NON-INVASIVE PRENATAL TESTING FOR ADULT-ONSET CONDITIONS: REPRODUCTIVE CHOICE AND THE WELFARE OF THE FUTURE CHILD

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Non-invasive prenatal testing ('NIPT') enables quick, safe and generally reliable screening during early pregnancy. NIPT is currently available in Australia for screening for conditions such as trisomy 21 ('Down Syndrome') which are immediately apparent at birth. NIPT may also be used in the future to detect adult-onset disorders and risk profiles for conditions which manifest later in life. Although expanded prenatal testing has the potential to enhance reproductive choice, the use of NIPT to detect adult-onset conditions raises significant ethical concerns about the welfare of the future child. In this article, we explore the legal and ethical issues raised by NIPT for adult-onset conditions. We make some preliminary proposals for regulating access to NIPT, which encourage prospective parents to consider the interests of the future child through careful genetic counselling.

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I Introduction

Prenatal genetic testing is often perceived as a routine aspect of antenatal care. With the introduction of non-invasive prenatal testing ('NIPT'), it is easier and safer to access screening for a wide variety of genetic conditions. NIPT involves testing a sample of a pregnant woman's blood, which contains cell-free fetal DNA ('cffDNA'). NIPT has high accuracy for certain conditions, is less invasive than other prenatal testing, and can be done very early in gestation (10–12 weeks).[1] NIPT is already publicly subsidised in some countries, such as Belgium,[2] and is available on a private basis in others, including Australia. NIPT is currently used to screen for conditions such as trisomy 21 ('Down Syndrome'), which are immediately apparent at birth. NIPT may also be used in the future to detect adult-onset disorders and risk profiles for conditions which manifest later in life, including through whole-genome sequencing.

Expanded prenatal testing has the potential to enhance reproductive choice and informed decision-making. However, the use of NIPT to detect adult-onset conditions raises significant ethical concerns about the welfare of the future child, particularly where a woman chooses to continue a pregnancy following a fetal diagnosis. Prenatal testing for disorders that are immediately apparent at birth, or apparent soon after, poses comparatively few ethical concerns compared to testing for those that manifest later in life. In this article, we focus

[1] For example, NIPT has a sensitivity of 99% for trisomy 21 ('Down Syndrome'): Sian Taylor-Phillips et al, 'Accuracy of Non-Invasive Prenatal Testing Using Cell-Free DNA for Detection of Down, Edwards and Patau Syndromes: A Systematic Review and Meta-Analysis' (2016) 6(1) *BMJ Open* 1, 1, 4, 6.

[2] Emilia Kostenko et al, 'Clinical and Economic Impact of Adopting Noninvasive Prenatal Testing as a Primary Screening Method for Fetal Aneuploidies in the General Pregnancy Population' (2019) 45(6) *Fetal Diagnosis and Therapy* 413, 414.

on the impact of predictive testing for adult-onset conditions on the welfare of the future child and whether this should influence the ability of prospective parents to access such prenatal testing. Prospective parents with a known genetic risk for a specific genetic disease generally undertake invasive diagnostic techniques such as chorionic villus sampling to find out if their fetus has this serious condition. In the near future, however, prospective parents with no known history of genetic disease (and who would not ordinarily consider invasive testing) may be persuaded by the low risk and ease of NIPT to test for adult-onset conditions as part of a suite of genetic tests. This is more likely if NIPT replaces the combined first-trimester screening test and is publicly subsidised.[3] NIPT differs from other forms of prenatal testing as it significantly lowers the barrier for accessing fetal information and is more likely to be implemented as a routine part of prenatal care than more invasive diagnostic tests. Trisomy 21 screening programs have a very high uptake, and NIPT is more accurate and soon may be more cost-effective for public healthcare systems than the commonly used current method of combined first-trimester screening.

Although NIPT is not specifically regulated by law, access to this technology is influenced by a combination of professional standards, clinical guidelines and common law principles. Current clinical guidelines highlight a fundamental tension between the interests of prospective parents in making genuinely informed reproductive decisions and the interests of a future child in protection from psychosocial harm, preservation of future autonomy, and non-discrimination. In this article, we unpack these interests by analysing relevant legal and ethical principles applicable to NIPT, with a view to clarifying the specific nature of the interests at stake, the ways in which they collide and intersect, and how they might be managed. We also briefly explore broader societal interests in limiting the scope of NIPT based on concerns about 'designer babies' and eugenics in determining what role, if any, the state has in regulating access to NIPT. Attempts by courts and policymakers to balance the interests of prospective parents with those of the future child and broader society typically employ the language of 'rights', which does not necessarily capture the complex and interconnected nature of the interrelated interests within families. In this article, we frame our discussion within a relational approach that acknowledges the various individual and interconnected interests at stake throughout pregnancy and beyond. There is currently some recognition of the relational nature of the interests of a child within a family in

[3] See Antina de Jong et al, 'Non-Invasive Prenatal Testing: Ethical Issues Explored' (2010) 18(3) *European Journal of Human Genetics* 272, 273.

international guidelines on predictive testing of minors. We build on this approach in proposing a regulatory framework for NIPT.

We begin in Part II by briefly explaining the current status of NIPT technology, before outlining existing guidelines on prenatal and genetic testing, some of which reflect a 'facilitative approach' and others a more 'restrictive approach' to testing. In Part III, we explore the interests of prospective parents and the future child, the interrelationship between them, and how the law has managed the tension between these interests in other contexts. In particular, we examine how the legal principles of bodily integrity and reproductive autonomy have been applied in cases involving refusal of treatment by pregnant women,[4] potentially harmful antenatal behaviour, and prenatal injury. We also critique the ethical and empirical bases for concerns raised about the welfare of the future child in the context of NIPT. Finally, we discuss the broader interests within society in promoting equity and reducing discrimination. In Part IV, we propose a facilitative approach to regulating NIPT for adult-onset conditions that promotes reproductive choice and protects the welfare of the future child. We draw on current regulation of pre-implantation genetic testing ('PGT') in Australia and propose a relational approach to accommodate the various interests at stake in NIPT. We conclude in Part V that this approach could be adapted to apply to NIPT more broadly as this procedure becomes commercially available for a broad range of single-gene disorders, polygenic disease susceptibilities, and other information that can be gained from the whole-genome sequencing of a fetus.

II NIPT and Current Relevant Guidelines

A *Current NIPT Technology and the Regulatory Landscape*

As previously stated, NIPT is safe, has high accuracy for certain conditions, and can be done early in pregnancy. However, it is important to note that NIPT is a screening rather than diagnostic test (that is, it cannot say definitively whether the fetus has the condition or not). This means that if a high-chance result is received (eg for trisomy 21), it is recommended that it be confirmed through prenatal diagnosis techniques such as amniocentesis before any decision is made regarding pregnancy management[5] as false positives and false negatives

[4] For simplicity, we use the phrase 'pregnant women' to describe gestational parents, acknowledging that not all gestational parents identify as women. To date, the discourse and case law have generally adopted this phrase to refer to pregnant people assigned female at birth.

