⁵³ While the term 'opportunism' possesses a range of definitions, broadly, it is the behaviour of taking advantage of a situation or circumstance, with little regard for potential consequence. With parents emphasising that its mere availability was a contributing factor to uptake, this may undermine purposeful decision-making.²⁵⁴ The significance of this finding

²⁴⁶ HCP Qualitative Findings, Section 6.5.3 and Parent Qualitative Findings, Section 5.4.1.

²⁴⁷ Neeltje M Crombag and others, '*Determinants affecting pregnant women's utilization of prenatal screening for Down syndrome: a review of the literature*', (2013) 26 J Matern Fetal Neonatal Med, 1676.

²⁴⁸ Emily Jackson, (n96), 14.

²⁴⁹ HCP Qualitative Findings, Section 6.5.3

²⁵⁰ Parent Qualitative Findings, Section 5.4.1.

²⁵¹ Parent Qualitative Findings, Section 5.4.1.

²⁵² Emily Jackson, (n96), 15.

²⁵³ Parent Qualitative Findings, Section 5.4.2.

²⁵⁴ Parent Qualitative Findings, Section 5.4.2; see also, Bettina Schone-Seifert and Chiara Junker, '*Making use of non-invasive prenatal testing (NIPT): rethinking issues of routinisations and pressure*', (2021) 49 Journal of Perinatal Medicine, 959.

rests on the indication that the reportedly opportunistic nature of screening could pose substantial challenges for HCPs to obtain consent.²⁵⁵

The mere availability of the provision to screen may imply that it is beneficial, and that parents should undertake screening to take advantage of this opportunity, rather than taking steps to reflect on whether it is the appropriate choice for them.²⁵⁶ While the provision of screening seeks to empower parents to take control of their healthcare choices, HCPs should remind parents that, while the provision of screening may increase medical knowledge, possession thereof does not necessarily equate to better healthcare outcome, or an increase in reproductive autonomy.²⁵⁷ HCPs must also remind parents its availability should not be determinative of their choice to accept it.

Opportunism, and the suggestion that consent may not be purposeful, raises unique challenges to the HCP's duty to secure consent, under the current 'trisomy' model of screening. Parents have the option to screen for DS only, ES and PS only, or all three together. However, parents consider ES and PS as accompaniments to the decision to screen for DS, interpreted as a 'screening package'.²⁵⁸ Thus, the current presentation of trisomy screening, as a genetic model, could mean that parents are providing *generic* consent for the three conditions, without having considered the unique implications of potential outcomes on decision-making.²⁵⁹

Indeed, providing 'generic' consent to screen for DS, ES and PS together, means that consent may not be bespoke to each condition.²⁶⁰ This is concerning, as the implications, consequences and subsequent decision-making, in the event of a high-risk result, will differ significantly between trisomies, due to their unique health characteristics and outcomes for

²⁵⁵ Dena S Davis, '*Opportunistic Testing: the Death of Informed Consent?*', (2013) 23 Health Matrix: Journal of Law-Medicine, 35.

²⁵⁶ Ibid.

²⁵⁷ Olivia Miu Yung Ngan and others, '*Parental expectations of raising a child with disability in decision-making for prenatal testing and termination of pregnancy: A mixed methods study*', (2020) 103 Patient Education and Counselling, 2373.

²⁵⁸ Parent Qualitative Findings, Section 5.4.2.

²⁵⁹ Parent Qualitative Findings, Section 5.4.2.

²⁶⁰ Emily Jackson, '*Regulating Non-Invasive Prenatal Testing: the view from the UK*', (2014) Japanese Journal of Law and Politics, 15.

both mother and baby.²⁶¹ Thus, the suggestion of a fragmentation of the trisomy pathway becomes increasingly pertinent in this regard.

The suggestion, to fragment the pathway, becomes increasingly desirable in light of parents' revelation that they were *unaware* trisomy screening also targets ES and PS, in addition to DS.²⁶² As discussed previously, this reported oversight of ES and PS, could rest on the tendency of HCPs to refer to 'trisomy' screening as 'Down Syndrome' screening.²⁶³ However, this must be treated with caution as other factors may be at play, namely the reported failure of parents to read the information packs. Another reason may be that DS typically takes centre stage during pre-screening consultations. While these conclusions are partially speculative, the objective for HCPs must be to enable parents to make *conscious* and deliberate decisions on screening, particularly since the introduction of ES and PS to the 'Down's Syndrome' pathway.

7.4.3 Conformity

While opportunism may be an influence on screening uptake, conforming to perceived social norms has also been identified as an influence on choice, in this study.²⁶⁴

Social conformity is often cited as the *principal* reason why parents decide to undertake screening. Mclean opines that if a patient experiences undue influence or pressure to exercise a particular choice, that decision is incapable of being defined as truly autonomous.²⁶⁵ These influences and pressures, it is noted, may extend from personal values or externally from the healthcare setting.²⁶⁶ However, Mclean suggests that a patient "is virtually never free from external influences, yet we nonetheless expect and receive respect for our actions (and inactions) outside of the healthcare context".²⁶⁷

All parents want to be 'good' parents; however, portraying the image of a 'good' and 'responsible' parent, often rests on conformity to existing social norms and values.²⁶⁸ A risk

²⁶¹ Ibid.

²⁶² Parent Qualitative Findings, Section 5.4.2.

²⁶³ Issue raised during the British Journal of Midwifery conference (12/04/2021).

²⁶⁴ Parent Qualitative Findings, Section 5.4.3.

²⁶⁵ Sheila A.M. McLean, *Autonomy, Consent and the Law*, (1st Edn, Taylor & Francis Group, 2009), 51.

²⁶⁶ Parent Qualitative Findings, Section 5.4.3.

²⁶⁷ Ibid.

²⁶⁸ Parent Qualitative Findings, Section 5.4.3; see also, Panagiota Nakou, 'Is routine prenatal screening and testing fundamentally incompatible with a commitment to reproductive choice? Learning from the historical context', (2021) 24 *Medicine, Health Care and Philosophy*, 73.

exists that conforming to social expectation as a ‘responsible parent’, to both HCPs and family, compels parents to engage with screening.²⁶⁹ This may also speak to the impact that the confined social structure surrounding the parents, in terms of both family and institutional support systems, has on their choice to accept or refuse screening.²⁷⁰ It may be that parents do not want to be seen as ‘responsible’ for having a baby with trisomy.²⁷¹ Thus, societal environment is a key influence on parents’ engagement with screening, and HCPs may therefore be required to address unique social contexts during counselling, without underestimating the social pressure.²⁷²

7.4.4 Early Signs of Trisomy Screening’s Routinisation

Conformity also raises concerns around screening’s routinisation; as the participant in this research stated, “... *I did not feel the need to question it because doesn’t everyone else have it?*”.²⁷³ It has been well documented from previous research that parents perceive DS screening as a routine part of their care, consistent with this study’s findings.²⁷⁴ However, seldom does any research on routinisation of screening proffer cogent suggestions as to where this perception originates, or how to manage it.

While social conformity may be one explanation, an imbalance of information on the care pathways – to refuse or undertake screening – could also be the cause. With a reported absence of information on the care pathway following a *refusal* of screening, compared to an abundance of information available on the pathway to undertake it, this may proffer another cause to the routinisation of screening.²⁷⁵

As it is human nature to fear the unknown, an absence of information on the care pathway following a refusal of screening, could impel parents to accept screening, due to anxiety and fear in this regard.²⁷⁶ It may also be that an imbalance of information subtly suggests that to

²⁶⁹ Parent Qualitative Findings, Section 5.4.3.

²⁷⁰ Gottfredsdottir H, Bjornsottir K and Sandall J, ‘*How do prospective parents who decline prenatal screening account for their decision? A qualitative study*’, (2009) 69 *Social Science & Medicine*, 277.

²⁷¹ *Ibid.*

²⁷² Olivia Miu Yung Ngan and others (n257), 2373.

²⁷³ Parent Quantitative Findings, Section 4.3 and Parent Qualitative Findings, Section 5.4.4.

²⁷⁴ HCP Qualitative Findings, Section 6.5.4 and Parent Qualitative Findings, Section 5.4.4.

²⁷⁵ Parent Quantitative Findings, Section 4.3 and Parent Qualitative Findings, Section 5.4.4.

²⁷⁶ HCP Qualitative Findings, Section 6.5.4 and Parent Qualitative Findings, Section 5.4.4.

undertake screening is the expected option, or standard practice, alluding to a directive model of care.

A directive model of care poses significant obstacles to the ability of parents to exercise a choice, in the context of screening.²⁷⁷ It has been noted that a directive model of care involves elements of persuasive coerciveness from individual HCPs, or broadly by the institution.²⁷⁸ As the Nuffield Council underline, “aside from inaccurate or absent information, risks to reproductive autonomy are posed when influence is exerted on women to undergo screening ... through personal, institutional or societal expectations, biases or pressures”.²⁷⁹ The Nuffield Council conclude that parents must make *voluntary* decisions to undergo examination and treatment, requiring HCPs to address and dispel misconceptions of trisomy screening, such as being a standard or routine part of antenatal care.²⁸⁰ In light of this, a risk exists that consenting to undertake screening may become a mere formality, lacking the desired *deliberate* decision-making by parents.²⁸¹ This may also speak to a hidden curriculum in pre-screening information, placing a subtle pressure on parents to undertake it.²⁸²

Another factor, which could exacerbate the routinisation of screening, could be the inability of parents to distinguish between essential and non-essential care.²⁸³ With the growing number of scans and tests being offered - not only on the trisomy pathway, but also for other antenatal and perinatal screening programmes - it is foreseeable that parents may become disorientated, and perceive trisomy screening to be ‘standard’ antenatal care.²⁸⁴ As a result, a risk exists of parents being wholly unprepared for the consequences of receiving a higher-risk result, or being entirely unaware that trisomy screening has even taken place.²⁸⁵ It may

²⁷⁷ HCP Qualitative Findings, Section 6.5.7; see also, Sanne L. van der Steen (n146), 235.

²⁷⁸ *Ibid.*

²⁷⁹ Nuffield Council Report, 28.

²⁸⁰ *Ibid.*, 28.

²⁸¹ Parent Qualitative Findings, Section 5.4.4.

²⁸² Tsouroufli M, ‘Routinisation and constraints on informed choice in a one-stop clinic offering first trimester chromosomal antenatal screening for Down’s syndrome’, (2011) 27 *Midwifery*, 431.

²⁸³ Parent Qualitative Findings, Section 5.4.4; see also, Tsouroufli M, ‘Routinisation and constraints on informed choice in a one-stop clinic offering first trimester chromosomal antenatal screening for Down’s syndrome’, (2011) 27 *Midwifery*, 431.

²⁸⁴ Parent Quantitative Findings, Section 4.3 and Parent Qualitative Findings, Section 5.4.4.

²⁸⁵ Heyman B and others, ‘On being at higher risk: A qualitative study of prenatal screening for chromosomal anomalies’, (2006) 62 *Social science & medicine*, 2360.

be that a clearer distinction can be drawn between essential and non-essential care, earlier in the pathway, to better empower reproductive autonomy.

7.4.5 Perception of the Trisomies

A significant factor going to the decision to undertake or refuse screening, in part, rests on the perception of disability.²⁸⁶ Parents and professionals are concerned that biased, outdated, and negative generalisations of the trisomies, currently influence the decision to uptake screening.²⁸⁷ While a “culture of institutional and professional bias” is often cited as the cause, the law may also be encouraging a culture of discrimination and prejudice, following decisions such as *Mordel*.²⁸⁸ Indeed, this decision has sent a negative and damaging message to the DS community.²⁸⁹

While Jay J sincerely contended that “*nothing I have said in this judgment should be interpreted as suggesting that the birth of a child with Down’s syndrome must be seen as unwelcome*”, the decision to award damages to the Claimant may be misinterpreted, and suggests otherwise.²⁹⁰ Agreeably, while the judgment itself focussed primarily on defective systems for securing parent consent, it sits uncomfortably, predominantly as parents were able to recover compensation due to the fact their baby was born with DS: this does not send a positive and inclusive message.²⁹¹

However, these reported discriminatory attitudes are currently being challenged by the DS community. A ground-breaking claim for judicial review, in 2021, brought by members of the DS community, sought a declaration of incompatibility under section 4 of the HRA, in respect of section 1(1)(d) of the Abortion Act 1967. In *R (Crowter and Ors) v SSHSC* the Claimants contended that section 1(1)(d) is incompatible with Articles 2, 3, 8 and 14 of the ECHR.²⁹² While Singh LJ and Lieven J disagreed with all of the counsel’s submissions, this case is

²⁸⁶ Gottfredsdottir H, Bjornsottir K and Sandall J, ‘How do prospective parents who decline prenatal screening account for their decision? A qualitative study’, (2009) 69 *Social Science & Medicine*, 274.

²⁸⁷ HCP Qualitative Findings, Section 6.5.8 and Parent Qualitative Findings, Section 5.4.5.

²⁸⁸ See also, *R (Crowter and Ors) v SSHSC* (2021) EWHC 2536.

²⁸⁹ Discussions with campaign groups supporting DS.

²⁹⁰ *Mordel* (n14), 153.

²⁹¹ Parent Qualitative Findings, Section 5.4.5.

²⁹² *R (Crowter and Ors)* (n288). Currently been granted appeal.

symbolic of a shift in momentum to expunge the culture of prejudice against the DS community, in favour of the emancipation and empowerment of disability rights.

In *R (Crowter and Ors) v SSHSC*, the Claimants argued that a Westernised view of disability is typically influenced by discriminatory and ‘outdated’ perceptions, originating from the medical community, and results to the stigmatisation of entire communities: in this case, Down’s Syndrome.²⁹³ It was argued by counsel for the Claimants that section 1(1)(d) sends the message that “the lives of persons with conditions, such as DS, are not worth living”.²⁹⁴ While parent decision-making and consent for *screening* was not central to the decision itself, Perrot and Horn opine that *R (Crowter and Ors)* reinforced the significance of training and specialist resources for HCPs to preserve reproductive autonomy, in addition to implementing balanced and research-led support systems for parents when screening, with a view to remove a “culture” of bias and directiveness.²⁹⁵

The Down’s Syndrome Act – given Royal Assent on the 28th of April 2022 – is also symbolic of a conscious effort to remove negative and damaging cultural stereotypes. This Act is said to offer “more rights” for those living with DS, and their families, in health, education, housing and social care.²⁹⁶ While the true scope of this legislation is yet to be determined, it may be that ‘health’ permeates areas such as trisomy screening, particularly in the context of non-invasive prenatal testing.

While these developments are certainly a step forward in the removal of the alleged “culture” of discrimination and stigmatisation towards the DS community, from medical practice, the ES and PS community are also reportedly subject to similar treatment. Parents of babies with ES and PS continue to battle outdated and harmful depictions of the conditions, often feeling marginalised or ostracised by society and/or the medical community.²⁹⁷

In *Re L (A Minor)* – a case concerning the birth of a baby with ES, discussed later in the chapter – the President introduced the facts as “*L has had the enormous misfortune to be born with a*

²⁹³ *R (Crowter and Ors)* (n288). 90.

²⁹⁴ *Ibid*, 95.

²⁹⁵ Adeline Perrot and Ruth Horn, ‘Preserving women’s reproductive autonomy while promoting the rights of people with disabilities?: the case of Heidi Crowter and Maire Lea-Wilson in the light of NIPT debates in England, France and Germany’, (2022) 0 *Journal of Medical Ethics*, 1.

²⁹⁶ Down’s Syndrome Act 2022.

²⁹⁷ Parent Qualitative Findings, Section 5.4.5.

genetic disorder, trisomy 18, known as 'Edwards' Syndrome'".²⁹⁸ While the President was correct by outlining that the child's condition is not curative, the language used throughout the judgment, which extended from the medical expert reports, was unduly negative and pessimistic.²⁹⁹

Both ASW and FASP stress that the provision of accurate, balanced and unbiased information, in terms of the trisomies health characteristics and prognosis, is imperative to informed choice and reproductive autonomy. Despite this, this research allures to the possibility that HCPs focus excessively on the negative implications of the trisomies, particularly in the context of ES and PS, or provide an incomplete account of the conditions.³⁰⁰

Many other research studies have explained this as being suggestive of a culture of bias. However, this is arguably a rather superficial conclusion. Instead, this could speak to HCP unfamiliarity, or lack of specialised training, with these rarer conditions.³⁰¹ Many HCPs have not received bespoke training on the trisomies from the perspective of those with lived experience.³⁰² Indeed, HCPs believe that there are *no existing cases* of babies surviving ES and PS beyond a couple of weeks, for example.³⁰³

This focus on the negative implications of the conditions may also speak to the existence of defensive practices by HCPs. Post-*Montgomery*, in addition to the legal uncertainty left in the wake of *Mordel*, professional anxiety will undoubtedly be exacerbated. Thus, rather than being biased, it may be that fear of litigation impels HCPs to prioritise the risks and consequences associated with ES and PS, to ensure their duty is discharged in this regard.

A disparity between professional and patient norms may also be misinterpreted as 'bias'. Indeed, healthcare is situated within social institutions, formed by context, culture, the interaction and relationship between professional and patient values, and moral principles. It could be that the perception of the trisomies, situated within these social contexts, differ between the position of providing healthcare and the patient's moral reasoning; therefore,

²⁹⁸ Re L (A Minor) (2004) EWHC 2713 (Fam), 3.

²⁹⁹ Ibid, 5-9.

³⁰⁰ HCP Qualitative Findings, Section 6.5.8 and Parent Qualitative Findings, Section 5.4.5.

³⁰¹ HCPs Qualitative Findings, Section 6.4.2.

³⁰² HCP Qualitative Findings, Section 6.2.3.

³⁰³ SOFT training workshop.

rather than being conclusively biased, this issue is far more nuanced and could reflect friction between social settings and institutions.

7.4.6 Decisions on Intervention and Striking an Appropriate Balance

Facing the option to continue or end a pregnancy, is arguably the most difficult choice to make, in any area of medicine. This option is one that parents would be wholly unprepared for: parents typically enter parenthood under the presumption that their baby is healthy.³⁰⁴ Thus, when faced with this unimaginably difficult crossroad, providing a balanced and unbiased account of the trisomies, and available care pathways, is vital.³⁰⁵ As Simons concludes, unbalanced counselling of the trisomies “could lead to decisions about termination that are not well informed”.³⁰⁶

However, this research indicates that parents are able to readily access an abundance of information on the termination pathway, following a positive diagnosis for a trisomy, compared to the limited information available on continuing with the pregnancy.³⁰⁷ The findings also suggest that the option to terminate is often returned to, or repeated, on multiple occasions in pregnancy.³⁰⁸ It has been suggested that terminations are often presented as the *first* choice, following a trisomy diagnosis.³⁰⁹ It is safe to conclude that this reported disparity and imbalance in information, risks precipitating parents towards the option to terminate.³¹⁰

A “culture of bias” is often the reported culprit behind the imbalance and disparity of information. Parents who have babies with a trisomy, particularly ES and PS, often ‘battle’ with the profession to obtain more information on continuing with the pregnancy, following

³⁰⁴ Liknur Yesilcinar, Memnun Seven, Seyma Lnciser Pasalak and Gulden Guvenc, ‘Interventions aiming to improve informed decisions on prenatal screening and testing: A scoping review of the literature’, (2021) 5 Journal of Genetic Counselling, 1.

³⁰⁵ Abi Merriel, Medhat Alberry and Sherif Abdel-Fattah, ‘Implications of non-invasive prenatal testing for identifying and managing high-risk pregnancies’, (2021) 256 European Journal of Obstetrics & Gynaecology and Reproductive Biology, 32.

³⁰⁶ Caroline Simons, ‘Incompatible with life: does article 40.3.3 permit abortion for ‘fatal foetal abnormality’, (2015) 21 Medico-Legal Journal of Ireland, 13.

³⁰⁷ HCP Qualitative Findings, Section 6.5.8 and Parent Qualitative Findings, Section 5.4.6.

³⁰⁸ Parent Qualitative Findings, Section 5.4.6.

³⁰⁹ HCP Qualitative Findings, Section 6.5.8 and Parent Qualitative Findings, Section 5.4.6; see also, Bret Asbury, (n198), 304.

³¹⁰ Parent Qualitative Findings, Section 5.4.6.

a diagnosis.³¹¹ Parents also feel that they have been left in the dark, reporting limited discussion with professionals on the option to continue with the pregnancy and available support provisions.³¹²

This experience may also indicate a lack of cohesion under existing maternity units, to support parents who decide to continue with the pregnancy. A widening of the scope to assess broader systemic deficiencies and disconnect, within maternity units, may permit a fair assessment of whether parents are able to readily access specialist support and counselling, rather than focussing on the conduct of individual HCPs.³¹³

On the other hand, HCPs believe parents typically fail to understand the gravity of the situation, particularly in the context of ES and PS.³¹⁴ Asbury found that some HCPs exhibit surprise or concern upon discovering that parents wish to continue with the pregnancy, post-diagnosis.³¹⁵ Thus, it may be that alterations in professional attitude towards a pregnancy, post-diagnosis of a trisomy, creates a model of care akin to paternalism, rather than partnership.

With parents reporting a medicalisation of their pregnancy post-diagnosis, and a lack of support following their decision to continue with the pregnancy, this begs the question whether embedded institutional attitudes and perceptions of the trisomies encourages directiveness in this regard.³¹⁶ Janvier and Watkins asserted that parents who did not follow the “usual path of termination” reported “incomprehension, negativity and sometimes a lack of support”.³¹⁷ A similar experience was also conveyed by a participant in Guon *et al.*'s research study: *'I was told many times that abortion was definitely the best option for us and I had full support to have an abortion ... but hardly any support for wanting to carry the*

³¹¹ Parent Qualitative Findings, Section 5.4.6.

³¹² Parent Qualitative Findings, Section 5.4.6.

³¹³ Rob Heywood (n5), 440.

³¹⁴ Priscilla Coleman, 'Diagnosis of Fetal Anomaly and the Increased Maternal Psychological Toll Associated with Pregnancy Termination', (2015) 30 Issues in Law and Medicine, 3.

³¹⁵ Bret Asbury, (n198), 304.

³¹⁶ Parent Qualitative Findings, Section 5.4.6; see also, Bret Asbury, (n198), at 334.