[5] The Royal Australian and New Zealand College of Obstetricians and Gynaecologists, *Prenatal Screening and Diagnostic Testing for Fetal Chromosomal and Genetic Conditions* (Statement, July 2018) 7 recommendation 8.

are still possible with NIPT.[6] However, recent developments using fetal cells rather than cffDNA may allow for these techniques to become diagnostic in the future.[7]

NIPT can also be used for sex determination, which has raised concerns about sex-selective termination of pregnancy.[8] In the near future, the availability of NIPT is likely to be significantly expanded to include testing for single-gene disorders[9] and high-depth whole-genome sequencing,[10] essentially enabling prospective parents to test for any condition with a genetic basis. Predictive testing can include predictors for adult-onset disease such as the *BRCA1* variant for breast or ovarian cancer,[11] or the *ApoE4* variant for early-onset Alzheimer's disease.[12] With the advent of whole-genome sequencing, NIPT could also be expanded to encompass genomic predictions for disease susceptibility, known as polygenic risk scores. These may include a variety of diseases with a polygenic basis, such as type 2 diabetes[13] or mental illness.[14] Predictive testing can also encompass serious single-gene disorders such as

6 Hannah Skrzypek and Lisa Hui, 'Noninvasive Prenatal Testing for Fetal Aneuploidy and Single Gene Disorders' (2017) 42 (July) *Best Practice & Research: Clinical Obstetrics & Gynaecology* 26, 34–5. See also Taylor-Phillips et al (n 1) 7–8.

7 See Pin-Jung Chen et al, 'Noninvasive Prenatal Diagnostics: Recent Developments Using Circulating Fetal Nucleated Cells' (2019) 8(1) *Current Obstetrics and Gynecology Reports* 1.

8 Hilary Bowman-Smart et al, 'Sex Selection and Non-Invasive Prenatal Testing: A Review of Current Practices, Evidence, and Ethical Issues' (2020) 40(4) *Prenatal Diagnosis* 398, 399 ('Sex Selection and Non-Invasive Prenatal Testing').

9 Skrzypek and Hui (n 6) 34–5.

10 See KC Allen Chan et al, 'Second Generation Noninvasive Fetal Genome Analysis Reveals de Novo Mutations, Single-Base Parental Inheritance, and Preferred DNA Ends' (2016) 113(50) *Proceedings of the National Academy of Sciences of the United States of America* E8159; Mary E Norton, 'Noninvasive Prenatal Testing to Analyze the Fetal Genome' (2016) 113(50) *Proceedings of the National Academy of Sciences of the United States of America* 14173; Malgorzata I Srebniak et al, 'Social and Medical Need for Whole Genome High Resolution NIPT' (2020) 8(1) *Molecular Genetics & Genomic Medicine* e1062:1–4, 2.

11 See Sining Chen and Giovanni Parmigiani, 'Meta-Analysis of *BRCA1* and *BRCA2* Penetrance' (2007) 25(11) *Journal of Clinical Oncology* 1329, 1329, 1332.

12 See Cornelia M van Duijn et al, 'Apolipoprotein E4 Allele in a Population-Based Study of Early-Onset Alzheimer's Disease' (1994) 7(1) *Nature Genetics* 74, 76.

13 Amit V Khera et al, 'Genome-Wide Polygenic Scores for Common Diseases Identify Individuals with Risk Equivalent to Monogenic Mutations' (2018) 50(9) *Nature Genetics* 1219, 1219, 1222.

14 Alicia R Martin et al, 'Predicting Polygenic Risk of Psychiatric Disorders' (2019) 86(2) *Biological Psychiatry* 97, 105–6.

Huntington's disease, for which NIPT has been demonstrated in a research context, although it is not currently available to consumers.[15]

Different adult-onset conditions vary in a number of important ways. One factor is treatability or actionability. Huntington's disease is not considered clinically actionable as there is little to be done to prevent or modify the course of the disease.[16] By way of contrast, detecting the *BRCA1* variant can lead to treatments to prevent the onset of breast cancer, such as a preventive mastectomy.[17] These conditions also differ in penetrance. Having the genetic basis for Huntington's disease means that one will develop the disease (although a lower number of trinucleotide repeats does have incomplete penetrance).[18] However, while having the *BRCA1* variant increases breast cancer risk considerably, it does not guarantee that one will develop breast cancer.[19] The use of polygenic risk scores raises its own questions; their validity, level of predictiveness and utility in the clinical setting have been criticised.[20] For example, they demonstrate an association, rather than a causal relationship.[21] Furthermore, pleiotropic effects, where a genetic variant can affect multiple traits, may mean that one score is associated with a variety of traits rather than just the one of interest.[22]

Different conditions may also vary in 'severity' or 'seriousness'.[23] Diabetes or many types of mental illness would generally not be considered as having as great an impact on quality of life as something like Huntington's disease, which

[15] See Jessica ME van den Oever et al, 'Noninvasive Prenatal Diagnosis of Huntington Disease: Detection of the Paternally Inherited Expanded CAG Repeat in Maternal Plasma' (2015) 35(10) *Prenatal Diagnosis* 945, 945–6.

[16] Samuel Frank, 'Treatment of Huntington's Disease' (2014) 11(1) *Neurotherapeutics* 153, 153.

[17] Kelly Metcalfe et al, 'Contralateral Mastectomy and Survival after Breast Cancer in Carriers of BRCA1 and BRCA2 Mutations: Retrospective Analysis' (2014) 348(7945) *BMJ* g226:1–11, 1–3.

[18] Francis O Walker, 'Huntington's Disease' (2007) 369(9557) *Lancet* 218, 220.

[19] Chen and Parmigiani (n 11) 1329.

[20] See, eg, Alicia R Martin et al, 'Clinical Use of Current Polygenic Risk Scores May Exacerbate Health Disparities' (2019) 51(4) *Nature Genetics* 584; L Duncan et al, 'Analysis of Polygenic Risk Score Usage and Performance in Diverse Human Populations' (2019) 10(3328) *Nature Communications* 1; A Cecile JW Janssens, 'Validity of Polygenic Risk Scores: Are We Measuring What We Think We Are?' (2019) 28(R2) *Human Molecular Genetics* R143.

[21] See, eg, Robert Plomin and Sophie von Stumm, 'Polygenic Scores: Prediction versus Explanation' (2022) 27(1) *Molecular Psychiatry* 49, 49.

[22] See Sarah Munday and Julian Savulescu, 'Three Models for the Regulation of Polygenic Scores in Reproduction' (2021) 47(12) *Journal of Medical Ethics* e91:1–9, 2.

[23] The authors acknowledge that these terms are subjective and likely to be influenced by an individual's lived experience, values, preferences, beliefs and (to some extent) access to services and supports.

affects many parts of the body and eventually results in death. Of course, the concept of severity depends on value judgements around what is necessary for a good quality of life and overall wellbeing. These factors, and the way that adult-onset conditions differ with respect to them, are relevant in determining the scope of NIPT. As we discuss later, there may be some threshold of seriousness that is required for NIPT to be either available or funded for certain adult-onset conditions.[24] However, where this threshold falls is a matter of considerable debate, as different parents may have different conceptions of a good life and what constitutes a severe condition. Furthermore, parents may have different risk appetites. For example, one parent may tolerate a 50% risk of an adult-onset condition, while another may not. We propose a flexible approach to such a 'threshold', depending on the impact on the family and future child within their particular context rather than a blanket clinical assessment of seriousness. Overall, parents should be informed that if a risk is low or uncertain (eg a variant of uncertain significance), then the utility of the information may be low and potentially burdensome.

The current scope and availability of NIPT in Australia is influenced by a combination of professional practice standards, clinical guidelines on prenatal and genetic screening, and common law principles relevant to the doctor–patient relationship. In addition, the manufacture and supply of NIPT tests are regulated in Australia by laws governing the quality, production and supply of medical devices.[25] The law regulating access to abortion is also indirectly relevant to NIPT, given that the information obtained by testing may result in a decision to terminate a pregnancy.[26] Specialists in reproductive medicine are regulated over and above the legal and ethical restraints on medical practitioners generally.[27] However, regulation in this specialisation remains

[24] See below Part III(D).

[25] See *Therapeutic Goods Act 1989* (Cth) ('*TGA*'). The blood test for NIPT and the instruments used to carry it out are regulated pursuant to the *Therapeutic Goods (Medical Devices — Specified Articles) Instrument 2020* (Cth) sch 1 item 2 and *TGA* (n 25) s 41BD(2B). The *TGA* (n 25) only regulates *therapeutic* devices so would not cover NIPT for non-medical uses such as sex selection: Isabel Karpin and Kristin Savell, *Perfecting Pregnancy: Law, Disability, and the Future of Reproduction* (Cambridge University Press, 2012) 300.

[26] In Australia, abortion is regulated by both common law and statute and varies from state to state: see, eg, *Termination of Pregnancy Act 2018* (Qld); *Criminal Law Consolidation (Medical Termination of Pregnancy) Regulations 2011* (SA); *Abortion Law Reform Act 2008* (Vic).

[27] In Australia, the doctor–patient relationship is governed by Medical Board of Australia, *Good Medical Practice: A Code of Conduct for Doctors in Australia* (Code of Conduct, October 2020) 27 <https://www.medicalboard.gov.au/codes-guidelines-policies/code-of-conduct.aspx>. This code of conduct is issued by the Medical Board of Australia pursuant to the *Health Practitioner Regulation National Law* (as in force in each state and territory): *Health*

'partial and selective'[28] and focuses primarily on assisted reproductive technology ('ART').[29] For example, access to ART — including PGT — in Australia is specifically regulated by national guidelines and legislation in various states.[30] By way of contrast, and consistent with other common law jurisdictions,[31] there is no regulation directly limiting access to NIPT in Australia. The common law principles of bodily integrity and reproductive autonomy are discussed below in Parts III(B)(1)–(2) of this article, as they are relevant to determining access to NIPT. This Part focuses on current clinical guidelines relevant to NIPT for detecting adult-onset conditions. We discuss the regulation of PGT in more detail in Part IV, as it raises similar issues to NIPT and provides a useful comparator for how NIPT might be regulated.

There are various national and international position statements and clinical guidelines which reflect a range of approaches to how much genetic information parents should be able to obtain about their future child. These include general guidelines on prenatal testing, including for adult-onset conditions,[32] more specific guidelines on prenatal testing for Huntington's

Practitioner Regulation National Law (ACT) Act 2010 (ACT) s 6; *Health Practitioner Regulation (Adoption of National Law) Act 2009* (NSW) s 4; *Health Practitioner Regulation (National Uniform Legislation) Act 2010* (NT) s 4; *Health Practitioner Regulation National Law (South Australia) Act 2010* (SA) sch 2 s 39; *Health Practitioner Regulation National Law Act 2009* (Qld) sch s 39; *Health Practitioner Regulation National Law (Tasmania) Act 2010* (Tas) s 4; *Health Practitioner Regulation National Law (Victoria) Act 2009* (Vic) s 4; *Health Practitioner Regulation National Law (WA) Act 2010* (WA) sch s 39.

[28] Margaret Brazier, 'Regulating the Reproduction Business?' (1999) 7(2) *Medical Law Review* 166, 169.

[29] See Michelle Taylor-Sands, *Saviour Siblings: A Relational Approach to the Welfare of the Child in Selective Reproduction* (Routledge, 2013) 130–2 ('*Saviour Siblings*'). See also Loane Skene, 'Why Legislate on Assisted Reproduction?' in Ian Freckelton and Kerry Petersen (eds), *Controversies in Health Law* (Federation Press, 1999) 266, 267.

[30] National Health and Medical Research Council, *Ethical Guidelines on the Use of Assisted Reproductive Technology in Clinical Practice and Research* (Guidelines, 2017) <https://www.nhmrc.gov.au/file/3776/download?token=QsZvWpw_>, archived at <https://perma.cc/PG6K-Z3WQ> ('*NHMRC Guidelines*'); *Assisted Reproductive Treatment Act 1988* (SA) ('*ART Act (SA)*'); *Assisted Reproductive Treatment Act 2008* (Vic) ('*ART Act (Vic)*'); *Human Reproductive Technology Act 1991* (WA) ('*HRT Act (WA)*').

[31] In the United Kingdom ('UK'), Canada and the United States ('US'), 'there is no direct regulation limiting access to' NIPT: Karpin and Savell (n 25) 298.

[32] See, eg, National Society of Genetic Counselors, *Prenatal Testing for Adult-Onset Conditions* (Position Statement, 26 June 2019) <https://www.nsgc.org/Policy-Research-and-Publications/Position-Statements/Position-Statements/Post/prenatal-testing-for-adult-onset-conditions-1>, archived at <https://perma.cc/G392-GBC4>.

disease[33] and general guidelines on predictive testing for minors.[34] We analyse how the different guidelines attempt to address the potentially conflicting interests of prospective parents and the (future) child, with a view to informing the development of more specific guidelines for NIPT. Broadly speaking, the various guidelines reflect two distinct approaches. The first approach favours the provision of information to prospective parents (the 'facilitative approach'), whereas the second approach prioritises the welfare of the future child by limiting access to certain information (the 'restrictive approach'). The guidelines also differ in how they approach the tension between the interests of prospective (or current) parents and the future (or existing) child. While some guidelines articulate specific rights that should be resolved in favour of either parents or children, other guidelines take a more nuanced approach that acknowledges the relational nature of interests within families and the role of parental discretion in evaluating the welfare of a child. In the following section, we briefly outline current relevant clinical guidelines before arguing in favour of a facilitative approach that acknowledges the relational nature of the interests at stake.

B *General Guidelines on Prenatal Testing*

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists ('RANZCOG') published a general statement on prenatal screening in 2018, which recommended that '[a]ll pregnant women should be provided with information and have timely access to screening tests for fetal chromosome and genetic conditions'.[35]

[33] 'Guidelines for the Molecular Genetics Predictive Test in Huntington's Disease' (1994) 31(7) *Journal of Medical Genetics* 555. These recommendations were drawn up by a committee of represenatives of the International Huntington Association and the World Federation of Neurology Research Group on Huntington's Chorea: at 555. See also R MacLeod et al, 'Recommendations for the Predictive Genetic Test in Huntington's Disease' (2013) 83(3) *Clinical Genetics* 221.