³¹⁷ Annie Janvier and Andrew Watkins, 'Medical interventions for children with trisomy 13 and trisomy 18: what is the value of a short disabled life?', (2013) 102 Acta Paediatrica, 1112.

pregnancy'.³¹⁸ As suggested by Coleman, this may endorse a narrative, among the profession, that to continue with a pregnancy, post-diagnosis of ES and PS, is not the 'sensible' option.³¹⁹

This narrative was particularly evident in the decision of *Re L (a Minor)*.³²⁰ The Claimant, in this case, had a baby born with ES. During the course of the pregnancy, the Trusts and parents conflicted in terms of whether to schedule a contingency care plan or withhold treatment, following the birth. In this case, an application was made by two NHS Trusts for a declaration regarding future care plans for L, specifically to withhold treatment. The Claimant felt that the bond between her and the baby, was "*consistently underestimated by the doctors*", and believed that "*the medical profession may be giving up on L too soon*".³²¹ If this is the experience of parents who have babies with ES and PS, it is unsurprising that termination is viewed as the first, and possibly the only, choice in pregnancy.³²²

This rationale is often justified on the basis that parents will experience less psychological trauma ending the pregnancy, compared to losing the baby spontaneously during the pregnancy, or shortly after birth.³²³ However, this research suggests that parents, who end the pregnancy post-diagnosis, experience greater physical and psychological trauma than those who continue with the pregnancy.³²⁴ Indeed, many parents feel their families were enriched and strengthened by the birth of their baby with a trisomy.³²⁵

Agreeably, it would be very unusual for a parent to have a child with a trisomy and characterise the outcome in negative terms: parents with a trisomy will inevitably love their baby, and view the outcome positively.³²⁶ The varying perspectives, in this context, are ultimately subjective, and while there is no 'solution' to this issue, the best approach may be to present both sides of the coin. Presenting information on termination, or returning to the option of termination, may stem from the HCP's duty to maintain a dialogue, and to verify

³¹⁸ Guon J, Wilfond and others, 'Our children are not a diagnosis: The experience of parents who continue their pregnancy after a prenatal diagnosis of trisomy 13 or 18', (2014) 164 Am J Med Genet Part A, 312.

³¹⁹ Priscilla Coleman, (n235), 15.

³²⁰ *Re L (A Minor)* (2004) EWHC 2713 (Fam).

³²¹ *Ibid*, 9.

³²² Parent Qualitative Findings, Section 5.4.6.

³²³ Nora Sullivan and Eoghan de Faoite, 'Psychological Impact of Abortion due to Fetal Anomaly: A Review of Published Research', (2017) 32 Issues in Law & Medicine, 30.

³²⁴ Parent Qualitative Findings, Section 5.4.6.

³²⁵ Parent Qualitative Findings, Section 5.4.6; see also Priscilla Coleman, (n235), 6.

³²⁶ Parent Qualitative Findings, Section 5.4.6.

that the initial decision still stands. Nevertheless, HCPs must be “careful and attentive to potential bias in their communication with patients, particularly to allow the less-likely choices to be made”.³²⁷

The provision of additional information on the decisions and experiences of those with experiential knowledge, in conjunction with the perspective of HCPs, is urgently required in these contexts.³²⁸ It may be that the NHS could facilitate evaluation groups, with these communities and HCPs, to produce specialised resources to further enhance parent decision-making and consent.

7.4.7 Incidental Findings

The identification of incidental/unanticipated findings are often unavoidable in prenatal screening.³²⁹ During the performance of an ultrasound, the sonographer is duty-bound to report any soft markers, or other biological indicators, that are suggestive of the baby being at risk of having a genetic/hereditary condition.³³⁰ Indeed, in the case of *A v The Barts*, the court found in favour of the Claimant for the sonographer’s failure to effectively communicate the risk of having a baby with ES, after soft markers had become known to the sonographer during the examination.³³¹ This was also an issue addressed in *A v East Kent Hospitals*.³³² The Claimant, in this case, revealed she was only aware of DS after consenting to screening, and did not consider the possibility of obtaining a risk score for other chromosomal conditions that may be discovered incidentally.³³³

The sonographer’s overriding duty to report an increased NT measurement to fetal medicine, after parents have *declined* the offer of trisomy screening, is currently creating substantial controversy.³³⁴ While this duty is founded on preserving the health and well-being of both

³²⁷ Anne Moller and others, ‘Danish Sonographers’ Experience of the Introduction of “Moderate Risk” in Prenatal Screening for Down Syndrome’, (2018) 43 *Journal of Pregnancy*, 6.

³²⁸ Lou S, Lanther MR, ‘This is the child we were given: A qualitative study of Danish parents’ experiences of a prenatal Down syndrome diagnosis and their decision to continue the pregnancy’, (2020) 23 *Sex Reprod Healthc*, 480.

³²⁹ Joanne Kelley and others, ‘Increased nuchal translucency after low risk noninvasive prenatal testing: What should we tell the prospective parents?’ (2021) 1 *Prenatal Diagnosis*, 2: Doi.org/10.1002/pd.6024.

³³⁰ *Ibid.*

³³¹ ‘Failure to diagnose Trisomy 18 (Edwards’ Syndrome) from antenatal scan: A v The Barts & London NHS Trust’ (2011) 17 *Journal of Patient Safety and Risk Management*, 1.

³³² *A v East Kent Hospital University NHS Foundation Trust* (2015) EWHC 1038, 38.

³³³ *Ibid.*

³³⁴ HCP Qualitative Findings, Section 6.5.6 and Parent Qualitative Findings, Section 5.4.7.

mother and baby, this overriding duty does raise questions over the ability of parents to act autonomously, and preserve their right not-to-know.³³⁵ Indeed, if parents exercise a choice *not* to receive information on their risk in these contexts, it is arguable that this choice should be respected, in accordance with the doctrines of autonomy and self-determination.³³⁶

The Nuffield Council explain that parents have the choice to decline having their bloods taken, which are necessary to complete the combined test, following a soft marker concern. However, it is highly improbable and unlikely that parents would decline to complete the combined test once they are placed in this situation.³³⁷ The pressure and gravity of the potential risks, to both mother and baby, would prey on the mind of parents, until a diagnosis is sought.³³⁸

While *Montgomery* acknowledged the doctrine as permitting HCPs to withhold information reasonably considered to be detrimental to the patient's health, the Supreme Court left the boundaries of the therapeutic exception largely undefined.³³⁹ Mulheron proposes that, by reference to comparative case law drawn from British, North American and Australian jurisprudence, the defence of therapeutic privilege has three distinct elements.³⁴⁰ Broadly, these elements are: is risk disclosure to the patient would foreseeably be adverse or damaging (divided out into *de minimis* level of damage and reasonable foreseeability); disclosure would have harmed the patient's health or 'best interests'; and the doctor's decision not to disclose was reasonable.³⁴¹ Importantly, *Montgomery* held that the exception should be applied *narrowly*. The application of this exception is subject to the characteristics and circumstance of the particular patient, and should therefore not be applied to patients generally.

The GMC also provide some limited guidance in this regard, highlighting that HCPs must believe the disclosure of information would cause "serious harm".³⁴² As the patient also has

³³⁵ Adeline Perrot and Ruth Horn (n295), 4.

³³⁶ Parent Qualitative Findings, Section 5.4.7.

³³⁷ HCP Qualitative Findings, Section 6.5.6 and Parent Qualitative Findings, Section 5.4.7.

³³⁸ Parent Qualitative Findings, Section 5.4.7; see also, Heyman B, Hundt G, Sandall J, Spencer K, Williams C, Grellier R, and Pitson L, "On being at higher risk: A qualitative study of prenatal screening for chromosomal anomalies", *Social science & medicine*, 2006, 62(10): 2360-2372.

³³⁹ *Montgomery* (n1), 88.

³⁴⁰ Rachel Mulheron, "Has Montgomery Administered the Last Rites to Therapeutic Privilege? A Diagnosis and a Prognosis" (2017), 2.

³⁴¹ *Ibid.*

³⁴² General Medical Council Guidelines on Consent (2008), 16-17.

a right not-to-know³⁴³, the GMC states that, “if the patient insists they do not want ... basic information, (HCPs) must explain the potential consequences of them not having it, particularly if it might mean that their consent is not valid”.³⁴⁴ The GMC suggests that HCPs must record that the patient declined the information, and make it clear that they can change their minds, at any point, to have more information.³⁴⁵

The Australian and Canadian courts recognised that situations may arise where a patient does not want to hear or accept unexpected news from the HCPs. The courts suggested that the HCPs must take reasonable precautions to document that the patient has clearly expressed they do not wish to be told the information. If the patient’s health presents as precarious, and that the disclosure of information would certainly trigger an adverse outcome causing unnecessary harm, the court outlined this may justify its non-disclosure.

7.4.8 Remembering the Partner’s Influence and Inclusion

Ultimately, the pregnant person holds the greatest degree of discretion and autonomy over their reproductive choices.³⁴⁶ While this statement is relatively commonplace, it would be an oversight not to also consider the influence of partners on decision-making. Upon review of the literature in this area, it is apparent that research into the partners’ influence on decision-making and consent for trisomy screening, is sparse.³⁴⁷

This research revealed that partners play an influential role on the process of decision-making for trisomy screening.³⁴⁸ With participants expressing that they valued their partner’s input *above* that of the healthcare staff when making decisions on trisomy screening, this suggests that the partners’ impact on decision-making and choice, in these contexts, has historically been underestimated.³⁴⁹

³⁴³ Montgomery, (n1), 85.

³⁴⁴ GMC, (n342), 15.

³⁴⁵ Ibid.

³⁴⁶ Alys Einion, *The medicalisation of childbirth: The Social Context of Birth* (3rd Edn, Routledge, 2017), 12.

³⁴⁷ Wätterbjörk, I. and others, ‘Decision-making process of prenatal screening described by pregnant women and their partners’, (2015) 18 *Health Expectations: An International Journal of Public Participation in Health Care & Health Policy*, 1582.

³⁴⁸ HCP Qualitative Findings, Section 6.5.9 and Parent Qualitative Findings, Section 5.4.8.

³⁴⁹ Parent Qualitative Findings, Section 5.4.8; see also, Y. Severijns and others, ‘To what extent do decision aids for prenatal screening and diagnosis address involvement of partners in decision-making? – An environmental scan’, (2021) 104 *Patient Education and Counseling*, 2952.

With this in mind, logic dictates that the partner's inclusion during pre-screening consultations, is a key factor going to decision-making. However, partners felt marginalised at consultations, and that the HCP's focus rested on the pregnant person.³⁵⁰ It would be unsophisticated to conclude that the partner's marginalisation was an intentional act by HCPs; this may speak to the lack of time at consultations to effectively promote and manage partners' input to decision-making.³⁵¹ It may also suggest a lack of available training on endorsing the partners' interests during counselling, consistent with this study's findings.³⁵²

The creation of a *partner*-specific clinic may be necessary to support, promote and manage the inclusion of partners during the decision-making process for trisomy screening.³⁵³ Implementing a partner-specific information packs and leaflets could encourage the desired inclusion, and may enable HCPs to effectively manage existing time constraints. The information could proffer suggestions to partners on managing their role, placing a focus on how to support the pregnant person in the event of a typical or atypical screening result, issues and considerations pertaining to paternal mental health, and support available to partners in the event of a positive trisomy diagnosis. Information on common questions from partners could also be a useful addition to the materials. Partner-specific clinics may also support the HCPs to monitor and manage any signs of coercion or pressure from partners during the decision-making process.³⁵⁴

³⁵⁰ Parent Qualitative Findings, Section 5.4.8.

³⁵¹ Anne-Marie Laberge and others, 'Canadian Pregnant Women's Preferences Regarding NIPT for Down Syndrome: The Information They Want, How They Want to Get It, and With Whom They Want to Discuss It' (2019) 41 J Obstet Gynaecol Can, 782.

³⁵² HCP Qualitative Findings, Section 6.5.9.

³⁵³ Fenton SK, Joscelyne T and Higgins S, 'Part 1: exploring views from fathers and perinatal practitioners on the inclusion of fathers by perinatal services', (2021) 29 British Journal of Midwifery, 1; and, Fran E Carroll and others, 'A qualitative investigation of the decision-making process of couples considering prenatal screening for Down Syndrome', (2012) 32 Prenat Diagn, 57.

³⁵⁴ Fran E Carroll and others, 'A qualitative investigation of the decision-making process of couples considering prenatal screening for Down Syndrome', (2012) 32 Prenat Diagn, 63.

7.5.0 Delineating Communication in the HCP-Parent Relationship: Role of the Midwife and Sonographer

While effective communication between HCPs and patients is vital to the *Montgomery*, *Mordel* and GMC shared decision-making model of care, sonographers and midwives experience difficulties executing this duty, in the context of trisomy screening.³⁵⁵ The overriding theme, in *Mordel*, was that negligence manifested within the disconnect and miscommunication between the sonographer and midwife, and ultimately the Claimant. However, communication between sonographers and parents is limited to brief introductions, and to merely record the decision from the booking appointment: to communicate with parents at the level expected in *Mordel*, is not established practice, according to sonographers in this study.³⁵⁶

Jay J outlined that logic mandates “*checking that there has been a discussion between patient and midwife*” before the sonographer secures consent.³⁵⁷ This requirement raises two distinct questions: (i) should the onus be on the patient or sonographer to confirm that discussion has previously occurred with the midwife; and (ii) what information should have been communicated within this ‘discussion’. With all the other information on pregnancy care, and regular consultations with HCPs during early pregnancy, the question remains whether it is reasonable or logical to place the onus upon the parent to confirm that discussion, on DS screening, has occurred with the midwife, or if the sonographer should take reasonable steps to confirm this themselves.

Mordel threw into question the ‘established’ system of communication between the parent and sonographer for the purposes of obtaining informed consent, and whether it was ‘reasonable’. Jay J underlined that, “*asking a short question and hearing the answer (maybe 3 seconds); explaining briefly what is about to happen and inviting the patient to lie down (maybe 5-6 seconds); and then turning to the computer and selecting the appropriate dropdown menu (a few more seconds ...)*”, was an inadequate system of communication between sonographer and parent.³⁵⁸

³⁵⁵ HCP Qualitative Findings, Section 6.6.1.

³⁵⁶ HCP Qualitative Findings, Section 6.2.1.

³⁵⁷ *Mordel* (n14), 89.

³⁵⁸ *Ibid*, 98.

Cooper v Royal United Hospital emphasised that the law is unable to guarantee that HCPs will be skilled communicators.³⁵⁹ However, *Deriche v Ealing Hospital Trust* asserted that effective communication, between HCPs and patients, is crucial to supporting informed decision-making.³⁶⁰

Robertson outlined the importance of establishing systems of communication between HCPs and units, in clinical practice.³⁶¹ This case suggested that establishing a systemic duty, rather than focussing on the individual error of HCPs, would better enhance the development and fortification effective communication.³⁶² *Robertson* explained that narrowing the scope to an individual breach of duty may cloud the fact that broader systemic failings exist, under which HCPs are expected to work.³⁶³

While *Mordel* alluded to possible systemic negligence, in the context of systems of communication between midwife, sonographer and parent, the question remains whether a ‘reasonable’ system of communication places an expectation to synthesise the sonographer’s duty with that of the midwife. Indeed, time and resource limitations during the examination appointment, compounded by the lack of training sonographers receive on ‘counselling’ parents, forces one to return to the suggestion that a *Bolitho* ‘reasonable’ system must be established to enhance and synthesise interdisciplinary practice, for the purpose of effectively securing parent consent.

7.5 Delineating Communication in the HCP-Parent Relationship: Key Considerations

7.5.1 A Growing Friction Between Consumerism and Montgomery’s Shared Decision-Making

The provision of healthcare, in the context of screening, is an “ideological mess”.³⁶⁴ This research unearthed a continuum in this regard, with a desire for HCPs to act solely as neutral information providers on one end, and a preference for HCPs to act as advisory figures on the

³⁵⁹ *Cooper v Royal United Hospital Bath NHS Trust* (2005) EWHC 3381.

³⁶⁰ *Deriche v Ealing Hospital NHS Trust* (2003) EWHC 3104.

³⁶¹ *Robertson v Nottingham Health Authority* (1997) 8 Med LR 1.

³⁶² *Ibid.*

³⁶³ *Ibid.*

³⁶⁴ Tara Latimer, Joseph Roscamp and Andrew Papanikitas, ‘Patient-centeredness and consumerism in healthcare: an ideological mess’, (2017) 110 *Journal of the Royal Society of Medicine*, 425.

other.³⁶⁵ Parents also fell in the middle of this continuum, preferring a model of care based on shared decision-making.³⁶⁶ These divergent preferences support a growing body of research which suggest our model of healthcare is comprised of differing values and ideologies.³⁶⁷

Scholars are beginning to gauge a growing friction between differing ideologies on the provision of healthcare.³⁶⁸ Indeed, the GMC and *Montgomery's* desired model of care is founded on values consistent with shared decision-making; however, parents' perception of the provision of healthcare currently stands as a 'one-stop' shop: parents compare selecting treatment options with purchasing a product from a market or business.³⁶⁹ It also seems that the HCP's role is becoming increasingly synonymous with 'Google', that being a passive source of information and lacking the desired interactivity.³⁷⁰

Indeed, a misunderstanding on the part of HCPs to avoid 'directiveness', is leading HCPs to adopt a model of care based on "passivity and defensive avoidance".³⁷¹ Non-directiveness has been framed as an 'active strategy', requiring HCPs to harness their counselling skills to empower patient autonomy and self-directiveness, rather than adopting defensive practices.³⁷² If this is the preferred model of care, by both HCPs and parents, the emergence of a business-like or consumerist model of healthcare – diverging from the GMC and *Montgomery* values, to promote interactivity and shared decision-making – is currently underway.³⁷³

Latimer *et al.* explain that the philosophy underpinning consumerism is based primarily on autonomy, with its provenance gauged from principles of liberty, established by John Mill, "... the liberty of thought and feeling ... (and) doing as we like, subject to such consequences as may follow ... even though they should think our conduct foolish, perverse or wrong".³⁷⁴

³⁶⁵ HCP Qualitative Findings, Sections 6.6.1 and 6.6.2 and Parent Qualitative Findings, Section 5.5.1.

³⁶⁶ Parent Qualitative Findings, Section 5.5.1.

³⁶⁷ Tara Latimer (n364), 426

³⁶⁸ *Ibid*

³⁶⁹ HCP Qualitative Findings, Section 6.6.1 and Parent Qualitative Findings, Section 5.5.1; see also; Emily Jackson (n96), 14.

³⁷⁰ HCP Qualitative Findings, Section 6.6.1 and Parent Qualitative Findings, Section 5.5.1.

³⁷¹ HCP Qualitative Findings, Section 6.6.2; see also, Sanne L. van der Steen (n146), 235.

³⁷² *Ibid*.

³⁷³ Parent Qualitative Findings, Section 5.5.1.

³⁷⁴ Tara Latimer (n364), 426.

Under a consumerist model of care, Arvind and McMahon explain that this seldom entails shared decision-making, but rather “the choice is the customer’s alone, and the provider’s role is to simply provide any information the customer might require to make a choice”, and distances HCPs from the decision-making process.³⁷⁵

However, while *Montgomery* is often cited as empowering a model of care based on shared decision-making, the decision itself used language consistent with a consumerist model of care, and arguably based its decision on principles of consumerism.³⁷⁶ Indeed, the decision drew an analogy between patients and consumers, in the context of information disclosure, ‘patients ... are also widely treated as consumers exercising choices: a viewpoint which has underpinned some of the developments in the provision of healthcare services’.³⁷⁷ Indeed, Arvind and McMahon argue that the decision in *Montgomery*, in fact, diverges from the GMC’s intended model based on partnership, between professional and patient.³⁷⁸

One explanation for the divergent approaches may be due to a lack of a single cogent definition of ‘patient-centred care’, as various interpretations of this concept have been appropriated.³⁷⁹ Cave highlights, “competition resulted in a mix of private and state-run NHS organisations which ... made the NHS more business-like and consumeristic”.³⁸⁰ However, Latimer et al. opine that, while “consumerism is explicitly used as a vehicle for the emancipation of patients in the name of patient-centred care ... in its purest form, patient-centred care never saw health as a commodity that could be bought and sold, dependent on the response to consumer choice for survival”.³⁸¹

While consumerism intends to promote self-determination, Teff argues that a model of care, that prioritises principles of self-determination, is as flawed as paternalism, due to the fact that it may hinder the HCPs from forming a therapeutic alliance or partnership for decision-

³⁷⁵ T. T. Arvind and Aisling M. McMahon, ‘Responsiveness and the Role of Rights in Medical Law: Lessons from *Montgomery*’, (2020) 28 *Medical Law Review* 445, 452.

³⁷⁶ Emma Cave and Caterina Milo, ‘Informing patients: The *Bolam* Legacy’, (2020) 20 *Medical Law International*, 103.

³⁷⁷ *Montgomery*, (n1) 75.

³⁷⁸ T. T. Arvind and Aisling M. McMahon, (n375), 452.

³⁷⁹ *Ibid*, 426.

³⁸⁰ Emma Cave and Caterina Milo, (n376), 119.

³⁸¹ Tara Latimer (n364), 426.

making.³⁸² Indeed, Teff posits that when the transmission of information is formal and impersonal, this replaces the necessary interactive and genuine dialogue between professional and patient.³⁸³ Jackson observes that the analogy drawn by the Supreme Court in *Montgomery*, between patients and consumers, is interesting “given the increasing recognition that consumers fail routinely to understand and use information disclosures”.³⁸⁴

In an era of responsive law, consumerism arguably has no place in trisomy screening practices. Indeed, HCPs must act as more than neutral information providers, particularly in contexts of epistemic uncertainty: many parents will not be familiar with trisomy screening.³⁸⁵ While those parents who were higher-risk, or had a positive diagnosis, demonstrated a stronger preference for a neutral information provider approach, their epistemic uncertainty and the unfamiliar circumstances would arguably have required the HCPs to adopt more than a purely information-provider approach.³⁸⁶ In these stressful and emotive circumstances, it would be highly undesirable for HCPs to appropriate a ‘google-type’ role, with their duty being devoid of any shared decision-making values and interactivity, and consisting of being only a passive source of information.³⁸⁷ This duty would point towards a retailer-customer relationship, and that being a far less intimate relationship than HCP and patient.³⁸⁸

It may be that their experience of screening has coloured their opinion in this regard, which points more towards issues of distrust, rather than preferring a consumerist model of care; this also alludes to an ‘us against them’ culture.³⁸⁹ Instead of distancing HCPs from decision-making, which would only exacerbate a culture of this type, establishing a *partnership* between HCPs and parents tackles epistemic uncertainty as a joint force, enhancing parents’ ability to make informed decisions.³⁹⁰ However, access to effective training protocols and

³⁸² Harvey Teff, ‘Consent to Medical Procedures: Paternalism, Self-Determination or Therapeutic Alliance?’ (1985) 101 Law Quarterly Review, 432.

³⁸³ *Ibid.*

³⁸⁴ Emily Jackson, (n96), 3.

³⁸⁵ T. T. Arvind and Aisling M. McMahon, (n375), 445.

³⁸⁶ Parent Qualitative Findings, Section 5.5.1.

³⁸⁷ *Ibid.*

³⁸⁸ Emily Jackson, (n96), 5.

³⁸⁹ HCP Qualitative Findings, Section 6.6.4 and Parent Qualitative Findings, Section 5.5.1.

³⁹⁰ T. T. Arvind, (n 375), 460.

guidelines are essential in this regard: a provision that has been lacking since the development of the trisomy pathway.³⁹¹

To the contrary, overdependence on HCPs is equally damaging. The dependency on HCPs' discretion – exhibited by parents who fell within the advisory and shared decision-making groups – may be borne from the increasing complexities of the screening pathway, medical technology, and knowledge.³⁹² The growing complexities, within the HCP-parent relationship, means that professional involvement in the decision-making process is becoming increasingly common, pointing to the potential benefits of selective paternalism.³⁹³

While parents want to retain the ability to make their own choices, when confronted with difficult or lifechanging decisions, parents' vulnerability and imbalance in expertise changes the dynamic of the HCP-parent partnership, possibly requiring a higher degree of professional dependency.³⁹⁴ Recent amendments to the screening pathway, such as the inclusion of additional conditions and new methods of screening, demands high standards of medical literacy from parents; this demand may reduce parents' levels of confidence to exercise informed choices, and preferring HCPs to hold the power in this dynamic. However, while growing complexities within the HCP-parent relationship may require increasing professional involvement in the decision-making process, this does not necessarily point to a "damaging" paternalistic model of care, as Hertig *et al.* suggests.³⁹⁵ Instead, this may point to the parents' and HCPs' joint endeavour to achieve the same healthcare objectives, promoting transparency, and addressing imbalances in knowledge and expertise.

7.5.2 Time Limitations and Parent-HCP Communication

A partnership between HCPs and parents requires the construction and fortification of rapport, founded upon interactive and meaningful supportive dialogue, operating under a

³⁹¹ Priscilla Coleman, "Diagnosis of Fetal Anomaly and the Increased Maternal Psychological Toll Associated with Pregnancy Termination", (2010) 30 *Issues in Law and Medicine*, 8.

³⁹² HCP Qualitative Findings, Section 6.6.1 and Parent Qualitative Findings, Section 5.5.1.

³⁹³ HCP Qualitative Findings, Section 6.6.1; see also, Solene Gouiher Hertig and others, 'Doctor, what would you do in my position?' Health professionals and the decision-making process in pregnancy monitoring', (2014) 40 *J Med Ethics*, 310.

³⁹⁴ Parent Qualitative Findings, Section 5.5.1; see also, Aune, I. and Möller, A., 'I want a choice, but I don't want to decide'—A qualitative study of pregnant women's experiences regarding early ultrasound risk assessment for chromosomal anomalies', (2012) 28 *Midwifery*, 14.

³⁹⁵ Solene Gouiher Hertig, 'Doctor, what would you do in my position?' Health professionals and the decision-making process in pregnancy monitoring', (2014) 40 *J Med Ethics*, 310.

‘reasonable’ system.³⁹⁶ However, time limitations risk interrupting or halting the desired interactivity completely.³⁹⁷ Counselling is becoming progressively demanding and time-consuming, principally due to the increasing complexity of the information that needs to be reviewed with parents, subject to recent developments in screening technology.³⁹⁸

Time limitations are responsible for the impersonal relationship parents report having with HCPs, with comparisons being drawn to a ‘revolving door’ system of trisomy screening, or on a ‘conveyor-belt’ of care.³⁹⁹ This research offers a timely illustration of the need for health services to review the impact of time limitations, in conjunction with systemic considerations, to assess the ‘reasonableness’ of the HCP’s duty to secure parent consent.

The desired partnership will be increasingly challenged by ever-constricting time limitations, in the wake of the implementation of two conditions, and a novel method of screening to the trisomy pathway. These additions have broadened the HCPs scope of information disclosure; however, the time afforded by the health service for HCPs to meet the requisite standards for information disclosure remains unchanged.

Parents’ comparison to a factory-type system of communication and care, alludes to the impracticality and unreasonableness of established systems to effectively secure consent, under growing time constraints, while maintaining a genuine and meaningful dialogue.⁴⁰⁰ Indeed, parent consent and decision-making for screening is at risk of becoming a mere formality. While Jay J acknowledged that the demand on HCPs to review an ever-increasing amount of information, within limited time periods, is indeed challenging, he failed to assess the true impact of time constraints on the ability of HCPs to perform their duty, whilst maintaining the desired interactivity and relationship between HCPs and parents.

Sonographers experience crushing time pressures to perform their role, in accordance with legal and professional expectation: introductions, performance of the scan, reporting the findings and review of the examination, currently takes place in a twenty-minute

³⁹⁶ McCourt C, ‘Supporting choice and control? Communication and interaction between midwives and women at the antenatal booking visit’, (2006) 62 *Social Science & Medicine*, 986.

³⁹⁷ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.6.3 and Parent Qualitative Findings, Section 5.5.2.

³⁹⁸ *Ibid.*

³⁹⁹ Parent Qualitative Findings, Section 5.5.2.

⁴⁰⁰ Parent Qualitative Findings, Section 5.5.2.

appointment.⁴⁰¹ Thus, the inclusion of Jay J's additional recommendation to also counsel and/or re-counsel parents on issues of information disclosure, understanding and choice, is fundamentally impractical and unrealistic.

Ideally, solving this issue points to providing additional time for HCPs to fulfil their duty. However, under *Bolitho's* assessment of reasonableness, developments in screening technology and knowledge will only add to existing complexities for the provision of counselling, exacerbating time pressures at consultations.⁴⁰² A sensible suggestion would be to review current clinical guidelines in line with established systems of communication, outlining what is *reasonably* expected from the HCPs. A *Bolitho* assessment of 'risk against benefit' should also encompass balancing the interests of executing HCPs' duty with maintaining interactivity, to achieve the desired partnership with parents. Making best use of the allocated time at consultations must be the objective of the review.

7.5.3 Allocation of Resources, Adversarial Interactions and its Impact on HCP-Parent Relationship

Most parents' experience of trisomy screening will be relatively unremarkable: in these contexts, the HCP-parent relationship is typically unproblematic and unchallenged.⁴⁰³ Indeed, medical research into the experience of lower-risk parents for DS screening, often conclude that the relationship between HCPs and parents is orderly, and routinely achieves the desired partnership.⁴⁰⁴ However, this is not representative of all parent groups.⁴⁰⁵ Cases, such as *Mordel*, underline the significance of delving into stakeholders' varying experiences of trisomy screening, for an appreciation of where challenges may raise for HCPs, to maintain the desired consent dialogue.

There is currently a conflict and/or frustration exhibited by parents, particularly those who had a baby with a trisomy, towards HCPs from their experience of decision-making and consent; this is unsurprising, as the emotive and high-pressured nature of trisomy screening

⁴⁰¹ HCP Qualitative Findings, Section 6.6.3 and Parent Qualitative Findings, Section 5.2.2.

⁴⁰² Sanne L. van der Steen (n146), 235.

⁴⁰³ Johan Christiaan Bester, 'Defensive practice is indefensible: how defensive medicine runs counter to the ethical and professional obligations of clinicians', (2020) 23 *Medicine, Health Care and Philosophy*, 413.

⁴⁰⁴ Seror. V, and Ville. Y, 'Prenatal screening for Down syndrome: women's involvement in decision-making and their attitudes to screening', (2019) 29 *Prenatal Diagnosis*, 120.

⁴⁰⁵ Parent Qualitative Findings, Section 5.5.3.

foreseeably induces adversarial interactions between parents and professionals, particularly in the context of ES and PS.⁴⁰⁶ However, the question as to *why* decision-making, in the context of the trisomies, causes such friction between HCPs and parents, remains largely unanswered.

Many academics, who conduct extensive research in this area, superficially conclude that the negative, biased and outdated attitudes of the medical community, following a diagnosis of a trisomy, sparks the conflict, and results to a breakdown in communication. While this may be true of some misguided HCPs, this would arguably be rare, and would not be conducive with all reported cases.

Undue focus upon the individual conduct of HCPs, risks overlooking systemic constraints and commitment to beneficence, under which they are required to communicate and counsel parents on their decision-making after a positive diagnosis. Adversarial interactions often ensue during counselling on whether to continue or end the pregnancy, subject to accessibility to specialist care and allocation of resources, in anticipation of birth: this narrative was particularly prominent in the context of care management for ES and PS.⁴⁰⁷ McCaffery explains that, as a result of ES and PS being labelled 'lethal', the provision of life-saving treatments are often withheld, and therefore the option to terminate is normalised.⁴⁰⁸

The case of *Bull v Devon Health Authority* conveyed how broader resource limitations may impact on HCPs' ability to provide the desired care.⁴⁰⁹ This decision outlined how resource limitations should be considered by the courts as a key factor going to the workability and reasonableness of NHS systems.⁴¹⁰ While this case concerned emergency obstetric care, it highlighted that the systems of communication between units were defective, primarily due to resource limitations, which should have been communicated to the Claimant as part of their care management.

⁴⁰⁶ HCP Qualitative Findings, Section 6.6.4 and Parent Qualitative Findings, Section 5.5.3.

⁴⁰⁷ HCP Qualitative Findings, Section 6.6.4 and Parent Qualitative Findings, Section 5.5.3.

⁴⁰⁸ Martin McCaffrey, 'Lives Worth Living', (2017) 32 *Issues in Law & Medicine*, 215.

⁴⁰⁹ *Bull v Devon Area Health Authority* (1993) 4 *Med LR* 117. See also, Charles Foster, 'Simple rationality? The law of healthcare resource allocation in England' (2007) 33 *Journal of Medical Ethics*, 404.

⁴¹⁰ *Ibid.*

However, there is a clear lack of appreciation by parents of the prognosis and health outcomes of the trisomies.⁴¹¹ If parents perceive ES and PS the same or just ‘worse versions’ of DS, it is likely that they do not fully understand the complex and multifaceted health needs of a baby with ES and PS, and the demand this outcome would have on available resources.⁴¹² Parents perceive communicating the *unavailability* of specialist resources as not caring for, or valuing, their child, and being demonstrative of a culture of ‘bias’ against this community.⁴¹³ They also believe that communicating the option to withhold treatment and access to specialist resources, in anticipation of the birth of their child, implies they should end the “hopeless” pregnancy, as the child’s life is “not worth living”.⁴¹⁴

While it is understandable that parents feel this way, disentangling HCPs’ and parents’ competing interests, to assess the professions’ justification and reasoning, is required. Hedley J, in *Re Wyatt (a child) (medical treatment: parents’ consent)*, had to assess the issue of decision-making and consent, following a disagreement between the parents and the Trust, on neonatal care management and planning.⁴¹⁵ An application was made by Portsmouth NHS Trust not to artificially ventilate, or otherwise aggressively treat, a baby born prematurely. Following initial consultations with the HCPs, the baby’s parents opposed this application. Hedley J explained that there are four stages in which a clinician could be in disagreement with the parents, in terms of whether to offer access to specialist care and resources.⁴¹⁶ These stages are:

*‘the position where a course of treatment was advised but rejected by the parents where the clinician concluded that it was an affront to professional conscience to withhold that treatment. The second stage was where the same circumstances existed but where the clinician although disagreeing with the parents could see that there was something to be said for their view. The third is the reverse of the second where the clinician advises against treatment which the parent wants but is able in conscience nevertheless to give it. The last stage is where to do what the parents want is not possible in good conscience’.*⁴¹⁷

⁴¹¹ Parent Qualitative Findings, Section 5.5.3.

⁴¹² HCP Qualitative Findings, Section 6.6.4.

⁴¹³ Parent Qualitative Findings, Section 5.5.3.

⁴¹⁴ Parent Qualitative Findings, Section 5.5.3.

⁴¹⁵ *Re Wyatt (a child) (medical treatment: parents’ consent)* (2004) EWHC 2247 (Fam).

⁴¹⁶ *Re Wyatt (a child)* (n415), 18.

⁴¹⁷ *Ibid.*

The instant case fell into the third stage, with the HCPs expressing the view that the treatment options available for the baby were not worthwhile, due to her developmental delays and prognosis, contrary to parental opinion.⁴¹⁸ After consideration of the principles of the sanctity of life and the ‘intolerable to that child’ concept, Hedley J held that it was not in the best interests of the baby to allocate aggressive life-saving intervention.⁴¹⁹

These principles were applied by Dame Elizabeth Butler-Sloss P, in *Re L (A Minor)*.⁴²⁰ She held that, “... *the test is ‘best interests’ which are interpreted more broadly than ‘medical interests’ and include emotion and other factors. There is a strong presumption in favour of preserving life, but not where treatment out be futile, and there is no obligation on medical professionals to give treatment which would be futile*”.⁴²¹ The President, in this case, implemented a benefits and disbenefits method of assessing the baby’s best interests, and whether potential treatment options should be offered.⁴²² *Obiter dictum*, Dame Elizabeth Butler-Sloss P stated that the “*mother had consistently and resolutely helped her son fight for his life*”, which brought “*considerable tensions between the mother and professionals*” during the course of the pregnancy, and into the period of postnatal care.⁴²³ The court found that it was in the best interests of the baby for the treatment options to remain open; however, “... *it is the duty of the mother, for the sake of L., to reduce those areas of conflict to a minimum, and to listen to what is proposed by those who have a great deal of medical experience*”.⁴²⁴

While *Re L (A Minor)* adopted a more holistic and nuanced approach than *Re Wyatt (a child)* to the assessment of best interests, this historical conflict between HCPs and parents may be borne from the courts’ failure to “*de-Bolamise best interests*” in the assessment of allocation and access to available resources.⁴²⁵ As Braizer explains, while *Bolam* is not strictly applied in

⁴¹⁸ Antje Pedain, ‘Doctors, parents, and the courts: legitimising restrictions on the continued provision of lifespan maximising treatments for severely handicapped, non-dying babies’, (2005) 535 *Child and Family Law Quarterly*, 1.

⁴¹⁹ *Re Wyatt (a child)* (n415), 24.

⁴²⁰ *Re L (A Minor)* (2004) EWHC 2713 (Fam).

⁴²¹ *Ibid*, 12.

⁴²² *Ibid*, 13.

⁴²³ *Ibid*.

⁴²⁴ *Ibid*, 32.

⁴²⁵ Margot Braizer, ‘An Interactable Dispute: When Parents and Professionals Disagree’, (2005) 13 *Medical Law Review*, 415.

this domain, its principles are certainly pervasive and have considerable impact in this regard.⁴²⁶

Braizer notes that “Charlotte Wyatt (the baby) will be just one of many babies needing specialist care ... the hidden spectre in all such tragic conflicts between parents and professionals is resources.”⁴²⁷ She concludes by underlining that “no hint is given in the judgment (*Re Wyatt*) that either party contemplated the presence of that spectre ... parents will fear that decisions about their baby are driven by economic concerns”, a point raised by parents of babies with ES and PS, in this study.⁴²⁸

Furthermore, Pedain argues that the courts have not appropriately distinguished between a “medically defensible and a medically indefensible parental treatment preference”.⁴²⁹ While many judges, in this domain, will inevitably appropriate the role of mediator to reconcile tarnished relationships, a lack of legal framework, compounded by the courts’ focus on justifying HCPs’ preferred choice in these circumstances, may catalyse parent and HCP conflict.⁴³⁰

Based on previous court decisions, Pedain surmises that courts attach greater weight to parental preferences on treatment refusal, and will enforce treatments on children, contrary to parental preference, only if parental preference is grossly unreasonable.⁴³¹ Conversely, the courts attach lesser weight to parental preferences if parents request costly treatments and resources, which HCPs and the hospital are reluctant to provide.⁴³² However, to conclude that this indicates a subtle judicial preference, as Pedain suggests, would be misguided; this is more likely to be an attempt at reflecting the reality and workability of NHS systems: the courts’ assessment must consider the allocation of finite NHS resources and capacity in antenatal, perinatal and neonatal care, which are often required to support multifaceted or complex health needs, such as ES and PS.

⁴²⁶ Ibid, 415 in footnote (14).

⁴²⁷ Ibid, 418.

⁴²⁸ Ibid, 418.

⁴²⁹ Antje Pedain, (n418), 535.

⁴³⁰ Margot Braizer, (n425), 418.

⁴³¹ Antje Pedain (n418), 535.

⁴³² Ibid.

The emergence of consumerism may also encourage a culture of expectation and unmitigated access to specialist resources.⁴³³ Latimer opines that “a consumerist model gives the patient what they want out of obligation .. but in a resource-strict setting ... this is not always achievable and so limits to patient choice are unavoidable”.⁴³⁴ Cave observes that, “consumerism can also lead to unmet patient expectations: it denotes a wide range of choice that is in practice limited ... choice is limited by resources, evidence of clinical need and beneficent obligations”.⁴³⁵ This issue was also acknowledged in *Montgomery*, stating that, “... the treatment which they can offer is now understood to depend not only upon their clinical judgment, but upon bureaucratic decisions as to such matters as resource allocation, cost-containment and hospital administration: decisions which are taken by non-medical professionals.”⁴³⁶

Both HCPs and parents bring different considerations to the table: “doctors in the issue’s biomedical dimension, and patients in how it affects the achievement of their aspirations”.⁴³⁷ Recognition of these plural values underlines the importance of a shared decision-making model of care, consistent with the GMC. This desired partnership, a dimension missed in *Montgomery*, must be championed by screening leads and programme co-ordinators, when seeking to address issues of best interests and decision-making.⁴³⁸ HCPs’ reporting a lack of training, in this context, must also be addressed.⁴³⁹ Building trust and improving communication is possible with supportive behaviours, realistic and compassionate support, and family-centred care.

⁴³³ Parent Qualitative Findings, Section 5.5.3.

⁴³⁴ Tara Latimer (n364), 426.

⁴³⁵ Emma Cave and Caterina Milo (n376), 119.

⁴³⁶ *Montgomery* (n1) 75.

⁴³⁷ T. T. Arvind and Aisling M. McMahon, (n375), 461.

⁴³⁸ *Ibid.*

⁴³⁹ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.6.3; see also, Johan Christiaan Bester, ‘Defensive practice is indefensible: how defensive medicine runs counter to the ethical and professional obligations of clinicians’, (2020) 23 *Medicine, Health Care and Philosophy*, 2020, 413.

7.6.0 Counselling Parents on ‘Risk’: Situating Professional Duty

There is currently a lack of clarity surrounding HCPs’ duty to counsel parents on ‘risk’.⁴⁴⁰ Traditionally, at the booking appointment, midwives would provide a broad overview of ‘risk’ and what this means, in the context of trisomy screening and/or testing; this duty did not fall to the sonographer at the examination appointment.⁴⁴¹ Nevertheless, it remains unclear whether the sonographer’s duty extends to counselling the parents on ‘risk’, in the context of both screening and diagnostic testing, following the decision in *Mordel*.

Counselling parents on ‘risk’, if done properly, should demand significantly more from the HCPs than ‘checking’ if parents understand the purpose of screening, as per *Mordel*. As the midwives underlined in the findings, this is arguably the most important element of screening for parents to understand, and significant time must be dedicated to exploring the information and parents’ understanding thereof.⁴⁴²

Risk, for screening, should include discussion on population and individual risk, and how these are calculated, following the performance of the ultrasound and serum analysis. A distinction should then be drawn between the screening high-and low-risk factors, and the need for diagnosis through testing. Naturally, this should lead counselling onto the fact that some screening tests do not reflect the diagnosis: false-positives and negatives are a possibility, due to the fact that ‘screening’ is unable to provide a definitive diagnosis.

Thus, it is clear that counselling parents on ‘risk’ is an arduous undertaking, for any HCP, at time-pressured appointments.⁴⁴³ Jay J briefly surmised that ‘risk’ is an issue of case-specific evaluation, tailored to the particular Claimant; however, the judgment did not reflect, or reasonably assess, the time required to perform this duty.⁴⁴⁴ On the issue of supporting parents’ understanding of ‘risk’, Jay J preferred the practice of the Claimant’s expert witnesses: *“both would make clear that the procedure about to be performed is an ultrasound for screening purposes involving no more than the measurement of the thickness of the foetal neck: this information, together with a blood test, will lead to the acquisition of more accurate*

⁴⁴⁰ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.7.1.

⁴⁴¹ HCP Qualitative Findings, Section 6.7.1.

⁴⁴² HCP Qualitative Findings, Section 6.7.3.

⁴⁴³ HCP Qualitative Findings, Section 6.7.3.

⁴⁴⁴ *Mordel* (n14), 148.

data as to the risk of Down's Syndrome"; however, this is insufficient, by itself, for a proper understanding of 'risk'.⁴⁴⁵

In addition, Jay J did not outline whether the sonographer, midwife, or both were required to support parents' understanding of 'risk' before acquiring consent. Thus, the question remains whether the duty of sonographers extend to counselling parents on 'risk', or to merely outline that the procedure (ultrasound scan and blood test) will produce a risk-score. Indeed, one could argue that to merely outline that the procedure will produce a risk-score, without counselling parents on what 'risk' means – consistent with Jay J's preferred practice – seems illogical, as it would not effectively support parent understanding.

It is foreseeable that Jay J's preferred approach to practice will prompt parents to ask questions on 'risk' at the scanning stage, subject to the counselling they received from the midwife at the booking appointment. To counsel and support parent understanding of 'risk', at the examination appointment, would be a significant undertaking for sonographers, as this is a complex notion and would require additional time. Under *Mordel's* 'reasonable' system, sonographers would be required to review a number of considerations at this stage in the pathway: why parents undertake trisomy screening and testing; how 'risk' is framed; the purpose of obtaining a risk status; and consequences of a higher-risk screening result. Sonographers are under increasing time pressures to effectively perform the scan within the allocated appointment, even *without* having to counsel parents on risk. Thus, the question remains whether it is reasonable or logical for sonographers to dedicate time to counsel parents on 'risk', under established systems of care.

7.6 Counselling Parents on 'Risk': Key Considerations

7.6.1 Trivialising the 'Risk' of ES and PS

Under the trisomy screening pathway, parents receive information on the prevalence of each condition within the general population: DS being the most common trisomy, with ES and PS following in second, and third place respectively. ES and PS were introduced to the traditional DS screening programme, as they were routinely identified as 'incidental findings' during the DS screening process, suggesting that they were more prevalent than once thought. Thus, the

⁴⁴⁵ Ibid, 93.

UK National Screening Committee (UK NSC) concluded that the number of reported incidences justified ES' and PS' inclusion into the programme.

While ES and PS, among the general population, are statistically less prevalent than DS, 1:800 will receive a screen positive result, and 1:3000-5000 parents will receive a positive diagnosis. This research identified that the risk of ES and PS, at the first instance, is being trivialised in the information packs and during counselling, routinely being labelled as 'unlikely' and 'rare'.⁴⁴⁶ While ES and PS are *statistically 'rare'*, the risk *still* exists. To overemphasise the unlikelihood of having a baby with ES or PS risks overlooking the possibility of receiving a higher-risk result, or indeed a positive diagnosis, for these conditions.⁴⁴⁷ The question remains whether the apparent trivialisation of risk for ES and PS is influenced by existing case law.