[34] See, eg, Danya F Vears et al, 'Human Genetics Society of Australasia Position Statement: Predictive and Presymptomatic Genetic Testing in Adults and Children' (2020) 23(3) *Twin Research and Human Genetics* 184.

[35] Human Genetics Society of Australasia and Royal Australian and New Zealand College of Obstetricians and Gynaecologists Joint Committee on Prenatal Diagnosis and Screening, *Prenatal Screening and Diagnostic Testing for Fetal Chromosomal and Genetic Conditions* (Statement C-Obs 59, July 2018) 6 recommendation 1 ('*Prenatal Screening and Diagnostic Testing*'). See also Royal Australian and New Zealand College of Obstetricians and Gynaecologists, *Prenatal Screening for Fetal Genetic or Structural Conditions* (Statement C-Obs 35, July 2019) 5 recommendation 1.

RANZCOG explicitly refers to NIPT as an acceptable first-line screening test for fetal chromosome 'abnormalities' in the first and second trimesters.[36] NIPT is currently used in Australia to detect chromosome conditions (as well as sex) but could potentially be used for a wide variety of purposes, including screening for adult-onset conditions. This presents regulators with difficult ethical questions. In 2017, the United Kingdom's ('UK's') Nuffield Council on Bioethics recommended a restrictive approach to regulating NIPT, stating that 'NIPT should not normally be used to test whether a fetus has a less significant medical condition or impairment or an adult onset condition'.[37]

Although the Nuffield Council was of the view that women should have access to NIPT, it believed NIPT should be provided in such a way as 'to minimise the risks of significant harm where such risks exist'.[38] In particular, the Nuffield Council was influenced by

> the possible harms of extending the use of NIPT beyond testing fetuses for information that could have a direct bearing on the immediate or early health of fetuses and future children. This includes future uses of NIPT to test for a range of genetic traits, including adult onset conditions where this is 'for information only', carrier status, less significant medical conditions and impairments, and non-medical traits.[39]

In essence, the approach recommended by the Nuffield Council prioritises the welfare of the future child over the interests of the prospective parents. Significantly, this approach treats the interests of the future child and those of the prospective parents as distinct and in conflict rather than enmeshed and interrelated. The recommendation of the Nuffield Council is arguably based on a flawed distinction between wanting to know 'for information only'[40] and wanting to know to inform a decision about termination of pregnancy. However, motivations for prenatal testing are not always easily separated out

[36] *Prenatal Screening and Diagnostic Testing* (n 35) 6 recommendation 4, 7 recommendation 6. The term 'abnormalities' is specifically used by RANZCOG, although this term is not adopted by the authors in this article.

[37] Nuffield Council on Bioethics, *Non-Invasive Prenatal Testing: Ethical Issues* (Report, March 2017) 126.

[38] Ibid 123 [5.33].

[39] Ibid 124 [5.37].

[40] Zuzana Deans, Angus J Clarke and Ainsley J Newson, 'For Your Interest? The Ethical Acceptability of Using Non-Invasive Prenatal Testing to Test "Purely for Information"' (2015) 29(1) *Bioethics* 19, 24.

from one another and are equally legitimate reasons for testing on the continuum of prenatal decision-making.[41]

Although the RANZCOG guidelines make a general recommendation that pregnant women be provided with timely access to prenatal screening tests to enable them to make informed reproductive decisions, the guidelines do not specifically address the use of NIPT for adult-onset conditions. The RANZCOG guidelines emphasise the voluntary nature of prenatal testing and recommend that screening for fetal chromosomal and genetic conditions should only be undertaken as an informed decision by the pregnant woman.[42] They also clarify that availability of testing should not be contingent on the future choices the pregnant woman intends to make:

> The offer of screening should be made to all people irrespective of the clinician's perception of what their likely choices might be. It is essential that the woman is not deprived of the opportunity to find out about the health of her fetus. It is not ethical to presuppose a course of action prior to this information being provided.[43]

The RANZCOG guidelines therefore differ from the proposal of the Nuffield Council, which recommends restricting NIPT in cases where prospective parents are seeking testing 'for information only'.[44]

In the United States ('US'), the position statement of the National Society of Genetic Counselors ('NSGC') on prenatal testing specifically addresses prenatal genetic testing for adult-onset conditions.[45] Whilst the NSGC 'does not recommend prenatal genetic testing for known adult-onset conditions if pregnancy or childhood management will not be affected',[46] it acknowledged in 2016 that:

> parents could not be offered or denied testing based on a hypothetical discussion of their intentions. Answering hypothetical questions is not a genuine proxy for being confronted with real results. … Additionally, parents who say they either

[41] Roger Brownsword and Jeff Wale, 'Testing Times Ahead: Non-Invasive Prenatal Testing and the Kind of Community We Want to Be' (2018) 81(4) *Modern Law Review* 646, 658–9. See below Part III(B)(2)(b).

[42] *Prenatal Screening and Diagnostic Testing* (n 35) 6 recommendation 2. See also *Prenatal Screening for Fetal Genetic or Structural Conditions* (n 35) 5 recommendation 1.

[43] *Prenatal Screening and Diagnostic Testing* (n 35) 10 [3.1.2].

[44] See Nuffield Council on Bioethics (n 37) 84–6 [3.32]–[3.35]. It is worth noting that the RANZCOG guidelines were arguably not written with considerations about testing for adult-onset conditions in mind.

[45] *Prenatal Testing for Adult-Onset Conditions* (n 32).

[46] Ibid.

> will or will not terminate a pregnancy can change their minds. Genuine reproductive choice is contingent on the access to information.[47]

In contrast to the approach proposed by the Nuffield Council, the NSGC recommends that 'any conflict between the right of prospective parents to obtain information and the right of the future child should generally be resolved in favor of the parents'.[48]

The RANZCOG and NSGC guidelines focus on supporting fully informed reproductive decision-making by pregnant women and (where relevant) their partners. To this end, the guidelines prioritise access to information and promote non-directive counselling. Importantly, the guidelines do not make the provision of information contingent on how that information will be used. This is in line with doctors' common law duty of care to provide pregnant women sufficient information to make informed reproductive choices. This duty requires clinicians to provide patients with adequate information regarding available treatment or care, including its implications, material risks and limitations.[49] It also 'extends to the performance, interpretation and communication of test results'.[50] We discuss this common law duty in more detail in Part III.

C *Specific Guidelines on Prenatal Testing for Huntington's Disease*

International guidelines dealing specifically with predictive testing for Huntington's disease take a more restrictive approach to access to testing. In particular, they recommend against testing for adult-onset conditions unless it will impact on the woman's decision to proceed with the pregnancy. For example, the European Huntington Disease Network ('EHDN') recommends that

[47] Laura Hercher et al, 'Prenatal Testing for Adult-Onset Conditions: The Position of the National Society of Genetic Counselors' (2016) 25(6) *Journal of Genetic Counseling* 1139, 1141.

[48] Ibid 1144.

[49] Jeanne Snelling, Nikki Kerruish and Jessie Lenagh-Glue, 'Non-Invasive Prenatal Testing: The Problem with "Fast Cars"' (2016) 24(1) *Journal of Law and Medicine* 203, 215; *Rogers v Whitaker* (1992) 175 CLR 479, 490 (Mason CJ, Brennan, Dawson, Toohey and McHugh JJ) ('*Rogers*'). See also *Montgomery v Lanarkshire Health Board* [2015] AC 1430, 1461–4 [82]–[94] (Lords Kerr and Reed JJSC, Lords Neuberger PSC, Clarke, Wilson and Hodge JJSC agreeing) ('*Montgomery*').

[50] Snelling, Kerruish and Lenagh-Glue (n 49) 215. See also *Rogers* (n 49) 490 (Mason CJ, Brennan, Dawson, Toohey and McHugh JJ).

> [t]he couple requesting prenatal testing must be clearly informed that if they intend to complete the pregnancy whether the fetus is a carrier of the gene expansion or not, there is no valid reason for performing the test.[51]

In its comments on this recommendation, the EHDN states:

> This is in line with the recommendation not to test minors. The child's autonomy regarding his/her future right to decide whether or not to undergo a pre-symptomatic test is violated if pregnancy is continued in the case of an abnormal prenatal test result.[52]

The EHDN guidelines prioritise the future child's autonomy to decide whether or not to undergo pre-symptomatic testing over the prospective parents' interest in testing by limiting access to prenatal testing to cases where termination is intended using directive counselling. The guidelines do not, however, overrule a couple's entitlement to change their mind about termination after receiving a high-risk test result, thereby acknowledging and accommodating human ambivalence.[53]

D *Guidelines on Predictive Testing of Minors*

Guidelines relating to the predictive testing of minors are also relevant because some commentators contend that prenatal testing without any intention to terminate is 'no different from testing a minor'.[54] We argue in Part IV that there are relevant features that can morally and practically distinguish these two scenarios. However, in light of this view, we will address these related guidelines.

Although the EHDN guidelines state their recommendation against prenatal testing is in line with international guidelines on predictive testing of minors, this is not strictly true. Some international guidelines are restrictive and recommend against predictive genetic testing of minors for adult-onset conditions until such time as the child can make their own decision, or unless it is in the child's best interest. However, other international guidelines take a more nuanced approach.

[51] MacLeod et al (n 33) 227 recommendation 7.1.7.

[52] Ibid 227–8 commentary 7.1.7.

[53] Felicity Wadrup et al, 'A Case-Note Review of Continued Pregnancies Found to Be at a High Risk of Huntington's Disease: Considerations for Clinical Practice' (2019) 27(8) *European Journal of Human Genetics* 1215, 1222.

[54] Rony E Duncan, Bennett Foddy and Martin B Delatycki, 'Refusing to Provide a Prenatal Test: Can It Ever Be Ethical?' (2006) 333(7577) *BMJ* 1066, 1066.

The position statement of the Human Genetics Society of Australasia ('HGSA') states that a decision about predictive genetic testing 'should be one that is made in the best interests of the child, although it is noted that what constitutes "best interests" is the subject of ongoing ethical debate'.[55]

In relation to predictive testing for adult-onset conditions in children who do not yet have decision-making capacity, the HGSA recommends

> the default position should always be to postpone testing until the child or young person achieves this capacity. This position values preserving the child or young person's future autonomy to make their own choice whether they wish to know their genetic risk of the condition.[56]

The HGSA concedes, however, that it may be appropriate in 'rare circumstances' for a child or young person to undergo predictive testing for an adult-onset condition where 'it is the best option to support the wellbeing of the child or young person through constructive family dynamics'.[57] In the event of a dispute between a child and their parents, the HGSA states the health professional should act as an advocate for the child but recognises the child as part of a family and recommends that counselling focus on the family and the child together and separately.[58]

The NSGC in the US similarly encourages deferring predictive genetic testing of minors for adult-onset conditions when results will not impact childhood medical management or significantly benefit the child.[59]

The EHDN takes a more restrictive approach than the HGSA by recommending a minimum age of 18 years for testing for Huntington's disease.[60] According to the EHDN, '[t]he decision to take the test is the sole choice of the person concerned'.[61] It states categorically that requests from third parties, be they family or otherwise, should not be considered.[62]

By way of contrast, other international guidelines take a more contextualised approach to the interests of the minor and recognise the role of parental discretion and the interrelated nature of interests within families in a similar way to the new HGSA guidelines. For example, the American Society of Human Genetics ('ASHG') recommends parents 'defer predictive or pre-

[55] Vears et al (n 34) 188.

[56] Ibid 189.

[57] Ibid.

[58] Ibid 188.

[59] *Prenatal Testing for Adult-Onset Conditions* (n 32).

[60] MacLeod et al (n 33) 223 recommendation 2.1.

[61] Ibid 223 recommendation 2.

[62] Ibid.

dispositional testing for adult-onset conditions until adulthood'[63] but expressly acknowledges that 'there has been a general shift toward greater parental discretion in the face of clinical uncertainty about the best interests of the child'.[64] The American College of Medical Genetics and Genomics ('ACMG') similarly recommends deferring genetic testing for adult-onset conditions until adulthood but notes that 'predictive genetic testing may be appropriate in limited circumstances'.[65] The ACMG states that although the focus must be on the best interests of the child, parents 'may also consider the potential psychosocial benefits and harms to the child and the extended family'.[66] The ACMG recognises the context of the family within which a child exists, noting:

> Extending consideration beyond the child's medical best interest not only acknowledges the traditional deference given to parents about how they raise their children but also recognizes that the interest of a child is embedded in and dependent on the interests of the family unit.[67]

The ACMG concludes that, after careful genetic counselling, predictive testing of a minor may be ethically acceptable in some cases, 'to resolve disabling parental anxiety or to support life-planning decisions that parents sincerely believe to be in the child's best interest'.[68]

Although the HGSA, the ASHG and the ACMG recommend deferring genetic testing for adult-onset conditions until the child can make their own decision, or unless testing is in the child's best interest, their guidelines promote a more nuanced and contextualised approach to testing. In particular, they recognise the role of parental discretion in evaluating the best interests of the child and acknowledge the relational nature of the interests of a child within a family. Moreover, the ASHG and ACMG guidelines treat the personal utility of testing for the parents of a minor, such as seeking reassurance, as a potentially legitimate reason for predictive testing. It is also worth noting the ACMG guidelines on incidental findings in genome sequencing, which require incidental variants (including adult-onset conditions and susceptibility genes)

63 Jeffrey R Botkin et al, 'Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents' (2015) 97(1) *American Journal of Human Genetics* 6, 8.

64 Ibid 7.

65 Laine Friedman Ross et al, 'Technical Report: Ethical and Policy Issues in Genetic Testing and Screening of Children' (2013) 15(3) *Genetics in Medicine* 234, 238.

66 Ibid.

67 Ibid (citations omitted).

68 Ibid.

to be 'reported *regardless of the age of the patient*'.[69] It would be inconsistent to require such incidental findings to be reported yet prohibit prospective parents to test for adult-onset conditions.