In *A v East Kent Hospitals*, Dingemans J surmised that the failure of HCPs to discuss the baby's risk of having a chromosomal condition (1:1000) was *justifiable*, as the risk was "theoretical", "background" and "negligible".⁴⁴⁸ Dingemans J concluded that "... *the decision in Montgomery affirms the importance of patient autonomy, and the proper practice set out in the GMC Guidance and the proper approach set out in Pearce and Wyatt. It is not authority for the proposition that medical practitioners need to warn about risks which are theoretical and not material.*"⁴⁴⁹

Indeed, in *Tasmin v Barts*, a case concerning the failure to perform a blood sample for the purpose of identifying the risk of transient ischaemic attack, Jay J highlighted that, "*a risk of 1:1000 is an immaterial risk for the purposes of Montgomery*".⁴⁵⁰ While *Montgomery* explained that materiality should not be reduced to percentages, a risk of 1:1000 fell below the undefined "borderline".⁴⁵¹

Rarity notwithstanding, parents must understand the risks of having a baby with ES and PS, and the implications of this outcome.⁴⁵² As was the case in *Pearce*, failure to appropriately

⁴⁴⁶ HCP Qualitative Findings, Section 6.7.2 and Parent Qualitative Findings, Section 5.6.1.

⁴⁴⁷ HCP Qualitative Findings, Section 6.7.2. and Parent Qualitative Findings, Section 5.6.1.

⁴⁴⁸ *A v East Kent Hospital University NHS Foundation Trust* (2015) EWHC 1038, 69; see also Sarah Devaney and others, 'The far-reaching implications of Montgomery for risk disclosure in practice', (2019) 24 *Journal of Patient Safety and Risk Management*, 25..

⁴⁴⁹ *A v East Kent Hospital University NHS Foundation Trust* (n448), 90.

⁴⁵⁰ *Tasmin v Barts Health NHS Trust* (2015) EWHC 3135 (QB), 115.

⁴⁵¹ *Ibid*, 115.

⁴⁵² Phoebe Winn, Krishna Acharya, Erika Peterson, and Steven Leuthner, "Prenatal counselling and parental decision-making following a fetal diagnosis of trisomy 13 or 18", (2018) *Journal of Perinatology*, 1.

discuss risk means that parents may be “blindsided”, if the risk materialises.⁴⁵³ *Montgomery’s* two-limbed assessment of materiality, suggests that parents should appreciate the physical and psychological risks of ES and PS, both to the pregnant person (trisomy-associated miscarriage or complex pregnancy) and baby (stillbirth and a shortened prognosis), before providing consent.

7.6.2 Differentiating Between Population and Individual Risk

Upon receiving information on population risk for the trisomies at the booking appointment, parents must then decide whether the risks warrant the undertaking of screening. If parents decide to screen, they receive an individualised, bespoke risk of DS and/or ES and PS. However, the transition between population and individual risk challenges the understanding of parents.⁴⁵⁴

The existing narrative is that older parents, in the general population, are more ‘at risk’ of having a baby with a trisomy.⁴⁵⁵ While this is objectively/statistically true, this narrative risks misleading younger parents to believe that they need not concern themselves with being higher-risk.⁴⁵⁶ Indeed, Engels *et al.* found that uptake of screening remains low among younger women, due to an overemphasis by HCPs that younger women are ‘low-risk’ of having a baby with a trisomy.⁴⁵⁷ While older parents are biologically more likely to have a baby with a trisomy, HCPs must communicate that this categorisation accounts for the generalised population risk, which is not reflective of parents’ individual risk.⁴⁵⁸

7.6.3 Parents’ Search for Certainty Amongst the Uncertainty

Jackson opines that, “information about risk is notoriously difficult to understand”.⁴⁵⁹ Making sense of the difference between population and individual risk arguably demands high levels of statistical and medical literacy. Conceptualising individual risk will require counselling on relative (group risk) and absolute risk (the actual risk itself, usually conveyed in percentages),

⁴⁵³ Jill Wieber Lens, ‘Medical Paternalism, Stillbirth, & Blindsided Mothers’, (2021) 106 *Iowa Law Review*, 674.

⁴⁵⁴ Parent Quantitative Findings, Section 4.3 and Parent Qualitative Findings, Section 5.6.2.

⁴⁵⁵ HCP Qualitative Findings, Section 6.7.3.

⁴⁵⁶ HCP Qualitative Findings, Section 6.7.3 and Parent Qualitative Finding, Section 5.6.2.

⁴⁵⁷ Melanie A J Engels and others, ‘Evaluation of the introduction of the national Down syndrome program in the Netherlands: age-related uptake of prenatal screening and invasive diagnostic testing’, (2014) 174 *Eur J Obstet Gynecol Reprod Biol*, 59.

⁴⁵⁸ *Ibid*, 63.

⁴⁵⁹ Emily Jackson, (n96), 12.

including specificity, sensitivity, positive and negative predicted values; factors which many parents' and HCPs' underlined as formidably difficult to understand.⁴⁶⁰ HCPs and parents are not trained statisticians; therefore, comprehending the information is challenging, to both the ability of HCPs to explain these concepts, and for parents to digest and understand the information, within the confines of the clinical appointment.

Parents' misinterpretation of a *screening*, believing it to be definitive as to whether their baby has/does not have a trisomy, was the most concerning misunderstanding of 'risk' in this study.⁴⁶¹ While many factors may be at play to contribute to this misunderstanding – predominantly parents' failure to read the information, and the pace of counselling at clinical appointments – it is certainly indicative of consent being less than informed.

This issue may also originate from parents' search for certainty among the uncertainty; the screening experience is often unfamiliar, confusing, and anxiety-inducing for parents.⁴⁶² Indeed, this may suggest that parents consent to screening, under the pretence that it will provide a diagnosis: an outcome which screening alone is unable to fulfil.⁴⁶³

Thus, it is essential that HCPs support parents' understanding of individual risk, including the possibility of false-positive and negative results. In many ways, as the participants conclude, the risk of a trisomy is often a 'lottery'.⁴⁶⁴ Despite developments in screening technology, the possibility of false-positives and negatives still exist, for *all* methods of *screening*. Coverage of the reasons why false-positive and negative results occur in screening – including available options in the event of this phenomenon, to inform parents decision-making – is required to clarify this distinction.

The US case of *Meleney-Distassio v Weinstein* highlighted the legal significance of counselling parents on screening's detection rates.⁴⁶⁵ In this case, the court found in favour of the Claimant, following the failure of the HCPs to warn the parents of the possibility of false-positive results. The HCPs, in this case, suspected that a baby had ES, due to an observation during a 3D ultrasound of club foot. Proper practice would have been to conduct appropriate

⁴⁶⁰ HCP Qualitative Findings, Section 6.7.4 and Parent Qualitative Findings, Section 5.6.3.

⁴⁶¹ HCP Qualitative Findings, Section 6.7.4 and Parent Qualitative Findings, Section 5.6.3.

⁴⁶² HCP Qualitative Findings, Section 6.7.5 and Parent Qualitative Findings, Section 5.6.3.

⁴⁶³ HCP Qualitative Findings, Section 6.7.5 and Parent Qualitative Findings, Section 5.6.3.

⁴⁶⁴ Parent Qualitative Findings, Section 5.6.3.

⁴⁶⁵ *Meleney-Distassio v Weinstein*, No. FSTCV136018746S, 2014 Conn. Super.

follow-up testing to obtain a diagnosis, before opting to terminate the pregnancy, due to the risk of false-positive results; this was not followed. The Claimants terminated the pregnancy, and later discovered that the baby did not have ES. This case reinforced the importance of explaining to parents that a high-risk screening result is not definitive of the baby's diagnosis, and that discordant results are still a possibility.

Clinical guidelines do not outline the HCP's duty in this regard, nor whether this duty falls to the midwife or sonographer. As it stands, undertaking the task of explaining and supporting parents' understanding of risk, including discordant results, within the confines of the clinical appointment, is arguably unreasonable.⁴⁶⁶ Deducing what is 'reasonable' may first require the publication of bespoke guidance on how to effectively support parents' understanding of this phenomena, under the current time-pressures of clinical practice; *Mordel's* assessment of a 'reasonable' system ultimately left this question unanswered.

7.6.4 Expression of 'Risk', and the Rigidity of High-and Low-Risk Categorisations

Post-screening, parents are placed in either high or low-risk groups, set by national and local policy; however, parents experience difficulty interpreting the concept of risk, in the context of their individual screening results, as their perception of high-and low-risk may differ to that set by the medical community, and national/local policy.⁴⁶⁷ Perception of risk is highly personal, and parents often disagree with the 'set boundaries' and rigidity of the high-and low-risk categorisations, as their objectivity overlooks parents' subjective and nuanced considerations going to decision-making.⁴⁶⁸

This finding speaks to the significance of the subjective limb in *Montgomery's* assessment of material risks. Indeed, while the profession has defined high-and low-risk groups as a matter of procedural efficiency for all three trisomies, this currently conflicts with parents' perception of high-and low-risk.⁴⁶⁹ This is further compounded by the varying definitions of 'high-risk', which is subject to local policy, ranging significantly between 1:150 and 1:800, or less.⁴⁷⁰

⁴⁶⁶ HCP Qualitative Findings, Section 6.7.4 and Parent Qualitative Findings, Section 5.6.4.

⁴⁶⁷ Parent Qualitative Findings, Section 5.6.4.

⁴⁶⁸ Parent Qualitative Findings, Section 5.6.4.

⁴⁶⁹ Parent Qualitative Findings, Section 5.6.4.

⁴⁷⁰ Liverpool Women's high-risk bracket is 1:800 or less, while University Hospital for Wales was 1:150 or less.

Due to the nature of ES and PS, and the consequences this has on decision-making for both mother and child, the objectivity of the high-and low-risk group categorisations may not be conducive with *Montgomery's* intended subjectivity, to provide a reasonable assessment of risk. It may be that the rigidity of the risk categories, which is currently formulated as a genetic model, deprives parents of the opportunity to undertake alternative methods of treatment (i.e invasive testing), in situations where *they* feel it is warranted. Furthermore, the gravity and magnitude of risk for ES and PS is greater than that of DS, which is not currently reflected in the traditional, objective grouping system.

It is no surprise, therefore, that a call for the abolition of the risk/chance categories emerged in this research, from both parents and HCPs.⁴⁷¹ The objectivity of the grouping system currently hinders parents' understanding of screening results. It may be that the objective statistical risk grouping system should also be accompanied by a subjective assessment of risk, in accordance with the particular patient and the individual trisomy. Thus, this raises interesting questions surrounding whether it be reasonable or logical to deprive parents of the opportunity to undertake invasive testing, based on the traditional 'low-risk' categorisation.⁴⁷²

In *A v East Kent Hospitals*, HCPs attempted to make use of 'decision trees' as a method of 'efficiently' explaining risk, under existing time constraints. Using statistical probability as a foundation to this tool, the HCP sought to evaluate which risks were 'material', and created predictions in relation to the decisions parents may make in each given circumstance.⁴⁷³ However, Arvind and McMahon explain that this tool is a "far cry" from the principles of *Montgomery*, which placed significance on shared decision-making, and is a product of "how doctors function in a heavily regulated organisational environment".⁴⁷⁴ This also raises the familiar question of establishing a reasonable system, under which HCPs can effectively discharge their duty in this regard.

⁴⁷¹ HCP Qualitative Findings, Section 6.7.4 and Parent Qualitative Findings, Section 5.6.4.

⁴⁷² Parent Qualitative Findings, Section 5.6.4.

⁴⁷³ T. T. Arvind and Aisling M. McMahon, (n375), 470.

⁴⁷⁴ *Ibid.*

7.6.5 Invasive Testing and the Challenge to Parents' Autonomy

When parents receive a high-risk or screen-positive result, they will be provided with the option to undertake invasive testing. It has been well-documented that supporting parents' autonomy, in the context of invasive testing, is fraught with legal and ethical difficulties.⁴⁷⁵ Parents' search for certainty from screening is central, not least because screening is unable to deliver a definitive result; therefore, HCPs must communicate that it is only invasive methods of testing, which carry a risk of procedural miscarriage, that can provide this certainty. However, the latter induces a stress, anxiety and emotional upheaval among parents.⁴⁷⁶

This research underlined the challenges posed to parent decision-making during the process of balancing the risk of procedural miscarriage, with obtaining a definitive diagnosis.⁴⁷⁷ As was the case in *A v East Kent Hospitals*, the court had to consider these competing factors, weighing the "negligible", "theoretical" or "background risk" of the baby having a chromosomal condition (1:1000), against the "real" risk of miscarriage (1-3:100).⁴⁷⁸ The Claimant submitted that, '*... the fear of having a baby with chromosomal defect would have been fore front in my mind and despite the risk of provoking premature labour ... would have opted for amniocentesis.*'⁴⁷⁹ Dingemans J dismissed the claim, expressing '*I do not accept if Mrs A had been given about the risk of B having a chromosomal abnormality that Mrs A would have had an amniocentesis, because the risks of having a disabled baby would have been greater from amniocentesis than from continuing with the pregnancy.*'⁴⁸⁰ however, this primarily objective assessment of materiality had arguably omitted the necessary subjectivity.

This balancing process and subsequent assessment of materiality, requires the necessary subjectivity, as *significant* psychological implications are experienced by parents upon the return of a higher-risk result, and the options it raises, due to the condition's very nature.⁴⁸¹

⁴⁷⁵ Frank Chervenak and others, 'Enhancing patient autonomy with risk assessment and invasive diagnosis: an ethical solution to a clinical challenge', (2008) 199 Am J Obstet Gynecol, 1.

⁴⁷⁶ Parent Qualitative Findings, Section 5.6.5; see also, Marie-Anne Durand and others, 'Information and decision support needs of parents considering amniocentesis: interviews with pregnant women and health professionals', (2010) 13 Health Expectations, 126.

⁴⁷⁷ Parent Qualitative Findings, Section 5.6.5.

⁴⁷⁸ *A v East Kent Hospital University NHS Foundation Trust* (2015) EWHC 1038, 69 and 96.

⁴⁷⁹ *Ibid*, 94.

⁴⁸⁰ *Ibid*, 102.

⁴⁸¹ Parent Qualitative Findings, Section 5.6.5.

Devaney *et al.* explain that while, objectively, one would agree with the assessment of Dingemans J in A, “this line of reasoning overlooked how the nature of this risk (of an unborn baby suffering from chromosomal abnormality) would have operated on the mind of the particular patient, both in terms of severity of consequence should it have materialised and how she would have responded had she been informed of it”.⁴⁸²

Indeed, without sufficient counselling, this period of uncertainty – or the parent’s ‘uncertain risk status’ – delays emotional attachment between mother and baby, which is proven to increase the likelihood of psychological harm.⁴⁸³ Thus, the significance of the subjective limb of materiality in *Montgomery* comes to the fore, safeguarding the patient’s ability to express “their experiences, interests and preferences to the process of shared decision-making.”⁴⁸⁴ This may also negate claims for solatium, which is a latent risk for the profession in this regard.⁴⁸⁵

7.6.6 Trivialising the Risk of Invasive Testing

The decision to undertake invasive testing, that carries a risk of miscarriage, is arguably one of the most challenging and ethically charged considerations, in any area of medicine. Ultimately, parents balance the risk of losing their baby, with the desire to know.⁴⁸⁶ However, ‘low’, ‘negligible’ and ‘trivial’ are adjectives commonly used to describe the procedural risk of miscarriage.⁴⁸⁷ Previous research has also found that parents are not provided with sufficient information on the ‘low’ risk of invasive testing, and the consequences of the procedure, to make an informed decision.⁴⁸⁸

Adjectives such as ‘low’ and ‘trivial’, to describe the risk of miscarriage, suggest that an effort is being made by HCPs to reassure or comfort parents. While this may be a technique employed to reduce parent anxiety, *Thefaut* underlined the risks this approach has on informed decision-making. In the instant case, the Defendant described the risk of the

⁴⁸² Sarah Devaney, (n448), 26.

⁴⁸³ Parent Qualitative Findings, Section 5.6.5; see also, Rowe H, Fisher J and Quinlivan, J, ‘Women who are well informed about prenatal genetic screening delay emotional attachment to their fetus’, (2009) 30 *Journal of psychosomatic obstetrics and gynaecology*, 34.

⁴⁸⁴ *Ibid.*

⁴⁸⁵ *Ibid.*

⁴⁸⁶ Parent Qualitative Findings, Section 5.6.6.

⁴⁸⁷ Parent Qualitative Findings, Section 5.6.6.

⁴⁸⁸ Green JM and others, ‘Psychosocial aspects of genetic screening of pregnant women and newborns: a systematic review’, (2004) 8 *Health Technology Assessment*, 1

procedure being “very small”, and provided “comforting and optimistic advice”.⁴⁸⁹ While this was evidently a technique used by the HCP to ease anxiety, the court found that the HCP placed undue emphasis on the risk of the procedure being “very small”. The judgment highlighted the damage overestimating success rates, and underestimating the risks, may have on the patient’s ability to exercise an informed choice.⁴⁹⁰

Use of such terminology risks being indictive or suggestive of a preferred course of treatment.⁴⁹¹ Overemphasis of a lower-risk could impel parents to undertake the procedure, rather than considering discontinuing treatment. The Canadian case of *Seney* found that if HCPs prefer one method of treatment, this does not absolve them of their duty to advise the patient of alternatives, which in the circumstance of invasive testing, would be to not receive further intervention.⁴⁹²

The functionality of the hybrid concept of materiality in *Montgomery* – being both an objective and subjective test – is called into question, considering that ‘risk’ is an actuarial concept in medicine, accounting for statistics and probabilities in its quantification.⁴⁹³ However, as highlighted by Arvind, cultural and social factors also play a role affecting patients’ attitude toward risk, arguing that “actuarial risk plays a minimum role” in this regard.⁴⁹⁴ Parents perception of risk, in this context, is highly personal.⁴⁹⁵ Thus, parents perception of ‘low’ may well differ to that of the HCP, as seen in *Ollosson*.⁴⁹⁶ Indeed, *Montgomery* reiterated that this assessment is fact-sensitive and attuned to the characteristics of the patient.⁴⁹⁷

Maintaining consistency is also a significant factor when communicating the risk of procedural miscarriage.⁴⁹⁸ While conflicting opinions is common among studies on medical science and care, inconsistent use of statistics by HCPs may exacerbate the difficulties parents experience

⁴⁸⁹ *Thefaut v Johnson* (2017) EWHC 497 (QB).

⁴⁹⁰ *Ibid.*

⁴⁹¹ Parent Qualitative Findings, Section 5.6.6.

⁴⁹² *Seney v Crooks* (1998), 223 A.R 145 (Alta. C.A) para 157.

⁴⁹³ T. T. Arvind and Aisling M. McMahon, (n375), 467.

⁴⁹⁴ *Ibid.*

⁴⁹⁵ Parent Qualitative Findings, Section 5.6.6.

⁴⁹⁶ Parent Qualitative Findings, Section 5.6.6; see also, *Ollosson v Lee* (2019) EWHC 784 (QB).

⁴⁹⁷ *Montgomery* (n1), 89.

⁴⁹⁸ Parent Qualitative Findings, Section 5.6.7.

processing and balancing the information on the risk, associated with invasive testing.⁴⁹⁹ Parents' ability to process complex numerical information has been proven to decrease under stressful and high-pressured situations, a factor which must also be taken into account in the context of invasive testing.⁵⁰⁰ Thus, maintaining consistency and clarity, when imparting information on risk at consultations and upon review of the paper-based materials, is required to support parents' decision-making in this regard.

⁴⁹⁹ Parent Qualitative Findings, Section 5.6.7.

⁵⁰⁰ Parent Qualitative Findings, Section 5.6.7; see also, Marie-Anne Durand, Mareike Stiel, Jacky Boivin and Glyn Elwyn, "Information and decision support needs of parents considering amniocentesis: interviews with pregnant women and health professionals", *Health Expectations*, 2010, 13(2): 125-138, at pp.126-127.

7.7.0 Counselling Parents on ‘Alternatives’: Situating Professional Duty

Montgomery underpinned the significance of providing patients with alternative therapeutic options when exercising an informed choice. *Montgomery* mandates that HCPs must disclose information on alternative treatment options, including the risks and benefits of the alternatives, before securing the patient’s consent.⁵⁰¹ In the context of trisomy screening, NIPT is framed as an ‘alternative’ to invasive testing, following a screen-positive result: “non-invasive prenatal screening is widely used as the alternative choice for pregnant women at high-risk of fetal aneuploidy”.⁵⁰²

While *Mordel* did not specifically address HCPs’ duty to counsel parents on NIPT, as the case preceded its introduction, the decision would suggest that both midwives *and* sonographers are required to review and counsel parents on the methods of screening, before obtaining consent. The operative portion of Jay J’s decision, in *Mordel*, raises the question whether the duty to counsel parents on NIPT, as an ‘alternative’ method of screening, should also fall to the sonographer.⁵⁰³

Indeed, sonographers are called upon by parents to answer questions on NIPT, at the examination appointments, with increasing frequency.⁵⁰⁴ However, sonographers’ CPD and training does not currently reflect this consideration, leaving them unequipped to counsel parents in this regard.⁵⁰⁵

‘Cascade’ training was provided to midwives, in 2016, in anticipation of NIPT’s rollout; however, the training was ineffective, and it was not offered to sonographers.⁵⁰⁶ While cascade training is desirable in terms of financial and resource constraints, the training model meant that information was diluted, and did not sufficiently equip midwives to counsel parents on NIPT’s implementation to the pathway.⁵⁰⁷

⁵⁰¹ *Montgomery* (n1), 90.

⁵⁰² Hui Zhu and others, ‘Efficiency of non-invasive prenatal screening in pregnant women at advanced maternal age’, (2021) 21 *BMC Pregnancy and Childbirth*, 86, <https://doi.org/10.1186/s12884-021-03570-6>.

⁵⁰³ *Mordel* (n14), 112.

⁵⁰⁴ HCP Qualitative Findings, Section 6.8.1.

⁵⁰⁵ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.8.1.

⁵⁰⁶ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.8.2.

⁵⁰⁷ HCP Qualitative Findings, Section 6.8.2.

Munthe opines that the introduction of NIPT will be difficult for HCPs “if existing programmes are problematic to start with”.⁵⁰⁸ It may be that reframing current systems for securing parent consent is required to accommodate counselling on NIPT, between sonographers and midwives. Although many trusts provide e-learning modules on NIPT for *all* HCPs, these are not bespoke to each area of professional practice. While synthesising the roles of sonographers and midwives on this issue is arguably desirable, guidelines must be clear on their *individual* duty within the broader system of counselling parents on NIPT, before this can be effectively executed. In addition, these training modules are not compulsory, nor are HCPs notified of any updates to their training.⁵⁰⁹ Thus, it may be necessary to create compulsory, bespoke training programmes, for both midwives and sonographers, to ensure accurate, balanced and regularly updated information on NIPT is available to effectively navigate their duty to secure parent consent.

Another solution may come in the form of creating bespoke NIPT consent clinics, whereby trained midwives and sonographers are able to establish a reasonable system for securing parent consent. While this may be idealistic in light of growing financial and time constraints within the NHS, this may enhance the efficiency and effectiveness of the provision of parent support and consent in this regard.

⁵⁰⁸ Munthe C, ‘A new ethical landscape of prenatal testing: individualising choice to serve autonomy and promote public health: a radical proposal’, (2014) 29 *Bioethics*, 39.

⁵⁰⁹ HCP Qualitative Findings, Section 6.8.2.

7.7 Counselling Parents on NIPT: Key Considerations

7.7.1 Understanding Why NIPT is Considered an ‘Alternative’ to Invasive Testing

NIPT is a method of *screening*, and is therefore *unable* to provide parents with a definitive diagnosis as to whether their child truly has a trisomy. However, the purpose of NIPT is being misinterpreted by parents and professionals.⁵¹⁰ Upon review of the information packs, medical research, clinical guidelines and the Nuffield Council’s report on NIPT, all recommend that HCPs offer NIPT as an ‘alternative’ to invasive testing, following a screen-positive combined test. While this issue seems ostensibly innocuous, presentation of NIPT, in this way, risks misleading parents to believe that they do not need invasive testing to obtain a definitive diagnosis.⁵¹¹

Without clear counselling in this regard, not only will parents be under the false pretence that they do not need amniocentesis or CVS following a screen-positive NIPT result, this misinterpretation of NIPT’s purpose risks depriving parents from exercising an informed choice over their reproductive options, to take earlier steps in their pregnancy to obtain a diagnosis.⁵¹² In some rare – albeit increasingly reported – situations, this false pretence may lead to the termination of a baby that does not have a trisomy. This concern became a tragic reality for Mr Kiely and Mrs Price in a recent case from Ireland.⁵¹³ This ‘wrongful termination’ case provided a timely illustration and reminder of the catastrophic impact defective counselling for NIPT’s accuracy, and effective systems for securing consent, may have on reproductive autonomy.⁵¹⁴

The parents, in this 2021 case, received an NIPT screen-positive result for ES. A failure to counsel the parents on the possibility of discordant results – and undertaking the wrong contingency tests during laboratory analysis of the sample – led the parents to believe that the baby had ES, and subsequently decided to terminate the baby.⁵¹⁵ It later transpired that

⁵¹⁰ HCP Qualitative Findings, Section 6.8.3 and Parent Qualitative Findings, Section 5.7.1.

⁵¹¹ Parent Qualitative Findings, Sections 5.7.1, 5.7.2 and 5.7.3.

⁵¹² Parent Qualitative Findings, section 5.7.2 and 5.7.3.

⁵¹³ The researcher followed the case closely as Dr Bryan Beattie – clinical supervisor for the researcher – acted as an expert witness for the Claimants.

⁵¹⁴ <https://www.theguardian.com/world/2021/jun/23/irish-couple-to-receive-damages-over-advice-that-led-to-unnecessary-abortion> (accessed 05/07/2021).

⁵¹⁵ The main issues in this case were a lack of appropriate counselling on the possibility of false-positive and negative results, and a lack of appreciation that chorionic villus sampling PCR explores the trophoblast (the same as NIPT), whereas the culture results look at the mesenchyme which is more likely to reflect the fetal

the parents received a false-positive NIPT reading resulting from confined placental mosaicism, meaning the baby did not have ES. Both the health service (the national maternity hospital and Greater Glasgow Health Board) and the private clinic admitted liability, resulting to an out of court settlement: remedy in damages. This case demonstrates the importance of effective counselling on NIPT's detection rate, in particular, drawing attention to NIPT's association with discordant results. It also underlined systematic failures which lead to uninformed decision-making and consent for termination. Anecdotally, more cases of 'wrongful termination', following discordant NIPT results, are coming to the fore under two separate health Trusts in England.

Clinical guidelines state that HCPs must direct parents to the information on the purpose of NIPT, in the context of both primary and secondary screening, to support parents' decision-making. Thus, it is necessary for HCPs to explain that the purpose of NIPT is to *reduce* the number of invasive tests being performed post-screening, and not to *replace* invasive testing. NIPT should be understood as an *initial alternative option* to invasive testing, as parents are still mandated to undertake invasive testing to confirm a screen-positive NIPT result, due to its inability to provide a definitive diagnosis.

7.7.2 Dispelling the Myth that NIPT is '99% Accurate'

A misunderstanding of NIPT's purpose may extend from an oversight of the test's disadvantages.⁵¹⁶ NIPT's 'accuracy', and the benefits this offers to patients, is often documented as '99% accurate' in the information packs and during counselling, referring to outdated clinical research.⁵¹⁷ This description of NIPT's accuracy is wholly misleading, as this does not account for the positive and negative predicated values.⁵¹⁸ This oversight may also extend to a lack of up to date training, provided by the health service, to deliver information on NIPT accurately, appropriately balancing the advantages and disadvantages to inform parent decision-making.⁵¹⁹

genetics i.e reduce the likelihood of obtaining a discordant result. Dr Bryan Beattie was the expert witness for the Claimant in this case.

⁵¹⁶ Parent Qualitative Findings, Section 5.7.3.

⁵¹⁷ Parent Qualitative Findings, Section 5.7.3 and 5.7.4.

⁵¹⁸ HCP Qualitative Findings, Section 6.8.4.

⁵¹⁹ HCP Quantitative Findings, Section 4.4 and HCP Qualitative Findings, Section 6.8.2.

The primary disadvantage of NIPT is its inability to provide a definitive diagnosis.⁵²⁰ To reiterate the point as a matter of clarity, NIPT is *not* diagnostic and can only provide a risk-score. However, NIPT's detection rate is currently a source of confusion among parents, often being described as being '99% accurate' or 'more or less diagnostic'.⁵²¹ Indeed, many patient information packs and online sources report NIPT as being 99% accurate.⁵²² Thus, it is no surprise to discover that parents report overlooking the possibility of discordant – or false-positive and negative – results.⁵²³

The commonly referred to statistic of '99% accurate' reflects population risk, rather than individual risk, and fails to account for the differing detection rates between the trisomies. To describe NIPT's detection rate in this way would be wholly incorrect, as the effectiveness of NIPT at detecting the common trisomies is dependent upon sensitivity, specificity, positive and negative predicted values. Green J in *Thefaut* opined, "*as the citation from paragraph [89] (Montgomery) above makes clear this can include caution in the use of percentages ... there is the risk that they can convey false degrees of certainty where, in truth, none really exists.*"⁵²⁴

While parents reported that the advantages of NIPT often dominated pre-testing discussion at consultations, this must be treated with caution.⁵²⁵ It may be that during the often uncertain and stressful position parents find themselves in post-screening, parents seek to be reassured and focus on any positive information from the HCPs. This may also be a tactic by HCPs to manage the anxiety parents experience at this time. Nevertheless, *Thefaut* addressed that overstating the benefits associated with a proposed intervention will corrode the patient's ability to make an informed choice.⁵²⁶ This raises questions surrounding the effective management of parent anxiety, and also being aware that ascertaining positive information will often be the objective of parents at consultations.

⁵²⁰ Parent Qualitative Findings, Section 5.7.3.

⁵²¹ HCP Qualitative Findings, Section 6.8.4 and Parent Qualitative Findings, Section 5.7.4.

⁵²² Parent Qualitative Findings, Section 5.7.4.

⁵²³ HCP Qualitative Findings, Section 6.8.4 and Parent Qualitative Findings, Section 5.7.4.

⁵²⁴ *Thefaut v Johnson* (2017) EWHC 497 (QB), at 59.

⁵²⁵ Parent Qualitative Findings, Section 5.7.3.

⁵²⁶ *Thefaut v Johnson* (n524), 59.

7.7.3 Early Signs of NIPT's Routinisation and its Impact on Decision-Making

Overstating NIPT's advantages could also lead to its routinisation.⁵²⁷ The overriding narrative, which emerges from the NHS and the private market, is that NIPT is 'more accurate' than combined screening, and is 'safer' than invasive testing.⁵²⁸ Thus, it is no surprise that a growing body of medical research is coming to the fore underlining the risk this may have on its routinisation, and parents ability to make deliberate and informed decisions.⁵²⁹

Heuvel *et al.* found that HCPs view the consent process for NIPT differently to that of invasive testing, spending considerably *less* time counselling parents on NIPT's implications and consequences.⁵³⁰ The Nuffield Council reports that due to NIPT's procedural simplicity and safety, significant concerns arise surrounding the level of explanation and discussion HCPs have with parents on NIPT's implications.⁵³¹ Time constraints will inevitably mean that HCPs only have limited periods with parents to discuss this information, and their subsequent choices, warranting a review of practice guidelines in this regard.⁵³²

This concern is becoming a reality under some NHS Trusts. With NIPT being an automatic contingency test under Trusts in England from 2019, a generalised consent model currently exists to use blood samples for both the combined *and* NIPT screening, if required. Thus, parents are consenting to a test they may or may not have, only becoming aware that their bloods have been used for the purposes of NIPT once they obtain their NIPT result, either by telephone or in the post. Indeed, this deprives parents of the opportunity to receive specialised counselling on NIPT, and the time required to reflect on their decisions and choices.

Along with parents in this study, Perrot and Horn also emphasise that the routinisation of NIPT could have a detrimental impact on the DS community, as its safety and accessibility

⁵²⁷ HCP Qualitative Findings, Section 6.8.5 and Parent Qualitative Findings, Section 5.7.3.

⁵²⁸ HCP Qualitative Findings, Section 6.8.5 and Parent Qualitative Findings, Section 5.7.3.

⁵²⁹ Adeline Perrot and Ruth Horn (n295), 1.

⁵³⁰ van den Heuvel A, 'Will the introduction of non-invasive prenatal diagnostic testing erode informed choice? An experimental study of health care professionals,' (2016) 78 Patient Education and Counselling, 8.

⁵³¹ Nuffield Council Report, 114.

⁵³² *Ibid*, 46.

risks increasing the number of terminations.⁵³³ They also believe that its introduction will feed existing discriminatory attitudes among parents and the profession.⁵³⁴

However, this concern is founded upon a fundamental misinterpretation of NIPT's purpose.⁵³⁵ As NIPT is more accurate than combined screening, it will ultimately lead to a smaller number of invasive tests needing to be performed, reducing the number of procedural miscarriages. As Leonard states, this does not mean that the number of terminations will necessarily increase. This speaks to the need for HCPs to effectively communicate NIPT's purpose in these contexts, ensuring that this misunderstanding does not negatively impact on parents' decisions to undertake or refuse NIPT.

7.7.4 The Risk of Secondary Findings

Uncovering secondary findings on maternal health is a possibility from undertaking NIPT.⁵³⁶ However, this begs the question whether the HCP's duty extends to counselling parents on the possibility of secondary findings, from NIPT screening. While there is a growing body of medical research identifying the risk of secondary findings from NIPT, the question of HCP's approach to counselling parents remains ambiguous and undecided. Consultants, in this study, identified the lack of direction HCP's receive in this regard, underlining the risk secondary findings pose to obtaining valid consent.⁵³⁷

While discordant – or false-positive and negative – NIPT results may be entirely innocuous and benign, they may also be indicative of underlying maternal biological factors. NIPT's rapid expansion, to detect a broader range of conditions, is giving rise to increasing incidents of secondary findings, both in terms of detecting a condition that was unanticipated, but also the presence of maternal malignancies.⁵³⁸ The suggestion of a maternal malignancy, in the context of NIPT screening, often becomes known following an investigation into discordant results. Brownsword and Wale question whether parents possess the right-to-know and the right not-to-know about secondary findings from NIPT screening, and the moral dilemmas

⁵³³ Parent Qualitative Findings, Section 5.7.5; see also, Adeline Perrot and Ruth Horn, (n295), 4.

⁵³⁴ Parent Qualitative Findings, Section 5.7.5; see also, Adeline Perrot and Ruth Horn, (n295), 4.

⁵³⁵ Parent Qualitative Findings, Section 5.7.2.

⁵³⁶ HCP Qualitative Findings, Section 6.8.6.

⁵³⁷ HCP Qualitative Findings, Section 6.8.6.

⁵³⁸ Malgorzata Ilona Srebniak, Maarten F.C.M Knapen, Marieke Joosten, Karin E.M. Diderich, Sander Galjaard and Diane Van Opstal, "Patient-friendly integrated first trimester screening by NIPT and fetal anomaly scan", *Molecular Cytogenetics*, 2021, 14(1): 1-6.

this rises during counselling.⁵³⁹ Some NIPT manufacturers are able to mask incidental findings, while others do not possess this ability.⁵⁴⁰

Clinical guidelines do not refer to the issue of secondary findings in this context; however, this raises the question whether HCPs are required to counsel parents on the risk of secondary findings from NIPT screening before obtaining consent, and upon whom does this duty rest. Dow *et al.* found that many NIPT consent forms do not disclose the possibility of incidental findings, such as cancer or malignant tumours.⁵⁴¹ These academics conclude that, despite the limited presence of practice guidelines and education in this regard, “the investigation and management of women with abnormal NIPT results requires a timely, thorough, and sensitive approach ... considering the potential impact not only on the women but also on her future child, fertility, and family.”⁵⁴² In such circumstances, the question remains whether HCPs are equipped to counsel parents sufficiently in this regard.⁵⁴³

Detection of a *translocation* trisomy also contributes to the complexities surrounding HCPs’ duty for information disclosure. Unlike mosaic or full trisomy, translocation is hereditary; there is a risk, therefore, of the condition being passed to later generations.⁵⁴⁴ While discussion of the law surrounding carrier status falls outside the ambit of this thesis, *ABC v St George’s & Ors* challenged the scope of the HCP’s duty, in the context of information disclosure for third-party carrier status.⁵⁴⁵ This case raised some important questions for further exploration on the HCP’s duty on securing consent, in the wake of NIPT’s broadening scope.

7.7.5 Latent Impact of the Private Market and Commercialisation of NIPT

To conceptualise consent as a continued dialogue, and not as a ‘one-off’ event, logically mandates consideration of the private sector on HCPs’ duty to secure it for screening. An

⁵³⁹ Roger Brownsword and Jeff Wale, ‘Testing Times Ahead: Non-Invasive Prenatal Testing and the Kind of Community We Want to Be’, (2018) 81 *Modern Law Review*, MLR 646

⁵⁴⁰ *Ibid*,

⁵⁴¹ Eryn Dow and others, ‘Cancer Diagnoses Following Abnormal Noninvasive Prenatal Testing: A Case Series, Literature Review, and Proposed Management Model’, (2021) *JCO Precision Oncology*, 1001.

⁵⁴² *Ibid*, 1010.

⁵⁴³ Nobuhiro Suzumori, ‘Retrospective details of false-positive and false-negative results in non-invasive prenatal testing for fetal trisomies 21, 18 and 13’, (2021) 256 *European Journal of Obstetrics & Gynaecology and Reproductive Biology*, 75.

⁵⁴⁴ Graeme Laurie, Shawn Harmon, and Edward Dove, *Mason and McCall Smith’s Law and Medical Ethics* (11th Edition, 2019), 206.

⁵⁴⁵ *ABC v St George’s NHS Healthcare Trust* (2020) EWHC 445 (QB).

assessment of whether the private sector adds strain or complicates existing NHS systems, for obtaining consent, must also be included in this conversation.⁵⁴⁶ As the Nuffield Council forewarned, “although women and couples accessing NIPT in the private sector will have actively sought out NIPT services rather than being offered them as NHS care, the information that they need in order to make informed choices and avoid harm is the same”.⁵⁴⁷

Research into the *private* NIPT market is scant; this is surprising, considering the potentially wide-reaching and detrimental influence the commercialisation of NIPT, and its expanding market, could have on NHS HCPs’ duty to secure parent consent. Indeed, discussion and decision-making on NIPT, accessed through the private sector, often overflows into ‘routine’ NHS consultations for trisomy screening, with increasing frequency.⁵⁴⁸

Parents who undertake screening privately, typically do so alongside ‘routine’ NHS care.⁵⁴⁹ As such, parents who experience unexpected outcomes from private NIPT screening often rely heavily on the NHS for further advice or for specialist care.⁵⁵⁰ While some private clinics provide midwives and/or consultants to support parents’ decision-making for NIPT, these vary significantly in quality and accessibility.⁵⁵¹ Jackson stressed that parents are likely to “return to the NHS for explanations, advice and reassurance”, in such circumstances, adding additional strain to the NHS screening programmes.⁵⁵²

This raises three important questions: (i) to what extent does the NHS HCP’s duty to secure consent extend to matters of private care; (ii) depending on the latter, whether NHS HCPs are sufficiently equipped to counsel parents on the implications of NIPT screening, from private care; and (iii) whether a ‘reasonable’ NHS system for securing consent on NIPT should be reframed, in accordance with the influence of the private market. In answering these

⁵⁴⁶ HCP Qualitative Findings, Section 6.8.7.

⁵⁴⁷ Nuffield Council Report, 99.

⁵⁴⁸ HCP Qualitative Findings, Section 6.8.7 and Parent Qualitative Findings, Section 5.7.6.

⁵⁴⁹ HCP Qualitative Findings, Section 6.8.7 and Parent Qualitative Findings, Section 5.7.6.

⁵⁵⁰ HCP Qualitative Findings, Section 6.8.7 and Parent Qualitative Findings, Section 5.7.6; see also, Panlai Shi and others, ‘The potential of expanded noninvasive prenatal screening for detection of microdeletion and microduplication syndromes’, (2021) 41 *Prenatal Diagnosis*, 1332.

⁵⁵¹ Parent Qualitative Findings, Section 5.7.6; see also, Panlai Shi and others, ‘The potential of expanded noninvasive prenatal screening for detection of microdeletion and microduplication syndromes’, (2021) 41 *Prenatal Diagnosis*, 1332.

⁵⁵² Emily Jackson (n260), 17.

questions, Mrs Price and Mr Kiely’s tragic case must be taken into consideration, to assess what reasonable steps can be taken to eliminate incidents of this nature in the future.

Considering the growing panel of conditions private clinics are able to offer screening for – including rare microdeletions, which are not available on the NHS – the question remains whether a ‘reasonable’ system should warrant NHS HCPs to support parent decision-making in this regard, both in the context of interpreting the result and/or follow-up care. Indeed, the Nuffield Council forewarned that, “increased use of NIPT in the private sector, particularly for conditions that have high rates of false results (rarer genetic conditions), conversely may lead to an increase in demand for invasive diagnostic testing and counselling on the NHS”.⁵⁵³

With the above implications in mind, it may be logical to reassess existing NHS systems on securing consent for screening, taking into account the growing influence of private care to the question of reasonableness. Returning to the premise of specialised and interprofessional clinics as a ‘reasonable’ solution, for those who have questions on undertaking NIPT screening, specialised clinics may be a resource – comprised of midwives, sonographers and consultants – to better support decision-making.⁵⁵⁴ While this would be costly and logistically complex, an interprofessional approach is vital to address any lacuna in medical coverage, better enhancing and protecting parent choice. In addition, it may be that routine training is provided to synthesise NHS HCPs’ knowledge of the developments under the private sector, as well as NHS programmes, to support HCPs’ clinical practice.

Appropriate training *must* include the influence of online advertising to undertake private NIPT screening. Social media and the internet currently operate as a primary source of information for parents on matters of pregnancy care.⁵⁵⁵ Upon review of the online information and social media on pregnancy care, encountering webpages of pharmaceutical giants, which promote and oversell NIPT, is unavoidable. Jackson explains, “there may be some atypical pockets of private practice where there is a risk of overselling” medical treatment.⁵⁵⁶ Indeed, many tactics are used to entice parents: enhancing convenience with

⁵⁵³ Nuffield Council Report, 101.

⁵⁵⁴ Abi Merriel and others (n305), 32.

⁵⁵⁵ HCP Qualitative Findings, Section 6.8.7 and Parent Qualitative Findings, Section 5.7.6; see also, Heather Skirton, and others, ‘Non-invasive prenatal testing for aneuploidy: a systematic review of Internet advertising to potential users by commercial companies and private health providers’ (2015) 35 Prenatal Diagnosis, 1.

⁵⁵⁶ Emily Jackson, (n96), 7.

‘direct-to-consumer’ kits; offering ‘whole panel screening’; screening packages which include discounts; sex determination and ‘gender reveals’, to name a few.⁵⁵⁷

This poses the risk of alluring parents, who are able to purchase it, to undertake NIPT, “rather than ... individuals in whom those tests are clinically indicated”.⁵⁵⁸ Thomas observes that, “new conditions are added to NIPT kits based on technological feasibility and profit motive, leading to widespread prenatal screening for incompletely understood genetic disorders.”⁵⁵⁹ The outstanding question, however, is what influence does this play on the parent’s decision to undertake NIPT screening, and the challenges this could pose for NHS HCPs on parent decision-making for trisomy screening. This also points to the risk posed by consumerism and commercialisation, with parents effectively ‘shopping’ for the best screening deals, raising the question whether product trumps partnership.⁵⁶⁰

7.7.6 Anticipating the Expansion of NIPT’s Purpose and Its Impact on HCPs’ Duty to Secure Consent

Due to the higher detection rate, safety and procedural ease of NIPT, HCPs and academics anticipate that it is only a matter of time before NIPT replaces the current combined test.⁵⁶¹ NIPT is also less demanding in terms of NHS resources, requiring only a midwife to take a blood sample. Gammon *et al.* opined that not only does this point to the replacement of combined screening, it is reasonable to assume with NIPT’s increased use and expansion into other non-trisomy related areas.⁵⁶²

While NIPT operates as a contingency test, in countries such as England, Wales, France and Germany, it has recently been introduced as a first-line screening test – replacing the traditional combined screening – in Belgium and the Netherlands.⁵⁶³ Indeed, the “TRIDENT-2” study is currently being conducted in the Netherlands, as part of an ongoing (2019-2023)

⁵⁵⁷ Parent Qualitative Findings, Section 5.7.6; see also, Daniel Navon and Gareth Thomas, ‘Screening before we know: radical uncertainties in expanded prenatal genetics’, (2021) 5 OBM Genetics, 12.

⁵⁵⁸ Emily Jackson, (n260), 17.

⁵⁵⁹ Daniel Navon and Gareth Thomas, (n557), 12

⁵⁶⁰ Parent Qualitative Findings, Section 5.7.6.

⁵⁶¹ Bettina Schone-Seifert and Chiara Junker, “Making use of non-invasive prenatal testing (NIPT): rethinking issues of routinisation and pressure”, *Journal of perinatal medicine*, 2021, 49(8): 959-964.