Given the potential for NIPT to be more broadly used to detect adult-onset conditions in the near future, specific guidance on the use of NIPT is warranted. The guidelines discussed in this Part reflect a range of approaches to prenatal and genetic testing, which provide some helpful insights for how more specific guidelines on NIPT might be shaped. The next Part explores in detail the various interests at stake in relation to NIPT for detecting adult-onset conditions, with a view to developing a facilitative regulatory framework for NIPT that acknowledges the interrelated nature of the relevant interests.

III Relevant Interests and Legal Principles

A *Introduction*

The guidelines outlined above highlight the various interests that arise in relation to prenatal testing for adult-onset conditions. Broadly speaking, there is a tension between the interests of a woman or prospective parents in making genuinely informed reproductive decisions or understanding their pregnancy, and the interests of a future child in protection from potential psychosocial harm, preservation of future autonomy, and non-discrimination. In this Part, we explore the nature of these interests in more detail, the ways in which they intersect, and how the law has dealt with this tension in other contexts. We also discuss broader societal interests in limiting the scope of NIPT based on concerns about 'designer babies' and eugenics.

The interests of a pregnant woman (and her partner) in reproductive choice are generally prioritised in general guidelines on prenatal screening and under common law.[70] There is a strong argument for any regulation around NIPT to support women and relevant partners in making informed decisions about NIPT, particularly in difficult cases. However, using NIPT to find out sensitive genetic information about a fetus has significant (and potentially harmful) implications for the fetus (if it is terminated) or the future child (if the pregnancy continues). Whilst the fetus arguably has some moral (if not independent legal) status warranting consideration, the law generally stops

[69] Robert C Green et al, 'ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing' (2013) 15(7) *Genetics in Medicine* 565, 569 (emphasis added). See also ACMG, 'Practice Guidelines: Incidental Findings in Clinical Genomics: A Clarification' (2013) 15(8) *Genetics in Medicine* 664 <https://www.hgsa.org.au/documents/item/582>.

[70] See above Part II(B).

short of protecting the interests of the fetus against those of the pregnant woman.[71] By way of contrast, the interests of the future child are more carefully protected. As outlined in Part II, specific guidelines on prenatal testing for Huntington's disease prioritise the future child's autonomy over the parents' interest in testing in certain cases.[72] In this Part, we explore in detail the interests of prospective parents, the future child, and broader society before proposing a facilitative framework for accommodating the relevant and often interrelated interests in Part IV.

It is worth noting that a woman may access NIPT in various different contexts, including through public or privately funded healthcare arrangements via a medical professional, or even possibly through direct-to-consumer testing ('DTC') in the future. Although this article focuses on regulating access to NIPT in a medical setting, it is important to acknowledge the limitations of regulation in this context given the scope and increasing prevalence of DTC genetic testing in recent years. There are also different considerations when dealing with private and public healthcare contexts. As NIPT is increasingly integrated into public healthcare systems, questions of resource allocation may also come into play when assessing whether these tests should be provided. We recognise that these are likely considerations, but believe they apply in general to all diagnostic tests and techniques, and thus we will not address them at length here.

B *Interests of Prospective Parents*

Prospective parents, in particular pregnant women, have an interest in accessing information that may enhance their reproductive choices. As explained in Part I, NIPT is a relatively safe and simple test that potentially enhances reproductive choice by promoting informed decision-making. The interest of prospective parents in seeking NIPT invokes two key principles that are recognised under common law in the UK and Australia — the principle of bodily integrity and the principle of reproductive autonomy.[73] The potential for NIPT to harm the future child raises a broader discussion about the role of law

[71] The moral status of a fetus is a contentious subject beyond the scope of this article. For a detailed discussion on the legal and moral status of the fetus in the context of harm in utero, see Dominic Wilkinson et al, 'Protecting Future Children from In-Utero Harm' (2016) 30(6) *Bioethics* 425.

[72] For example, where testing is carried out for 'information only', although, as noted above in Part II(B), this assumption is questionable.

[73] *Secretary, Department of Health and Community Services v JWB* (1992) 175 CLR 218, 233–4 (Mason CJ, Dawson, Toohey and Gaudron JJ) ('*JWB*'). The first principle focuses on the pregnant woman whereas the second principle applies more broadly to prospective parents.

in balancing the interests of prospective parents with those of the future child. This discussion has played out in the context of refusal-of-treatment cases, harmful antenatal behaviour cases, and prenatal injury cases, which offer some additional insight into the interrelationship between the interests of the pregnant woman and the future child.

Courts and policymakers have struggled to balance the interests of prospective parents, particularly pregnant women, with those of the fetus or future child. Domestic courts typically frame these interests in terms of 'rights' of relevant individuals.[74] Whilst a 'substantive model of children's rights' grounded in international law can provide a relevant framework for evaluating the complex interests of a child, courts have not consistently applied a substantive rights analysis in their judgments.[75] As a consequence, the language of rights can sometimes be unhelpful and potentially polarising in debates about the welfare of the child in certain contexts.[76] We choose instead to use the language of 'interests' to frame our discussion within a relational approach that squarely acknowledges the interconnectedness of the various interests at stake.

1 *Principle of Bodily Integrity*

The law generally allows pregnant women to make their own medical treatment decisions, even where this may harm the future child. The requirement for consent to medical treatment is underpinned by the legal right to self-determination, which is based on the principle of bodily integrity.[77] A legally valid consent to treatment operates as a defence to assault in criminal law and trespass to person under civil law.[78] UK courts have applied the principle of bodily integrity in refusal-of-treatment cases to prioritise the interests of a pregnant woman over any potentially conflicting interests of the future child.[79] The principle of bodily integrity arguably extends to all medical decisions made

[74] See, eg, *Re T (Adult: Refusal of Treatment)* [1993] Fam 95, 102 (Lord Donaldson MR) ('*Re T*').

[75] John Tobin, 'Judging the Judges: Are They Adopting the Rights Approach in Matters Involving Children?' (2009) 33(2) *Melbourne University Law Review* 579, 582, 593–609.

[76] For a discussion of how the language of rights has been applied in the context of donor conception, see Michelle Taylor-Sands, 'Removing Donor Anonymity: What Does It Achieve?' (2018) 41(2) *University of New South Wales Law Journal* 555. See also John Tobin, 'Donor-Conceived Individuals and Access to Information about Their Genetic Origins: The Relevance and Role of Rights' (2012) 19(4) *Journal of Law and Medicine* 742.

[77] *Schloendorff v Society of New York Hospital*, 211 NY 125, 129–30 (Cardozo J) (1914).

[78] See, eg, ibid; *Rogers* (n 49) 490 (Mason CJ, Brennan, Dawson, Toohey and McHugh JJ) citing *Chatterton v Gerson* [1981] 1 QB 432, 443 (Bristow J); *JWB* (n 73) 234 (Mason CJ, Dawson, Toohey and Gaudron JJ).

[79] See, eg, *Re MB (Medical Treatment)* [1997] 2 FLR 426, 444 (Butler-Sloss LJ) ('*Re MB*').

by pregnant women, including decisions to pursue prenatal testing during the pregnancy. As mentioned in Part I, NIPT involves a blood test on the pregnant woman to obtain genetic information about the fetus, which is contained in the maternal bloodstream.