⁵⁶² HCP Qualitative Findings, Section 6.8.7; see also, Betsy L Gammon and others, ‘Implementing Group Prenatal Counseling for Expanded Noninvasive Screening Options’, (2018) 27 *J Genet Couns*, 894.

⁵⁶³ Kris Van Den Bogaert, and others, ‘Outcome of publicly funded nationwide first-tier noninvasive prenatal screening’, (2021) 23 *Genet Med*, 1137.

research group, to evaluate the use of NIPT as a “first-tier” screening test.⁵⁶⁴ It is also currently being trialled as a first-line screening test in Russia, with health care services anticipating a full implementation by 2022-2023.⁵⁶⁵

With pharmaceutical companies continuously seeking to develop and broaden NIPT’s scope, to include an ever-expanding panel of conditions, it may be that NIPT technology provides an opportunity for the NHS to establish additional screening programmes, to include other chromosomal aneuploidy.⁵⁶⁶ As Leonard opines, enabling an expansion of conditions affords parents to obtain more information about their baby, and allows early intervention, if required.⁵⁶⁷ For example, medical research into DiGeorge Syndrome has revealed that the condition is more prevalent among the general population than once thought, suggesting that DiGeorge should also have its own NHS screening programme, or to be added to current pathways.⁵⁶⁸

On its current trajectory, it is foreseeable that NIPT may not be confined to only the common trisomies, under the NHS. The NHS may follow the models adopted by other European countries, which also screen for sex chromosome conditions (i.e Turners Syndrome) and single gene disorders like microdeletions (i.e 22q11 deletion/DiGeorge Syndrome), as part their first-trimester screening programmes.⁵⁶⁹ As the prevalence of DiGeorge Syndrome is higher than once thought, it may be that the NHS will introduce screening programmes for this condition, as they did for both ES and PS, in the near future.⁵⁷⁰

These factors will create unprecedented challenges for HCPs to effectively counsel parents on NIPT. Bedei *et al.* argue that the expanding scope of NIPT will make it “nearly impossible” to obtain informed consent, as it will become too generalised.⁵⁷¹ They argue that consent ceases

⁵⁶⁴ Karuna ven der Meij, ‘TRIDENT-2: National Implementation of Genome-wide Non-Invasive Prenatal Testing as a First-Tier Screening Test in the Netherlands’, (2019) 105 AJHG, 1120.

⁵⁶⁵ Anton S. Olenov and others, ‘Adoption of a non-invasive prenatal test (NIPT) in prenatal screening in Moscow: first results’, (2021) 10 Russian Open Medical Journal, 110.

⁵⁶⁶ Yun Chen and others, ‘The application of expanded noninvasive prenatal screening for genome-wide chromosomal abnormalities and genetic counselling’ (2021) 34 J Matern Fetal Neonatal Med, 2710.

⁵⁶⁷ Samantha J Leonard, “Reproductive genetic screening for information: evolving paradigms?” (2021) 49 Journal of Perinatal Medicine, 998.

⁵⁶⁸ Yuan Zhao and Dong-Zhi Li, ‘Noninvasive prenatal testing for DiGeorge syndrome: is it ready for clinical practice?’, (2021) 23 Fetal Diagnosis and Therapy.

⁵⁶⁹ Daniel Navon and Gareth Thomas, (n557), 12.

⁵⁷⁰ Welsh Obstetrics Conference (20/05/2021).

⁵⁷¹ Ivonne Bedei and others, ‘Chances and Challenges of New Genetic Screening Technologies (NIPT) in Prenatal Medicine from a Clinical Perspective: A Narrative Review’, (2021) 12 Genes, 501.

to be bespoke, subject to the growing options of conditions available for screening.⁵⁷² Supporting parents' ability to navigate the rapidly evolving genetic testing landscape – not only for aneuploidy, but also carrier screening – must be urgently evaluated, in line with NIPT's impending expansion.⁵⁷³ This will pose significant challenges for HCPs, at pre-screening consultations, requiring an urgent review of the current resources to effectively inform parent decisions-making. Patient input, in the form of patient evaluation groups or workshops, would be invaluable for constructing an appropriate decision-making framework.

Self-determination and patient autonomy, the principles underpinning *Montgomery's* decision, will be challenged like never before in the face of NIPT's expansion. Many medical studies across the world are beginning to unveil a common theme, following the expansion of NIPT: 'responsible' motherhood, and its impact on participating in prenatal screening.⁵⁷⁴ Furthermore, it is becoming increasingly difficult for parents to decline NIPT screening, due to its safety, ease and expansion into detecting other conditions, pointing to its potential routinisation. Thus, a pressure, both in the healthcare setting and among social institutions, is beginning to ensue, as moral barriers become less prominent, in terms of circumventing invasive testing and the increased probabilistic nature of NIPT, compared to combined screening.⁵⁷⁵

⁵⁷² Ibid.

⁵⁷³ Ruth Farrell and others, 'Decision-Making for Prenatal Genetic Screening: How Will Pregnant Women Navigate a Growing Number of Aneuploidy and Carrier Screening Options', (2021) Research Square, 20.

⁵⁷⁴ Elisa Garcia and others, 'Non-Invasive Prenatal Testing (NIPT) and Pregnant Women's Views on Good Motherhood: A Qualitative Study', (2021) European Journal of Human Genetics, doi: 10.1038/s41431-021-00945-3.

⁵⁷⁵ Bettina Schone-Seifert and Chiara Junker (n561), 959.

Conclusion and Recommendations

This study has identified that parent and professional interests, in the context of securing consent for trisomy screening, currently conflict. It seems that the conflict of interests stems from a disconnect between the expectations of parents and the practical challenges professionals face to discharge their duty to secure consent for trisomy screening.

This study has underlined the paramount importance of appreciating consent as an ongoing process, with both parents and professionals being able to engage into a continued dialogue for the purpose of protecting reproductive autonomy and self-determination; this requires the upholding of *Montgomery's* commitment to patients' rights when disclosing information on trisomy screening and establishing *Bolitho* 'reasonable' systems for professionals to effectively secure consent.

Enhancing interprofessional practices, between the midwife and sonographer, is certainly a key recommendation to improve current systems for securing parent consent. With *Mordel* underlining the fragility of established systems, concluding that they were *Bolitho* unreasonable, this calls for the need to form clear systems of communication between the professions to effectively secure consent; whether this be in the form of requiring sonographers to adopt a chameleonic role, extending midwives' role to include involvement with consent at later stages in the screening process, or the formation of interprofessional consent clinics, these could mark the beginning of restoring professional standards for securing parent consent.

COVID-19 served as an example that established systems can be radically modified and overhauled in light of urgent demand; HCPs demonstrated that they have the ability to react, adapt and engage effectively with interprofessional practices. While COVID is rarely referred to positively in the context of healthcare, it did highlight the capability of maternity units to re-evaluate and implement reformed systems to uphold professional duty for securing consent for trisomy screening.

The inclusion of ES and PS to trisomy screening programmes has raised additional concerns going to parent decision-making and choice. Under a 'trisomy' screening model, consent and decision-making has become generalised, rather than bespoke to the consequences of receiving that often unexpected result. A fragmentation of the pathway would enable parents

and HCPs to better understand the conditions and their consequences and may assist programme co-ordinators in their quest for a 'reasonable' and streamlined system for securing parent consent.

The influence of consumerism on healthcare must also be considered for the purpose of delineating parent and professional interests. The increasing demand of parents to have access to treatment and finite resources, conflicts with the profession's commitment to beneficence, under a time and resource limited clinical environment.

The introduction of NIPT to NHS screening programmes will only exacerbate the influence and impact of consumerism. With the private market luring parents to undertake NIPT screening, through home-testing kits and discounts for whole panel screening, the provision of support within NHS units will come under increasing stress, posed the apart commercialisation of the product. Bespoke NHS NIPT consent clinics may be required to sufficiently evaluate both parent and professional interests before delivering and securing consent in this regard.

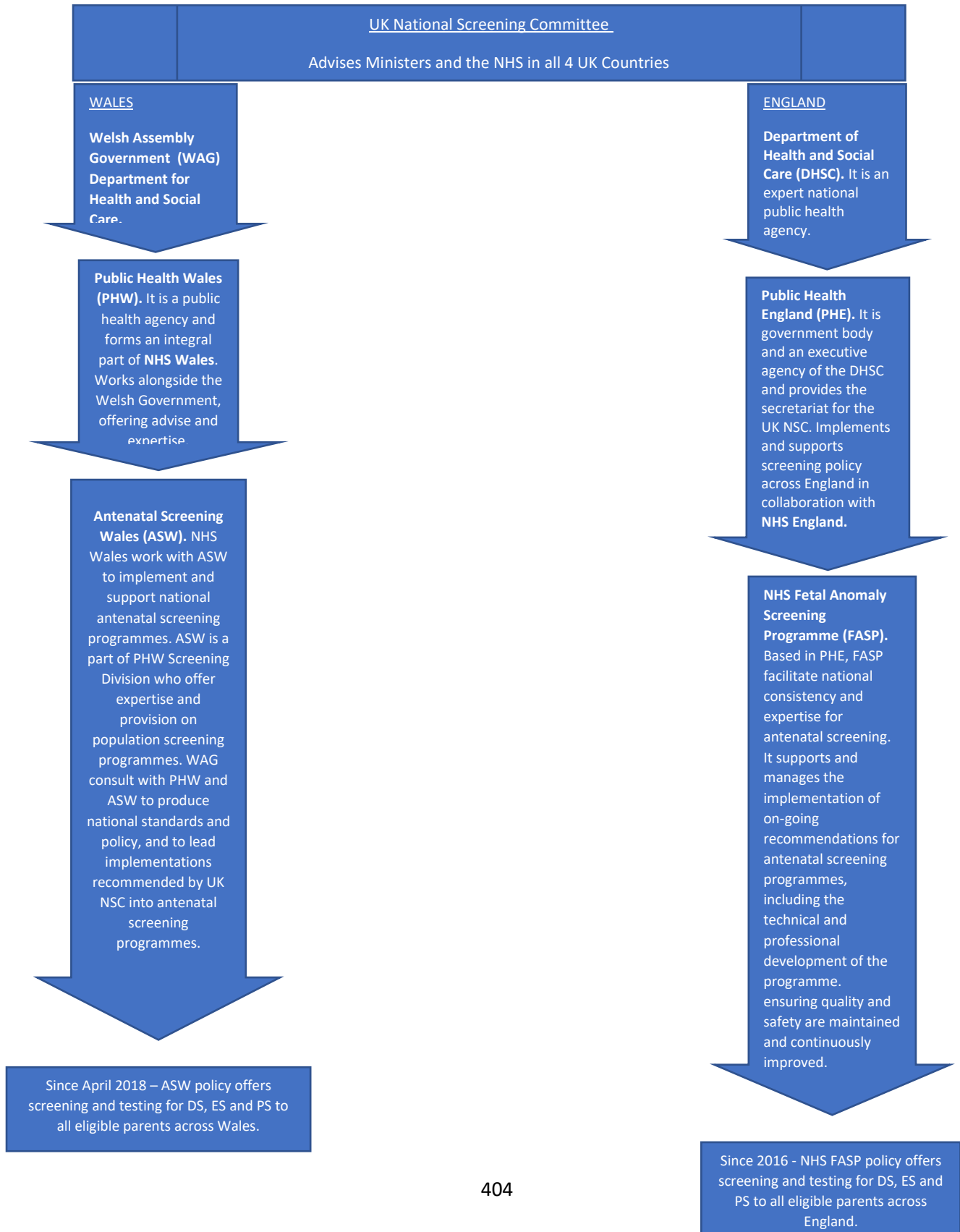
Providing HCPs, who perform a role going to the provision of support for parent decision-making and consent for trisomy screening, with the requisite up-to-date training must be a key consideration for Public Health England and Wales. With the study underlining HCPs' lack of confidence supporting parent choice and decision-making on the conditions and new methods of screening, an urgent need for review is required in this regard.

The purpose of this study was not to provide a definitive answer on how to restore the interests of both parents and professionals in the context of securing consent for trisomy screening; its purpose was to identify existing concerns for other researchers to explore in the future. While this study serves as a foundation for highlighting key considerations going to the restoration of interests, further research must be conducted to provide a comprehensive review of legal and ethical concerns and keep pace with the rapid development of the pathway.

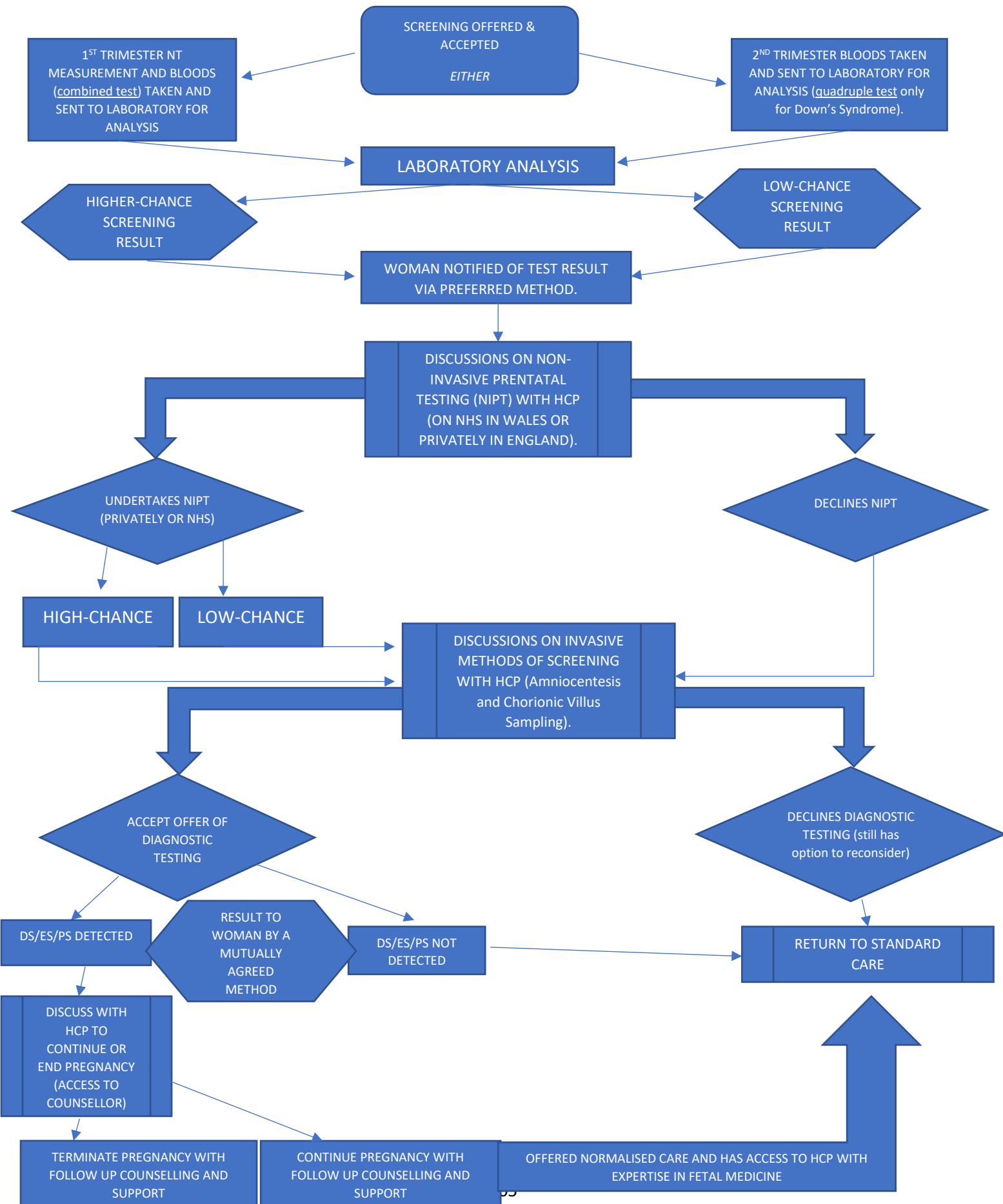
While it would be naïve and idealistic to conclude that parent and professional interests could ever reach a point of symbiosis, the key objective must be to narrow the margin between the party's interests, to enable effective delivery and obtention of consent for trisomy screening.

APPENDICES

Appendix 1: Chart of Structures and Relationships Between Bodies for Trisomy Screening in England and Wales:



Appendix 2: Diagram of the Current Trisomy Screening and Testing Pathway:



Appendix 3: Clinical Guidelines on Consent and Trisomy Screening/Testing

Antenatal Screening Wales Policy, Standards and Protocols 2019 – 7.0 and 8.0 Antenatal Screening for DS, ES and PS.

Association of Early Pregnancy Units.

British Medical Association – Committee on Medical Ethics: The Impact of the Human Rights Act 1998 on Medical Decision-Making, 2000.

British Medical Association – Consent and Refusal by Adults with Decision-Making Capacity – A Toolkit for Doctors, 2019.

British Medical Ultrasound Society (BMUS): Statement on Patient Information and Informed Consent.

British Society of Gynaecological Imaging.

Care Quality Commission. Regulation 11: Need for Consent. Health and Social Care Act 2008 (Regulated Activities) Regulations 2014: Regulation 11.

Department of Health: Maternity Matters: Choice, Access and Continuity of Care in a Safe Service (2007).

Department of Health: Reference Guide to Consent for Examination or Treatment, Second Edition. (2009).

Department of Health: The NHS Constitution (2015).

Fetal Anomaly Screening Programme Handbook for Ultrasound Practitioners (April 2015).

General Medical Council – Consent: Patients and Doctors Making Decisions Together (2008).

Health and Care Professions Council (HCPC) Standards. Regulating Health and Care Professionals.¹⁷¹⁰

NHS (England) Fetal Anomaly Screening Programme. Antenatal Screening – Working Standards for DS, 2007 (replaced by FASP Standards 2015-16 although original version still heavily relied upon and persuasive in recent case law on consent standards).

NHS (England) Screening Programmes: Fetal Anomaly Screening Programme Standards 2015-16 (policy to unify standards across England to ensure informed choice from women).

NHS England. Public Health England. NHS Fetal Anomaly Screening Programme Handbook, Valid from August 2018.

NICE. Society of Radiographers (2018) – Obtaining Consent: A Clinical Guideline for the Diagnostic Imaging and Radiotherapy Workforce.

Nuffield Council on Bioethics – Non-invasive prenatal testing: ethical issues. 2017.

Nursing & Midwifery Council (2018) The Code: Professional Standards of Practice and Behaviour for Nurses, Midwives and Nursing Associates.

¹⁷¹⁰ They oversee the NMC.

Public Health England. NHS Public Health Functions Agreement (2018). Service Specification no.16. NHS Fetal Anomaly Screening Programme – Screening for DS, ES and PS (Trisomy 21, 18 & 13).¹⁷¹¹

Public Health England. NHS Public Health Functions Agreement (2019-2020). NHS Fetal Anomaly Screening Programme – Screening for DS, ES and PS (Trisomy 21, 18 & 13).

Royal College of Obstetricians and Gynaecologists. Obtaining Valid Consent. Clinical Governance Advice No.6 2015. (standards in private practice).

Royal College of Obstetricians and Gynaecologists: Amniocentesis and Chorionic Villus Sampling (Green-top Guideline No.8).

Royal College of Obstetricians and Gynaecologists: Non-invasive Prenatal Testing (Green-top Guideline No.74).

Royal College of Radiologists. Standards for Patient Consent Particular to Radiology (2012).¹⁷¹²

Royal College of Surgeons. Consent: Supported Decision-Making – A Guide to Good Practice (2016).

The Health and Social Care Act 2008 (Regulated Activities) Regulations 2014. Section 2 Fundamental Standards.

The International Society of Ultrasound in Obstetrics and Gynaecology (ISUOG). ISUOG Practice Guidelines: Performance of First-Trimester Fetal Ultrasound Scan (2013).

The National Institute for Health and Care Excellence (2008 updated 2019) Antenatal Care for Uncomplicated Pregnancies. Clinical Guideline (CG62).

The Society & College of Radiographers. BMUS: Guidelines for Professional Ultrasound Practice (2019).

¹⁷¹¹ For the purposes of the study, focus is placed on this service specification, however, it should also be read in conjunction with Service Specification no.17 Fetal Anomaly Screening (preferred use to scan for only ES and PS (95% detection but only 50% for DS) – under the 11 auditable conditions).

¹⁷¹² While radiology and oncology are not specific to the thesis, this document could be useful to inform antenatal screening programmes. Several good points on patient information and consent.

Appendix 4: Information Sheet and Consent form for Parents

Introduction

My name is Emyr Wile and I am currently studying for a PhD in Law at Swansea University. I would like to invite you to take part in the research study. As part of the PhD research, I am seeking to recruit parents to obtain their experiences of the NHS antenatal trisomy screening and testing pathway across **Wales and England**. That is, screening and testing for Down's Syndrome (DS), Edwards' Syndrome and Patau's Syndrome (PS). Please note that this research will not explore your experience of the 20-week fetal anomaly scan. All participants would have received trisomy screening and testing within the **last 12-months**. Your participation in the study will be entirely voluntary and I will not have access to any of your medical records. I would like to brief you on the implications of the study and why it is being conducted before you decide to participate in the research. Section (A) will explain the purpose of the study and what will happen if you take part and section (B) will give you further detailed information about the conduct of the study. If you would like to participate in the research, please contact me by email, telephone or letter. If you would like to meet with me personally before conducting the study, I would be happy to do so.

Section (A) – The Implications of the Study and its Components

Why am I conducting the study?

Often, antenatal screening and testing is a difficult and overwhelming experience for parents. With the advancements in screening technology and access to testing, which is deemed to be more accurate, it is becoming increasingly common for prospective parents to embark on the antenatal trisomy screening and testing pathway to determine their chance of having a baby with, commonly, Down's Syndrome, Edwards' Syndrome and Patau's Syndrome. This has significant legal and ethical implications on professionals and patients. I aim to explore whether prospective parents are being given sufficient information and support at all stages of the antenatal screening and testing pathway to deliver truly informed consent. Consent is a dynamic process and I wish to discover whether this is sufficiently appreciated throughout the screening and testing pathway. With the recent implementation of NIPT by the NHS in Wales in 2018 (and its anticipated implementation by the NHS in England), it only amplifies the need to explore this issue. I will decipher whether current legal standards of care are being met in the context of trisomy screening and testing. The research will examine a range of potential themes to identify any demographic trends or patterns in the standard of care being delivered by the NHS. Examples of some of the issues that will be explored are:

- To evaluate whether healthcare professionals (HCP) are meeting current legal standards of care in the context of informed consent.
- Whether expectant parents *understand* the information received on antenatal screening and testing to sufficiently facilitate informed decision-making and consent.
- To evaluate whether balanced and accurate information is being delivered by HCP on antenatal screening and testing to facilitate informed consent.
- To discover whether women understand that it is their choice to have antenatal screening and testing, or do they feel that it is part of their antenatal care?
- Whether HCP make it clear to parents that they can disembark from the pathway or decline further investigative testing at any time.
- To ensure that HCPs remain non-directive and non-biased when communicating information in pre-and post-test consultation and counselling.
- To obtain the opinion and views of parents towards the trisomy screening and testing pathway.