Decisions by pregnant women about medical treatment and interventions potentially carry implications for the fetus or future child.[80] Refusal-of-treatment cases provide some insight into how the law has managed the conflict between the interests of a pregnant woman and the future child. Although a decision to refuse treatment can be distinguished from a decision to seek an arguably 'non-essential' medical intervention or test such as NIPT, there is a similar tension in both scenarios between the interests of the pregnant woman and the future child.[81] The following analysis is not intended as a legal justification for allowing broader access to NIPT but is used to show how the law has responded to the tension between these potentially conflicting interests based on the principle of bodily integrity.

In the case of *Re MB (Medical Treatment)*, the Court of Appeal of England and Wales highlighted a pregnant woman's right to self-determination and privacy in relation to consent to (and refusal of) medical treatment.[82] In that case, the Court of Appeal held:

[80] For example, medications for a variety of conditions, including high blood pressure, mental illness, thyroid conditions and acne may potentially have harmful effects on a developing fetus: see, eg, 'Pregnancy: Medication, Drugs and Alcohol', *Better Health Channel* (Web Page, 31 May 2012) <https://www.betterhealth.vic.gov.au/health/healthyliving/pregnancy-medication-drugs-and-alcohol>, archived at <https://perma.cc/JP4F-4XBD>; Carrie Armstrong, 'ACOG Guidelines on Psychiatric Medication Use during Pregnancy and Lactation' (2007) 78(6) *American Family Physician* 772. Certain medical interventions also carry specific risks of harm for the fetus or future child. For example, amniocentesis carries a low risk of miscarriage, needle injury, or transmission of infection to the fetus: 'Amniocentesis', *Mayo Clinic* (Web Page, 12 November 2020) <https://www.mayoclinic.org/tests-procedures/amniocentesis/about/pac-20392914>, archived at <https://perma.cc/8KX4-JB56>.

[81] Although there is no positive duty on a medical practitioner to facilitate each and every medical intervention sought by their patients, the discussion below in Part III(B)(2)(b) suggests that the duty to inform patients potentially extends to NIPT where there is either some clinical or personal utility to such testing.

[82] *Re MB* (n 79). The concept of privacy in this context has been criticised by feminist theorists as problematic insofar as it frames debates around reproductive choice in terms of opposing rights of fetus and pregnant woman: Jennifer Nedelsky, 'The Practical Possibilities of Feminist Theory' (1993) 87(4) *Northwestern University Law Review* 1286, 1288. See also Regina Graycar and Jenny Morgan, *The Hidden Gender of Law* (Federation Press, 2nd ed, 2002) 208–9. An alternative, more nuanced, approach is to view the fetus as inextricably linked with the pregnant woman.

> A competent woman who has the capacity to decide may … choose not to have medical intervention, even though the consequence may be the death or serious handicap of the child she bears, or her own death.[83]

Lady Justice Butler-Sloss recognised the ethical concern about potential harm to the future child but upheld the woman's right to self-determination, stating 'this is not a court of morals'.[84] Interestingly, the Court of Appeal ultimately held that the woman had a temporary loss of capacity and approved the provision of treatment on the basis it was in the pregnant woman's best interests.[85] Although there is no judicial authority in Australia on refusal of treatment by a pregnant woman, Australian courts are likely to follow UK authority and resolve such disputes on the basis of the patient's capacity.[86]

Courts have struggled with where the line should be drawn in refusal-of-treatment cases involving pregnant women. In an earlier UK case, the High Court of England and Wales made a declaration that a caesarean section and any consequential treatment could be lawfully performed if it was in the interests of the patient and her unborn child, despite the patient's refusal to give consent.[87] The Court of Appeal later upheld the right of a pregnant woman to refuse medical treatment even where this might result in the death of the woman and her unborn child.[88] In Australia, a New South Wales judge suggested in 2009 that a court might override a pregnant woman's refusal of treatment to protect a fetus 'if the treatment is necessary to save the life of a viable unborn child'.[89] Such a decision would effectively impose what some have

[83] *Re MB* (n 79) 436–7 (Butler-Sloss LJ).

[84] Ibid 440.

[85] Ibid 438–9 (Butler-Sloss LJ). The pregnant woman had consented to a caesarean section and was aware of medical need for it but suffered needle phobia and refused the needle for an anaesthetic: at 428–30.

[86] Bernadette Richards, 'General Principles of Consent to Medical Treatment' in Ben White, Fiona McDonald and Lindy Willmott (eds), *Health Law in Australia* (Thomson Reuters, 3rd ed, 2018) 135, 152 [5.100]. It could be argued that, by closely scrutinising a pregnant woman's capacity, courts are stretching the notion of incapacity to achieve the results they consider ethical: see also Belinda Bennett, 'Pregnant Women and the Duty to Rescue: A Feminist Response to the Fetal Rights Debate' (1991) 9(1) *Law in Context* 70, 71, 79.

[87] *Re S (Adult: Refusal of Medical Treatment)* [1992] 4 All ER 671, 672 (Sir Stephen Brown P). It is noteworthy that in this case, in the absence of treatment, the life of the pregnant woman was also at risk (in addition to the risks of harm to the fetus/future child). See also *Re T* (n 74) 102, 115–16 (Lord Donaldson MR).

[88] *St George's Healthcare NHS Trust v S* [1999] Fam 26, 50 (Judge LJ for the Court).

[89] *Hunter and New England Area Health Service v A* (2009) 74 NSWLR 88, 97 [40] (McDougall J). See discussion in Wilkinson et al (n 71) 430.

described as a moral 'duty to rescue'.[90] Even if such a moral duty exists, 'the presence of an ethical imperative is by no means a *sufficient* justification for the imposition of a legal obligation'.[91]

The principle of bodily integrity lends support to allowing access to NIPT to assist in reproductive decision-making, even where there is potential conflict with the interests of the future child. The next section discusses the related principle of reproductive autonomy, including the legal duty to inform and the clinical and personal utility of prenatal testing.

2 *Principle of Reproductive Autonomy*

The second (and related) principle which supports the interests of prospective parents seeking NIPT is the principle of reproductive autonomy. This principle applies to both pregnant women and their partners as prospective parents. The principle of reproductive autonomy provides that prospective parents should be able to make informed reproductive choices[92] and underpins the common law duty to inform pregnant women about screening and treatment options during pregnancy where a failure to inform may lead to unacceptable harm.[93] Information about a pregnancy has both clinical and personal utility for prospective parents.

(a) Duty to Inform Pregnant Women about Screening and Treatment Options

In relation to medical treatment in general, Australian law adopts a patient-centred approach to the duty to inform under the tort of negligence. In the leading case of *Rogers v Whitaker*, the High Court held that a health

[90] John A Robertson, 'Procreative Liberty and the Control of Conception, Pregnancy, and Childbirth' (1983) 69(3) *Virginia Law Review* 405, 456. See discussion in Graycar and Morgan (n 82) 242–3. See also Bennett (n 86) 83–5. This idea was more recently developed by Wilkinson et al (n 71) to support certain 'soft' prenatal interventions to protect the fetus from harm: at 430.

[91] Anne Morris and Susan Nott, 'The Law's Engagement with Pregnancy' in Jo Bridgeman and Susan Millns (eds), *Law and Body Politics: Regulating the Female Body* (Dartmouth, 1995) 53, 60 (emphasis in original). See also Graycar and Morgan (n 82) 243.