Why have you been invited to take part in the research?

This study would significantly benefit from your participation as you have experienced the NHS antenatal screening and testing pathway within the last 12-months in Wales or England. I am hoping to obtain your experience and knowledge of the screening and testing pathway exclusively post-testing.

Do you have to take part in the research?

Your participation is entirely voluntary. I am happy to describe the study and go through this information sheet with you before you make a decision. I will ask you to sign a consent form before your participation. Your information will remain anonymous unless stated otherwise. I will irrevocably strip the information of any direct identifiers such as your name, address, race or gender. Your identity will be replaced by a study number. I could also meet with you at a neutral/convenient/comfortable location if you are concerned that your confidentiality or anonymity will be breached. You are free to withdraw from the research at **any** time, without giving a reason for doing so. Section (B) provides an explanation of the implications of withdrawal.

What is required from you if you decide to take part in the research?

If you kindly decide to take part in the research, you will be required to fill a questionnaire. This can be conducted online, via telephone or face-to-face. Following the completion of the questionnaire, there is an optional follow-up interview. Again, this can be conducted by telephone or face-to-face. Participants can attend on their own or with partners – it is entirely personal preference. If you are a single mother or father, this does not have any effect on your participation. Depending on whether you agree to partake in the interview, you will be required to answer a series of questions and comment on identified issues. You must be **over 18 years of age** to participate in the research for ethical purposes.

The location of the interview is highly flexible. I am able to conduct the interview with yourself at a convenient location, such as your home or a nearby facility. The interview should take approximately 40 minutes to an hour, depending on any additional comments you would like to contribute. The interview will be recorded through an available audio-device which will be later transcribed. Your consent would be required for me to do so. You are able to withdraw your consent to this at any stage of the research, without explanation. If you would like me to record the interview through another method, I would be happy to oblige. Unless stated otherwise, any direct identifiers to yourself will be removed to the best of my ability.

Are there any risks or disadvantages to the research?

As part of the research interviews, you will be required to comment on potentially sensitive issues. For example, your experience of receiving a high-chance result, being subjected to negative attitudes, discussion of termination or an overall unsatisfactory experience of the trisomy screening and testing pathway. This may cause discomfort, stress or anxiety. Your physical and psychological wellbeing is paramount. My supervisors and I, along with the advice from support groups, have carefully designed the research to ensure that your wellbeing is protected. My study has been supported by organisations such as the Down's Syndrome Association (DSA) and I have been working closely with a Down's syndrome campaign group called "Don't Screen Us Out". I have also met with several health professionals such as obstetricians and neonatal consultants, as well as leading professors, academics and specialists in the area of antenatal screening. They have supported my research and closely followed its development. They have also given me advice on how to best manage the study to ensure optimal outcomes. I have also received training in conducting and designing research during my master's degree in Social Science Research Methods in 2017-2018 at Swansea University.

Further support

If the study induces unpredicted emotions such as anxiety or distress, support is available to you. Antenatal Results and Choices (ARC) and Support Organisation for Trisomy 13 and 18 (SOFT) provide direct support and are happy to deliver further information. The Down's Syndrome Association (DSA) is an organisation that exists to support individuals with Down's Syndrome and their families and is available to provide information, advocacy and support at all times. You should also be reminded that you are free to withdraw from the research at any point, without having to provide an explanation. All of the information you have provided would then be immediately deleted and destroyed.

What are the possible benefits of taking part in the research?

I cannot promise that the study will help you directly. However, the information that I obtain from the study will help improve the experiences of other parents who embark on the trisomy screening and testing pathway. It will also further improve current standards of care for the benefit of both parents and professionals. This will provide a strategy to better inform practice guidelines and the decision-making/choices of parents throughout the screening pathway.

What happens after the research study has finished?

Following the completion of the study, you will receive a summary of the research findings. You will be able to double-check that you are anonymised and that the quotations you provided are accurate and representative. The information and data that I have obtained from yourself will be published in my PhD thesis which will be examined and approved by Swansea University. The data will be read by external assessors, also. Potentially, subject to your approval, the data could be used as part of an academic publication such as journal articles, book chapters and books.

What if there is a problem?

Any complaint in terms of the research such as the way you have been dealt with during the study or any possible harm you have suffered will be addressed. Section (B) contains detailed contact information if this circumstance should arise.

Will your taking part in the research be kept confidential?

Yes. Ethical and legal practice will be strictly followed, and all information provided by yourself will be handled in confidence. Further details on this matter is contained in Section (B).

Section (B) – Further Information

Contact details

If you have any questions in relation to the research or the information provided, please feel free to contact me directly (telephone - [REDACTED], or by email - [REDACTED]). If you would like to speak to my supervisors, again, please do not hesitate to contact them (Professor Karen Morrow; [REDACTED] and Trish Rees; [REDACTED]).

Complaints procedure

If you have a concern about *any* aspects of the research study, please contact myself on my telephone or email and I will do my best to answer your questions. If you are still unsatisfied or unhappy and wish to complain formally, please contact my supervisors via their emails provided above.

Can you withdraw from the study?

Yes. You are able to withdraw from the study at *any* time, without needing to provide an explanation. If you do wish to withdraw from the research, please let me know at your earliest convenience so that I can act immediately on your request. All information and data will be instantly deleted and destroyed. If you wish to re-engage with the research, you are able to do so at any time.

Will your information and data be kept confidential at all times?

Yes. Unless you specify otherwise, any direct identifiers will be removed such as your name, race and gender and will be replaced with a study number if you deem that to be appropriate. The data will be collected via audio-recordings and field notes. You have the right to check the accuracy of the data collected and to correct any errors. All the data you provide such as audio-recordings, field notes, transcribed interviews and your personal data will be stored in a locked filing cabinet on a secure Swansea University premises or on a computerised file secured by a password which is only accessible to myself and agreed members of staff. I understand that information that contains personal or identifiable data will fall underneath the ambit of the Data Protection Act. I will ensure that I follow the University's protocols for Data Protection. Your information and data will be held on file (computerised and non-computerised) at the University in a location which is only accessible to myself and my supervisors (Karen Morrow and Trish Rees). Firewalls, anti-virus software and other measures will be undertaken to ensure data protection. I will not store any data on my personal devices as they could get lost or stolen. After the research has completed, all data will be deleted and destroyed.

Who has reviewed the study?

The research study has been approved by the ethics research committee at Swansea University.

Financial/Organisational elements of the research?

My PhD research is being sponsored by Swansea University and is funded by the Economic and Social Research Council (ESRC). If you have any questions in relation to the funding body, please do not hesitate to ask. I am the Swansea University student representative for the ESRC.

Does this have any effect on the data?

As I am funded by the ESRC, a requirement for researchers is that the data will be given to the UK Data Archive (UKDA). As such, this will include the transcribed interview from yourself. However, any potential identifiers will be stripped from the data so that it is entirely unidentifiable and anonymised. In accordance with Swansea University's data retention period, the materials (notes of the interviews and the audio-recordings) will be kept for a minimum of five years after the research has finished. After this period, the data will be destroyed in accordance with Swansea University's adherence to legislation, such as the Data Protection Act and Freedom of Information Act.

Further details

If there is any further information that you would like to obtain in relation to the research, please do not hesitate to get in contact with myself or my supervisors. If you would like to meet with me in person to discuss any of the information provided, I will be more than happy to do so. Thank you for taking the time to read this information sheet.

Consent Form for Parents

Please provide your initials below each statement:

1. I have had time to carefully read and understand the information sheet provided by the researcher for the study. I have been given the opportunity to consider all the information and ask questions, either by email, Skype, telephone or face-to-face, about any aspect of the study. All questions I have asked has been answered clearly and satisfactorily, enabling me to deliver truly informed consent for the research.

Initials

2. I acknowledge that my participation for the research is entirely voluntary. I am aware that I can withdraw from the research at any stage without having to provide an explanation. I understand that there will not be any implications or repercussions after withdrawing from the research.

Initials

3. I understand that I am able to partake in the interview. I acknowledge that all the information and data provided will be audio-recorded and later transcribed. I am aware that **all** information and data will be confidential and anonymised, unless stated otherwise by myself.

Initials

4. I am aware that the information and data provided could be used in the PhD thesis and other academic publications.

Initials

5. I consent for the researcher to conduct the interviews via telephone or enter my home or another convenient location/facility to conduct the interviews. I acknowledge that the discussion will be audio-recorded at this time.

Initials

6. I have been made aware that I am able to request a summary of the study at any time after the completion of the research.

Initials

Providing your name, date and signature indicates that you agree to participate in the study.

Name Date Signature

Appendix 5: Information Sheet and Consent form for Professionals

Introduction

My name is Emyr Wile and I am currently studying for a PhD in Law at Swansea University. I would like to invite you to take part in the research study. As part of the PhD research, I am seeking to recruit professionals to obtain their experiences of the NHS antenatal trisomy screening and testing pathway across **Wales and England**. That is, screening and testing for Down's Syndrome (DS), Edwards' Syndrome and Patau's Syndrome (PS). Please note that this research will not explore your experience of the 20-week fetal anomaly scan. Your participation in the study will be entirely voluntary. I would like to brief you on the implications of the study and why it is being conducted before you decide to participate in the research. Section (A) will explain the purpose of the study and what will happen if you take part and section (B) will give you further detailed information about the conduct of the study. If you would like to participate in the research, please contact me by email, telephone or letter. If you would like to meet with me personally before conducting the study, I would be happy to do so.

Section (A) – The Implications of the Study and its Components

Why am I conducting the study?

Often, antenatal screening and testing is a difficult and overwhelming experience for parents. With the advancements in screening technology and access to testing, which is deemed to be more accurate, it is becoming increasingly common for prospective parents to embark on the antenatal trisomy screening and testing pathway to determine their chance of having a baby with, commonly, Down's Syndrome, Edwards' Syndrome and Patau's Syndrome. This has significant legal and ethical implications on professionals and patients. I aim to explore whether prospective parents are being given sufficient information and support at all stages of the antenatal screening and testing pathway to deliver truly informed consent. Consent is a dynamic process and I wish to discover whether this is sufficiently appreciated throughout the screening and testing pathway. With the recent implementation of NIPT by the NHS in Wales in 2018 (and its anticipated implementation by the NHS in England), it only amplifies the need to explore this issue. I will decipher whether current legal standards of care are being met in the context of trisomy screening and testing. The research will examine a range of potential themes to identify any demographic trends or patterns in the standard of care being delivered by the NHS. Examples of some of the issues that will be explored are:

- To evaluate whether healthcare professionals (HCP) are meeting current legal standards of care in the context of informed consent.
- Whether expectant parents *understand* the information received on antenatal screening and testing to sufficiently facilitate informed decision-making and consent.
- To evaluate whether balanced and accurate information is being delivered by HCP on antenatal screening and testing to facilitate informed consent.
- To discover whether women understand that it is their choice to have antenatal screening and testing, or do they feel that it is part of their antenatal care?
- Whether HCP make it clear to parents that they can disembark from the pathway or decline further investigative testing at any time.
- To ensure that HCPs remain non-directive and non-biased when communicating information in pre-and post-test consultation and counselling.
- To obtain the opinion and views of parents towards the trisomy screening and testing pathway.

Why have you been invited to take part in the research?

This study would significantly benefit from your participation as you perform a professional role across the NHS trisomy pathway.

Do you have to take part in the research?

Your participation is entirely voluntary. I am happy to describe the study and go through this information sheet with you before you make a decision. I will ask you to sign a consent form before your participation. Your information will remain anonymous unless stated otherwise. I will irrevocably strip the information of any direct identifiers such as your name, address, race or gender. Your identity will be replaced by a study number. I could also meet with you at a neutral/convenient/comfortable location if you are concerned that your confidentiality or anonymity will be breached. You are free to withdraw from the research at **any** time, without giving a reason for doing so. Section (B) provides an explanation of the implications of withdrawal.

What is required from you if you decide to take part in the research?

If you kindly decide to take part in the research, you will be required to fill a questionnaire. This can be conducted online, via telephone or face-to-face. Following the completion of the questionnaire, there is an optional follow-up interview. Again, this can be conducted by telephone or face-to-face. Participants can attend on their own or with partners – it is entirely personal preference. If you are a single mother or father, this does not have any effect on your participation. Depending on whether you agree to partake in the interview, you will be required to answer a series of questions and comment on identified issues. You must be **over 18 years of age** to participate in the research for ethical purposes.

The location of the interview is highly flexible. I am able to conduct the interview with yourself at a convenient location, such as your home or a nearby facility. The interview should take approximately 40 minutes to an hour, depending on any additional comments you would like to contribute. The interview will be recorded through an available audio-device which will be later transcribed. Your consent would be required for me to do so. You are able to withdraw your consent to this at any stage of the research, without explanation. If you would like me to record the interview through another method, I would be happy to oblige. Unless stated otherwise, any direct identifiers to yourself will be removed to the best of my ability.

Further support

You should be reminded that you are free to withdraw from the research at any point, without having to provide an explanation. All of the information you have provided would then be immediately deleted and destroyed.

What are the possible benefits of taking part in the research?

I cannot promise that the study will help you directly. However, the information that I obtain from the study will help improve the experiences of other parents who embark on the trisomy screening and testing pathway. It will also further improve current standards of care for the benefit of both parents and professionals. This will provide a strategy to better inform practice guidelines and the decision-making/choices of parents throughout the screening pathway.

What happens after the research study has finished?

Following the completion of the study, you will receive a summary of the research findings. You will be able to double-check that you are anonymised and that the quotations you provided are accurate and representative. The information and data that I have obtained from yourself will be published

in my PhD thesis which will be examined and approved by Swansea University. The data will be read by external assessors, also. Potentially, subject to your approval, the data could be used as part of an academic publication such as journal articles, book chapters and books.

What if there is a problem?

Any complaint in terms of the research such as the way you have been dealt with during the study or any possible harm you have suffered will be addressed. Section (B) contains detailed contact information if this circumstance should arise.

Will your taking part in the research be kept confidential?

Yes. Ethical and legal practice will be strictly followed, and all information provided by yourself will be handled in confidence. Further details on this matter is contained in Section (B).

Section (B) – Further Information

Contact details

If you have any questions in relation to the research or the information provided, please feel free to contact me directly (telephone - [REDACTED], or by email - [REDACTED]). If you would like to speak to my supervisors, again, please do not hesitate to contact them (Professor Karen Morrow; [REDACTED] and Trish Rees; [REDACTED]).

Complaints procedure

If you have a concern about *any* aspects of the research study, please contact myself on my telephone or email and I will do my best to answer your questions. If you are still unsatisfied or unhappy and wish to complain formally, please contact my supervisors via their emails provided above.

Can you withdraw from the study?

Yes. You are able to withdraw from the study at *any* time, without needing to provide an explanation. If you do wish to withdraw from the research, please let me know at your earliest convenience so that I can act immediately on your request. All information and data will be instantly deleted and destroyed. If you wish to re-engage with the research, you are able to do so at any time.

Will your information and data be kept confidential at all times?

Yes. Unless you specify otherwise, any direct identifiers will be removed such as your name, race and gender and will be replaced with a study number if you deem that to be appropriate. The data will be collected via audio-recordings and field notes. You have the right to check the accuracy of the data collected and to correct any errors. All the data you provide such as audio-recordings, field notes, transcribed interviews and your personal data will be stored in a locked filing cabinet on a secure Swansea University premises or on a computerised file secured by a password which is only accessible to myself and agreed members of staff. I understand that information that contains personal or identifiable data will fall underneath the ambit of the Data Protection Act. I will ensure that I follow the University's protocols for Data Protection. Your information and data will be held on file (computerised and non-computerised) at the University in a location which is only accessible to myself and my supervisors (Karen Morrow and Trish Rees). Firewalls, anti-virus software and other measures will be undertaken to ensure data protection. I will not store any data on my personal devices as they could get lost or stolen. After the research has completed, all data will be deleted and destroyed.

Who has reviewed the study?

The research study has been approved by the ethics research committee at Swansea University.

Financial/Organisational elements of the research?

My PhD research is being sponsored by Swansea University and is funded by the Economic and Social Research Council (ESRC). If you have any questions in relation to the funding body, please do not hesitate to ask. I am the Swansea University student representative for the ESRC.

Does this have any effect on the data?

As I am funded by the ESRC, a requirement for researchers is that the data will be given to the UK Data Archive (UKDA). As such, this will include the transcribed interview from yourself. However, any potential identifiers will be stripped from the data so that it is entirely unidentifiable and anonymised. In accordance with Swansea University’s data retention period, the materials (notes of the interviews and the audio-recordings) will be kept for a minimum of five years after the research has finished. After this period, the data will be destroyed in accordance with Swansea University’s adherence to legislation, such as the Data Protection Act and Freedom of Information Act.

Further details

If there is any further information that you would like to obtain in relation to the research, please do not hesitate to get in contact with myself or my supervisors. If you would like to meet with me in person to discuss any of the information provided, I will be more than happy to do so. Thank you for taking the time to read this information sheet.

Consent Form for Professionals

Please provide your initials below each statement:

- 7. I have had time to carefully read and understand the information sheet provided by the researcher for the study. I have been given the opportunity to consider all the information and ask questions, either by email, Skype, telephone or face-to-face, about any aspect of the study. All questions I have asked has been answered clearly and satisfactorily, enabling me to deliver truly informed consent for the research.

Initials

- 8. I acknowledge that my participation for the research is entirely voluntary. I am aware that I can withdraw from the research at any stage without having to provide an explanation. I understand that there will not be any implications or repercussions after withdrawing from the research.

Initials

- 9. I understand that I am able to partake in the interview. I acknowledge that all the information and data provided will be audio-recorded and later transcribed. I am aware that **all** information and data will be confidential and anonymised, unless stated otherwise by myself.

Initials

10. I am aware that the information and data provided could be used in the PhD thesis and other academic publications.

Initials

11. I consent for the researcher to conduct the interviews via telephone or enter my home or another convenient location/facility to conduct the interviews. I acknowledge that the discussion will be audio-recorded at this time.

Initials

12. I have been made aware that I am able to request a summary of the study at any time after the completion of the research.

Initials

Providing your name, date and signature indicates that you agree to participate in the study.

Name Date Signature

Appendix 6: Survey Questions on Trisomy Screening and Testing

This survey will focus on your experience of informed consent along the NHS trisomy screening and testing pathway. Trisomy screening is the medical term to screen and test for Down's Syndrome, Edwards' Syndrome and Patau's Syndrome. Please note that this survey will not explore your experience of the 20-week fetal anomaly scan. Please avoid writing anything that will directly identify you as your participation is meant to be confidential and anonymous. You will be given an opportunity to expand on your answers from the survey in a follow-up interview.

Before completing the survey questions, can you please confirm some background information:

- If you are over the age of 18:

YES NO

- The country in which you received NHS trisomy screening/testing:

ENGLAND WALES Prefer not to say

- If you began trisomy screening/testing within the last 12 months:

YES NO If YES, what year: 2018 2019

- If you are happy to confirm which conditions you decided to screen for (Down's Syndrome and/or Edwards' Syndrome and Patau's Syndrome), please state the detected risk-category following screening:

Down's Syndrome Edwards' Syndrome Patau's Syndrome

Detected risk-category: _____ Prefer not to say

- Whether you received a positive diagnosis for Down's Syndrome, Edwards' Syndrome or Patau's Syndrome if you decided to have diagnostic testing (amniocentesis or CVS):

YES NO Did not receive diagnostic testing Prefer not to say

- Whether you had a child with Down's Syndrome, Edwards' Syndrome or Patau's Syndrome from the pregnancy in question:

Down's Syndrome Edwards' Syndrome Patau's Syndrome Prefer not to say

Survey Questions

1) After receiving information (paper-based, online and/or verbal) from the healthcare professional (HCP) at first contact or the booking appointment, do you feel they provided the information you needed on the purpose of trisomy screening and testing?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

2) After receiving information from the HCP(s) on trisomy screening and testing at first contact or the booking appointment, did the information help support your understanding of the conditions being screened for, that is, Down's Syndrome and/or Edwards' Syndrome and Patau's Syndrome?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

3) After receiving information from the HCP(s) on trisomy screening and testing at first contact or the booking appointment, did the information help support your understanding of the methods of screening and testing?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

4) Did you understand that the decision to have trisomy screening and/or testing was entirely your choice?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

5) Did the HCPs make you aware that you were able to refuse trisomy screening or testing at any time?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

6) Do you feel that the HCP(s) adopted an unbiased approach when supporting your decision-making throughout the pathway?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

7) Do you feel that the HCP(s) effectively communicated with you to support your decision-making for trisomy screening?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

8) Did you understand what high-chance (high-risk) and low-chance (low-risk) meant in relation to your trisomy screening result?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

9) Do you feel that the HCP(s) placed *equal* importance on supporting your understanding of both the advantages and disadvantages of your chosen methods of screening and/or testing?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

10) Do you feel that there were any barriers to communication between you and the HCPs before consenting to decisions on trisomy screening and/or testing?

Strongly agree

Agree

Neutral

Disagree

Strongly Disagree

N/A

Any other comments?

Appendix 7: Survey Questions on Trisomy Screening and Testing for Healthcare Professionals

This survey will focus on the interests of healthcare professionals for securing informed consent along the NHS trisomy screening and testing pathway. Please note that this survey will not include questions on the 20-week fetal anomaly scan. Please avoid writing anything that will directly identify you as your participation is meant to be confidential and anonymous. Thank you for your time and cooperation.

Before answering the survey questions, can you please confirm:

What your professional role is on the NHS trisomy screening and testing pathway:

The country in which you work: _____

1) Do you believe there are appropriate systems in place to secure parents' consent?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

2) Are you clear on your role for securing parents' consent for trisomy screening?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

3) Are you confident supporting parents' informational needs on trisomy screening?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

4) Are you confident supporting parents' understanding of trisomy screening?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

5) Do you feel confident supporting parents' choices along the trisomy pathway?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

6) Do you feel confident supporting parents' understanding of 'risk' in the context of their screening results?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

7) Do you feel that there are barriers to communication between professional and parent along the trisomy pathway?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

8) Do you feel you have received up to date training on securing consent since the implementation of the trisomy pathway?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

9) Do you feel that you have received up to date training on supporting parent understanding of DS, ES and PS since the implementation of the pathway?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

10) Do you feel that you have received up to date training on non-invasive prenatal testing (NIPT) for the purpose of supporting parent decision-making?

Strongly Disagree *Disagree* *Neutral* *Agree* *Strongly Agree* *N/A*

Please explain your answer here:

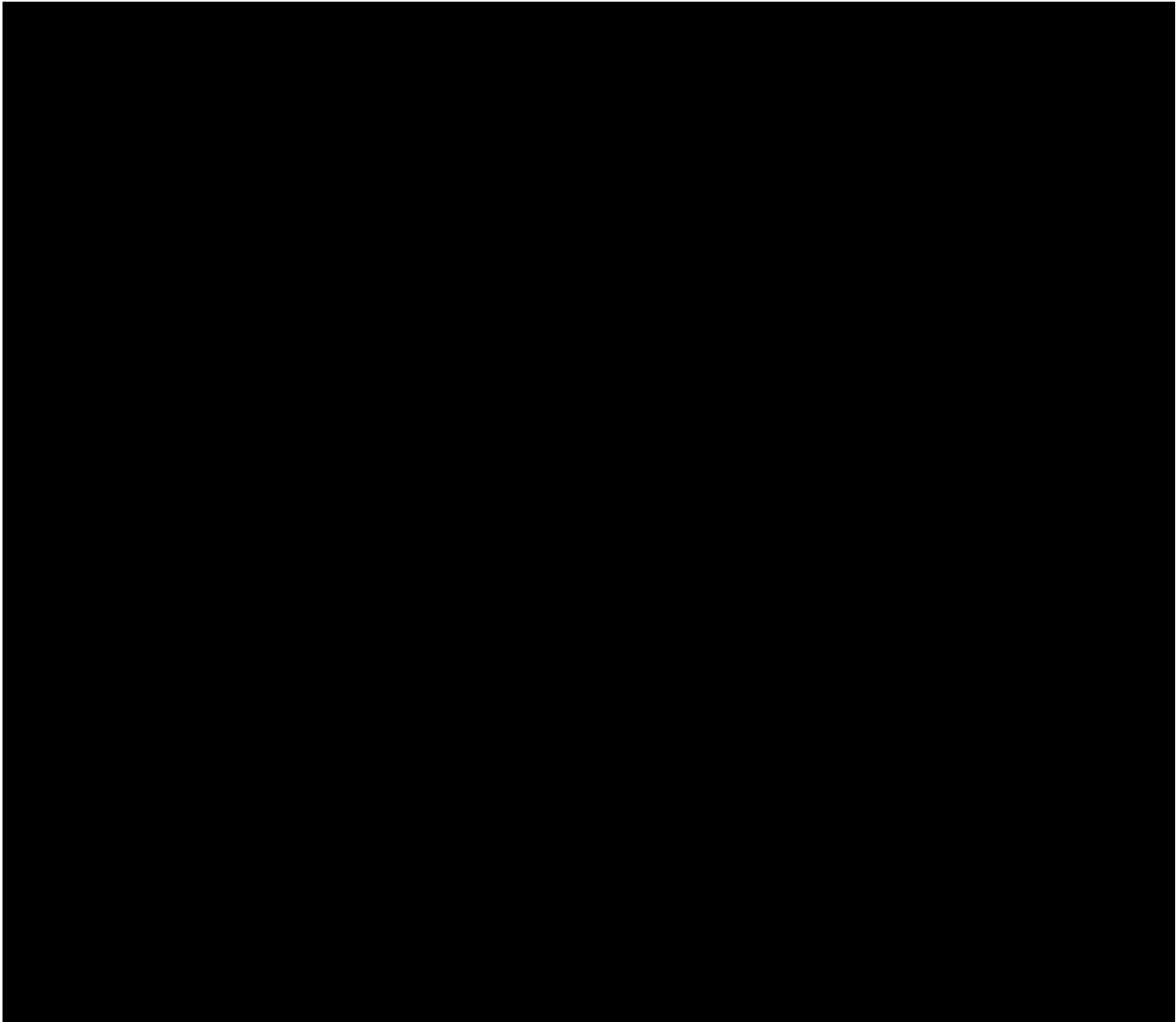
Thank you for taking part in the survey.

Appendix 8: Interview Questions for Parents

Thank you for completing the survey and for taking part in the follow-up interview. As you may notice, similar questions from the survey appear in the interview. This is designed to give you an opportunity to explain the reasons behind the answers you gave for the survey questions. If you are in possession of the information materials you received on trisomy screening as part of your pregnancy information pack, please bring these to the interview. There are questions in the interview which relate to non-invasive prenatal testing (NIPT). If you feel that you cannot provide a comment to the questions on NIPT, you do not have to answer them. If you *received* NIPT either privately or on the NHS, I kindly ask if you could take part in a separate interview to explore your experience of it. If it can be helped, please try to avoid telling me anything in the interview that could reveal your identity. Thank you for your time and cooperation.

- 1) Following your first visit or booking appointment, how would you suggest healthcare professionals (HCPs) ensure that expectant parents are provided with the information they need (paper-based, online and/or verbal) on the purpose of trisomy screening and testing?
- 2) Following your first visit or booking appointment, how would you suggest HCPs ensure that expectant parents are provided with the information they need on the methods of trisomy screening and testing?
- 3) Following your first visit or booking appointment, how would you suggest HCPs ensure that expectant parents are provided with the information they need on the conditions being screened for, that is, Down's Syndrome, Edwards' Syndrome and/or Patau's Syndrome?
- 4) Following your first visit or booking appointment, how would you suggest healthcare professionals (HCPs) support expectant parents understanding of the purpose of trisomy screening and testing? Please provide reasons for your answer.
- 5) Following your first visit or booking appointment, how would you suggest HCPs support expectant parents understanding of the methods of trisomy screening and testing? Please provide reasons for your answer.
- 6) Following your first visit or booking appointment, how would you suggest HCPs support expectant parents understanding of the conditions being screened for, that is, Down's Syndrome and/or Edwards' Syndrome and Patau's Syndrome? Please provide reasons for your answer.
- 7) From your experience, how would you suggest HCPs ensure that expectant parents understand the decision to have trisomy screening and/or testing is entirely their choice before consenting to it? Please give reasons for your answer.
- 8) From your experience, how would you suggest HCPs emphasise the importance of securing your informed consent for trisomy screening and/or testing?
- 9) From your experience, how would you suggest HCPs support expectant parents understanding of the difference between screening and diagnostic testing before giving their consent? Please provide reasons for your answer.
- 10) From your experience, what is your opinion on HCPs withholding information about certain aspects of the trisomy screening and testing pathway? Please provide reasons for your answer.

- 11) From your experience, how would you suggest HCPs support expectant parents understanding of the decisions they may be required to make along both the low-chance (low-risk) and high-chance (high-risk) pathways? Please provide reasons for your answer.
- 12) From your experience, how would you suggest HCPs place equal importance on supporting the understanding of expectant parents of both the advantages and disadvantages of the chosen methods of screening and/or testing? Please provide reasons for your answer.
- 13) From your experience, how would you suggest HCPs support expectant parents' understanding of the terms high-chance (high-risk) and low-chance (low-risk) in relation to their screening result before consenting to trisomy screening? Please provide reasons for your answer.
- 14) From your experience, if you were given the option to have non-invasive prenatal testing (NIPT), how would you suggest HCP(s) support the understanding of expectant parents of the purpose of NIPT? Please provide reasons for your answer.
- 15) From your experience, if you were given the option to have NIPT, how would you suggest HCPs place equal importance on supporting the understanding of expectant parents on both the advantages and disadvantages of NIPT? Please provide reasons for your answer.
- 16) From your experience, how would you suggest HCPs check that any gaps in the expectant parents understanding are filled before being required to consent to their decisions on trisomy screening and/or testing? Please provide reasons for your answers.
- 17) From your experience, how would you suggest HCPs approach questions from expectant parents if they want more information or support before giving consent to screening and/or testing? If you felt the need to ask questions, what questions did you ask and why did you feel the need to ask them? Please provide reasons for your answer.
- 18) From your experience, how would you suggest HCPs have an unbiased approach when communicating information on trisomy screening and testing, and maintain this approach throughout the trisomy screening and testing pathway? Please provide reasons for your answer.
- 19) If you had a partner present during trisomy screening and/or testing, how would you suggest HCPs give partners the opportunity to be included in the decisions made on trisomy screening and/or testing? Please provide reasons for your answer.
- 20) Please tell me what you think is the most important part of the HCP(s) role when making decisions on trisomy screening and testing? When answering this question, tell me what you expect from the role of the HCP(s).
- 21) From your experience, what type of barrier could disrupt communication between HCPs and expectant parents before consenting to decisions on trisomy screening and/or testing? Please provide reasons for your answer.
- 22) From your experience, how would you suggest that HCPs check that expectant parents have received information that is relevant to their own needs and expectations when making decisions on trisomy screening and/or testing? Was there anything more the HCPs could have done to tailor the information to your individual situation? Please provide reasons for your answer.
- 23) From your experience, how would you define informed consent and what are the most important aspects of it?



Appendix 9: Interview Schedule and Open-Survey Questions for Healthcare Professionals

Thank you for completing the survey and for taking part in the follow-up interview. As you may notice, similar questions from the survey appear in the interview. This is designed to give you an opportunity to explain the reasons behind the answers you gave for the survey questions.

Question Schedule

Question 1: How do you secure parent consent for trisomy screening?

Question 2: How do you support parents' informational needs on trisomy screening before securing consent?

Question 3: How do you support parents' understanding of trisomy screening before securing consent?

Question 4: How do you support parents' choices along the trisomy screening pathway?

Question 5: How do you effectively communicate with parents along the trisomy screening pathway?

Question 6: How do you support parents' understanding of 'risk' associated with trisomy screening, in terms of the available methods of screening and testing?

Question 7: How do you support parents' understanding of non-invasive prenatal testing (NIPT) before securing their consent?

Question 8: What barriers do you face in practice that could impact on parents delivering informed consent for trisomy screening?

Question 9: How do you overcome these barriers to effective secure parent consent? Please provide an example.

Question 10: What changes, if any, would you make to existing systems to ensure that parents are effectively delivering informed consent?

Appendix 10: Glossary of Terms

The content of the glossary of terms for this thesis is heavily reliant on the NHS FASP Programme Handbook (2018),¹⁷¹³ Antenatal Screening Wales (ASW)¹⁷¹⁴ and NHS Scotland Screening Programme glossary of terms. It is an amalgamation of these NHS programme documents. The document is a reliable means of explaining the medical terms used throughout the thesis. It represents common usage, making the thesis more accessible to readers. Most terms are referenced directly from the NHS FASP Programme Handbook, Antenatal Screening Wales (ASW) and NHS Scotland Programme glossary of terms. Additional information was sourced from other scholarly/medical literature which had not been covered by the NHS programme glossary of terms.

Amniocentesis: An invasive procedure undertaken from about 15 completed weeks (15+0) onwards to obtain a sample of amniotic fluid (liquor) surrounding the fetus. Using an aseptic technique whilst under continuous ultrasound guidance, a sterile needle is passed through the mother's abdomen, uterus and amniotic sac. A sample of amniotic fluid is aspirated with a syringe and sent for analysis to test for a range of chromosomal and inherited disorders. Up to 1 out of every 100 women who have an amniocentesis will miscarry.

Amniotic fluid: Also known as 'liquor', this is the fluid surrounding the fetus during pregnancy. It contains substances and cells from the fetus, which can be removed by amniocentesis and examined.

Antenatal: The period from conception to birth.

Biochemical markers: Analytes (commonly referred to as markers) measured by the laboratory that are used to calculate the likelihood of a pregnancy being affected by a condition or syndrome.

Chance: The likelihood that an event will occur.

Chance cut-off: Determines those women who are in the 'higher chance' group and considered 'screen positive'.

Chorionic villus sampling (CVS): An abdominal or cervical procedure performed under continuous ultrasound guidance after 10 completed weeks in pregnancy to obtain a sample of placental tissue for chromosomal or genetic analysis (between 10-13 weeks gestation). The range of chromosomal and genetic conditions that can be detected is similar to those for amniocentesis. Up to 1 out of every 100 women who have a CVS will miscarry.

Chromatid: Each chromosome is comprised of two chromatids, that is, the strand-like structure. In a chromatid, the DNA molecules are unwound.

Chromosome: Structures found in the nucleus of cells, composed of DNA and proteins. Normally humans have 46 chromosomes in each cell, 23 from each parent. Of these, 22 are autosomes and one is a sex chromosome.

Chromosome anomaly: A change in the number or arrangement of the normal 23 pairs of chromosomes.

Combined test: Between 11+2 weeks and 14+1 weeks of pregnancy, a combination of the nuchal scan measurement and a blood sample from the mother which measures the concentration of pregnancy

¹⁷¹³ NHS England. Public Health England. NHS Fetal Anomaly Screening Programme Handbook, Valid from August 2018.

¹⁷¹⁴ Antenatal Screening Wales Policy, Standards and Protocols 2019 – 7.0 and 8.0 Antenatal Screening for DS, ES and PS.

associated plasmprotein-A(PAPP-A), and free beta human chorionic gonadotrophin(freebetahCG). Together with the mother's age, the gestation of the pregnancy and the crown rump length (CRL) measurement (between 45.0mm and 84.0mm), these are used to estimate the chances that the fetus is affected with Down's syndrome, Edwards' syndrome or Patau's syndrome.

Confirmed Result: The results of initial screening tests are not usually 100% certain and are often called presumptive results. The results of screening tests are NOT confirmed results. They are confirmed later, with further diagnostic tests.

Congenital: Present at or shortly after birth.

Congenital anomaly: An anomaly present at birth, although not necessarily hereditary.

Crown rump length (CRL): Ultrasound measurement between the top of the head (crown) to the bottom of the buttocks (rump). To be eligible for first trimester combined screening as part of the NHS screening programme the CRL should measure between 45.0mm and 84.0mm.

Detection rate: The proportion of affected individuals with a positive screening result.

Diagnostic test: Refers to the process involved in obtaining a definite diagnosis. For example the diagnostic test on an amniocentesis sample (invasive procedure) is the full karyotype or QF-PCR.

DNA (deoxyribonucleic acid): The molecule that encodes genetic information. DNA is a double-stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contained the bases: adenine (A), guanine (G), cytosine (C), and thymine (T). In nature, base pairs form only between A and T and between G and C.

Embryo: A fertilised ovum (egg) in the early stage of development. In humans the term is reserved for the first eight weeks of development.

False-negative result: Screening tests divide people into lower and higher-risk groups. Some people with a negative screening test result do actually have the condition being screened for. These people are said to have a 'false-negative' result.

False-positive result: Screening tests divide people into lower and higher-risk groups. Some people with a positive screening test result do not actually have the condition being screened for. These people are said to have a 'false-positive' result.

Family history: History of a condition in at least one of the following family members: parent, sibling, grandparent, great-grandparent, aunt, uncle, nephew, niece or cousin or child.

Fetal anomaly: Structural abnormalities with how the fetus has developed.

Fetal anomaly ultrasound scan: A detailed ultrasound scan, sometimes called the mid-pregnancy or 20-week scan. It is a screening test offered to all pregnant women and is usually carried out between 18 and 21 weeks of pregnancy. It produces a 2-dimensional black and white image that gives only a side view of the baby and it checks for major physical anomalies in the baby; although it can't pick up every anomaly. This scan permits prospective parents to consider screening as an option, identify serious abnormalities in the fetus which can result in a continuation or termination of the pregnancy and identifies whether an abnormality can benefit from any kind of early treatment.

The 20-week fetal anomaly ultrasound scan is used to detect major structural differences in the baby. There are 11 conditions that are screened for using this method. The 20-week scan is not associated with screening for Down's syndrome as it is not a reliable means for doing so – only detects around

50% of baby's with Down's syndrome. There is an association with an enlarged nuchal translucency (NT) measurement at 12-weeks, but this is not diagnostic.

Edwards' and Patau's syndrome are one of the detectable conditions on the 20-week fetal anomaly scan. There are structural differences that strongly incline whether the baby has Patau's and Edwards' syndromes, such as non-division of the brain, non-formation of the brain, facial clefts, significant heart defects and talipes to name just a few. Some of these characteristics are detected at the 12-week scan, however the majority are not visible until the 20-week anomaly scan. The anomaly scan itself is not specifically designed to scan for chromosomal anomalies in the same manner that the 12-week scan is for Down's syndrome. While particular characteristics that point towards chromosomal aneuploidy are detected on the anomaly scan, HCP's use the scan to only screen for structural defects. T21, 18 and 13 are said to commonly be incidental findings on the 20-week scan.

Gene: The unit of a chromosome through which particular characteristics are inherited from one or both parents.

Genetic counselling: Information and support provided by an appropriately trained health professional, to individuals who have known conditions in their families or who are concerned about the future possibility of genetically inherited conditions.

Genetic counsellor: A health professional with specialised training in genetics and counselling who can provide information and support for individuals or families with concerns about a genetic disorder that may run in the family.

Genetic testing: Examination of an individual's genetic material to identify alterations that may cause a disorder.

Genetics: 1. The study of the structure and function of genes. 2. The genetic features which occur in individuals, families and populations.

Gestational age: The duration of an ongoing or completed pregnancy, measured from the first day of the last menstrual period (usually about two weeks longer than that measured from conception). Gestational age is usually measured in weeks and days.

Homologous: Similarity in internal or chromosomal structures.

Incidence: The number of new instances of a specific condition occurring during a certain period in a specified population.

Inheritance: The passing of familial characteristics from one generation to the next.

Inherited: Having a hereditary characteristic; there are many inherited characteristics, including eye colour, hair colour and health disorders.

Invasive diagnostic procedure: A method used to obtain a sample used to aid diagnosis, for example, amniocentesis or chorionic villus sampling.

IVF: This stands for in vitro fertilisation. This simply means that an egg is fertilised with a sperm under controlled laboratory conditions and is then inserted into the woman's uterus.

Karyotype: A photomicrograph of an individual's chromosomes arranged in a standard format showing the number, size, and shape of each chromosome type; used to correlate chromosomal anomalies with the characteristics of specific diseases. Karyotyping is often used for antenatal diagnosis of conditions such as Down's Syndrome, Edwards' Syndrome and Patau's Syndrome.

Marker: An identifiable physical location on a chromosome whose inheritance can be monitored. Markers can be expressed regions of DNA (genes) or some segment of DNA with no known coding function but whose pattern of inheritance can be determined.

Miscarriage: Loss of a fetus before the 24th week of pregnancy.

Morbidity: The extent of being affected by a disease or condition. In epidemiology, the morbidity rate is the prevalence of a disease within a particular number of the population.

Morbidity rate: In epidemiology, the prevalence of a disease within a population, usually expressed as cases per 100,000.

Mortality/mortality rate: The incidence of death in a population in a given period.

Mosaic: An individual who has some cells with an unusual genetic or chromosomal make-up while the rest of the cells in the body have the typical genetic or chromosomal constitution.

Mutation: A change in the gene resulting from an error made when the gene is being copied. It may result in altered gene function. 'Alteration' may be more acceptable to women and their families.

Non-invasive: A procedure that does not require incision into the body or the removal of tissue.

Non-invasive prenatal testing (NIPT): Non-invasive Prenatal Testing. A blood test taken early in gestation to identify the chances of the mother having a baby with a tested condition.

Nuchal scan (Nuchal translucency scan NT): Between 11+2 weeks and 14+1 weeks of pregnancy the thickness of fluid in the tissue space within the nape of the fetal neck, the nuchal translucency can be measured. An increased amount of fluid may indicate that the fetus has Down's syndrome, structural or genetic anomaly. By combining the mother's age and the gestation of the pregnancy with information from the scan an individual statistical chance of an anomaly can be given for that particular pregnancy. If the chance is between 1 in 2 and 1 in 150 a diagnostic test, such as CVS, will be offered.

Placenta: The structure that provides the fetus with nourishment during development. It is attached to the wall of the uterus and connects to the fetus through the umbilical cord.

Polymerase chain reaction (PCR): A rapid diagnostic test for the most common chromosomal and genetic anomalies. Using a small amount of amniotic fluid, PCR amplifies and enables specific regions of the DNA molecule to be quantified from uncultured amniocytes. The test is used to provide a definitive diagnosis of Down's syndrome, haemoglobin disorders and other single-gene disorders.

Predisposition: A situation in which a person, due to their inherited genetic makeup, may have a particular susceptibility to a condition if exposed to the appropriate environmental triggers.

Prenatal: Relating to the period before birth.

Prevalence: The proportion of people in a population who have a given disease or attribute.

Prevalence rate: The number of people with the condition or attribute, divided by the population at risk.

Prognosis: Predicted course and outcome of a disorder, based on all the knowledge related to a specific case, e.g., age, sex, the course of the disorder in other patients.

Quadruple test: Second trimester test (14-20 weeks gestation) to calculate the chance of the pregnancy being affected by Down's syndrome, usually based on the measurement of AFP, uE3, free beta hCG (or total hCG), and inhibin-A together with the woman's age. The head circumference (HC) is also measured to determine whether it is equal to or more than 101.0mm.

Quality assurance (QA): A system for monitoring and maintaining high standards in every aspect of a screening programme.

Screening: Testing people who do not have or have not recognised the signs or symptoms of the condition being tested for, either with the aim of reducing risk of an adverse outcome, or with the aim of giving information about risk.

Screening pathway: The whole system of activities needed to deliver high quality screening. It ranges from identifying and informing those to be offered screening through to the treatment and follow up of those found to have abnormality, and support for those who develop disease despite screening.

Screen positive rate (SPR): The number of women who receive a higher chance result.

Screen negative rate (SNR): The number of women who receive a lower chance result.

Screening programme: The whole system of activities needed to deliver high quality screening. It ranges from identifying and informing those to be offered screening through to the treatment and follow up of those found to have abnormality, and support for those who develop disease despite screening.

Screening safety incident: An unintended or unexpected incident(s) that could have or did lead to harm to one or more persons who are eligible for NHS screening; or to staff working in the screening programme.

Screening test: A test or inquiry used on people who do not have or have not recognised the signs or symptoms of the condition being tested for. It divides people into lower and higher chance groups.

Syndrome: Combination of symptoms and signs grouped together to form a disorder.

Termination of pregnancy: The medical expulsion or extraction from the uterus of a fetus in the first, second or third trimester of pregnancy.

Trisomy: Three copies of a particular chromosome rather than two.

True-negative result: Screening tests divide people into low and higher risk groups. Most of the people with a negative screening test result do not have the condition being screened for. These people are said to have a 'true-negative' result.

True-positive result: Screening tests divide people into low and higher risk groups. Some people with a positive screening test result do have the condition being screened for. These people are said to have a 'true-positive' result.

Twins: May be genetically identical (monozygous) when they arise from a single fertilised egg or non-identical (dizygous) when they arise from two separate eggs.

Ultrasound scan: A ultrasound scan is a safe and painless test that uses sound waves to make images. It is like radar.

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