[92] For an in-depth philosophical analysis of the principle of reproductive autonomy which critiques the standard negative model of reproductive freedom based on noninterference and proposes a form of positive freedom or 'self-making', see Catherine Mills, 'Reproductive Autonomy as Self-Making: Procreative Liberty and the Practice of Ethical Subjectivity' (2013) 38(6) *Journal of Medicine and Philosophy* 639.

[93] See, eg, *Cattanach v Melchior* (2003) 215 CLR 1, 31 [66] (McHugh and Gummow JJ) 71 [188]–[191] (Hayne J); *Waller v James* (2015) 90 NSWLR 634, 658–9 [129]–[132] (Beazley P); *Rogers* (n 49) 490 (Mason CJ, Brennan, Dawson, Toohey and McHugh JJ); *Montgomery* (n 49) 1464–5 [94] (Lords Kerr and Reed JJSC, Lords Neuberger PSC, Clarke, Wilson and Hodge JJSC agreeing).

practitioner must disclose all *material risks* involved in treatment.[94] Material risks are those that a reasonable patient in the patient's position would regard as significant or that the doctor should reasonably be aware that the particular patient would regard as significant.[95] This patient-centred approach to the duty to inform was more recently adopted in the UK in the 2015 landmark case of *Montgomery v Lanarkshire Health Board* ('*Montgomery*').[96] In that case, the UK Supreme Court found the obstetrician was negligent in failing to inform the patient of the risk of shoulder dystocia and the option of a caesarean birth.[97] Although the Supreme Court did not specifically mention NIPT or prenatal testing, *Montgomery* arguably reinforces the importance of taking a pregnant woman's autonomy, choices and consent seriously in reproductive settings,[98] and doctors may now be legally obliged to inform women about particular prenatal tests.[99]

As with all prenatal testing, the potential for routinisation of testing and increased societal pressure to test using NIPT requires careful thought about the nature of communication and counselling around testing.[100] In the event

[94] *Rogers* (n 49) 490 (Mason CJ, Brennan, Dawson, Toohey and McHugh JJ).

[95] Ibid. In this case, the surgeon failed to warn his patient of the risk of 'sympathetic ophthalmia' in her good left eye, for which there was a 1 in 14,000 chance arising out surgery to her injured right eye: at 482. Following her surgery, the patient developed this condition and ultimately lost all sight in her left eye resulting in almost total blindness.

[96] *Montgomery* (n 49). This case involved an obstetrician who failed to advise her patient about the risk of shoulder dystocia inherent in childbirth and the option of having a caesarean birth: at 1442 [2] (Lords Kerr and Reed JJSC, Lords Neuberger PSC, Clarke, Wilson and Hodge JJSC agreeing). The child born as a result of vaginal delivery experienced complications owing to shoulder dystocia, resulting in hypoxic insult with consequent cerebral palsy: at 1446 [22]. The patient, who had diabetes and was of small stature, had previously asked if the baby's size might be a potential problem: at 1443 [7], 1445 [17].

[97] Ibid 1464–5 [94].

[98] Brownsword and Wale (n 41) 650. However, Roger Brownsword and Jeff Wale acknowledge these principles need to be unpacked.

[99] Nuffield Council on Bioethics (n 37) 22 [1.45]–[1.46]. This raises a logistical issue, in relation to whole-genome sequencing, that the sheer amount of information available makes this an almost impossible task. It is worth noting alternative models of informed consent that have been developed in relation to genomic testing, including 'binning': Angela R Bradbury et al, 'Development of a Tiered and Binned Genetic Counseling Model for Informed Consent in the Era of Multiplex Testing for Cancer Susceptibility' (2015) 17(6) *Genetics in Medicine* 485, 487.

[100] For detailed discussions of these concerns about prenatal testing, see de Jong et al (n 3); SE Kelly and HR Farrimond, 'Non-Invasive Prenatal Genetic Testing: A Study of Public Attitudes' (2012) 15(2) *Public Health Genomics* 73; Ananda van den Heuvel et al, 'Will the Introduction of Non-Invasive Prenatal Diagnostic Testing Erode Informed Choices? An Experimental Study of Health Care Professionals' (2010) 78(1) *Patient Education and Counseling* 24, cited in Jade Bennett, Lyn Chitty and Celine Lewis, 'Non-Invasive Prenatal Diagnosis for BRCA Mutations: A Qualitative Pilot Study of Health Professionals' Views' (2016) 25(1) *Journal of Genetic Counseling* 198, 204.

that the scope for NIPT is greatly expanded (such as through whole-genome sequencing), there is a challenge as to what extent healthcare professionals are required to inform patients about the possible outcomes of a prenatal test, and how they obtain consent. For example, the amount of information that can be generated can be enormous and complex, with many variants having unknown risks and benefits, and there is a lack of consensus about which model of consent to use in the context of genomic sequencing.[101] Using NIPT to produce this information in a prenatal setting may result in 'information-overload', and it is a recognised challenge to be able to balance this with continuing to facilitate meaningfully informed choice.[102] These are challenges that will need to be addressed in the genetic counselling context.

(b) Clinical and Personal Utility of NIPT

NIPT has the potential to enhance reproductive choice by providing prospective parents with information that has both clinical and personal utility. Information about genetic conditions of the fetus has important clinical utility as it may be relevant to decisions that may impact on whether or not prospective parents continue with a pregnancy or consider termination. NIPT has several benefits over other forms of prenatal testing as it is easily performed and carries no risk of miscarriage. As NIPT is performed early in the pregnancy, it also allows parents more time to make decisions about invasive testing and termination. In addition to informing decisions about the current pregnancy, NIPT may also support parents to prepare for impending parenthood and make life-planning decisions to support the welfare of their future child and other family members. In cases where parents may not know of their carrier status, testing for adult-onset conditions as part of a suite of tests may reveal genetic predisposition to certain genetic conditions otherwise unknown. This information may impact on whether, when, and under what conditions the prospective parents will attempt any future pregnancies.[103] The NSGC expressly acknowledges the importance of prenatal testing in promoting reproductive choice in a variety of ways, including 'to prepare for the birth

[101] Shannon Rego et al, 'Informed Consent in the Genomics Era' (2020) 10(8) *Cold Spring Harbor Perspectives in Medicine* a036582:1–13.

[102] Wybo Dondorp et al, 'Non-Invasive Prenatal Testing for Aneuploidy and Beyond: Challenges of Responsible Innovation in Prenatal Screening' (2015) 23(11) *European Journal of Human Genetics* 1438, 1444.

[103] See Duncan, Foddy and Delatycki (n 54) 1067. For example, couples may choose to use pre-implantation genetic diagnosis ('PGD') for future pregnancies or delay another pregnancy until they are financially secure to cope with an affected child: see Hanane Bouchghoul et al, 'Prenatal Testing in Huntington Disease: After the Test, Choices Recommence' (2016) 24(11) *European Journal of Human Genetics* 1535, 1539.

and future needs of [the] offspring, to make an adoption plan, or to end a pregnancy'.[104]

Beyond its clinical utility in reproductive decision-making, NIPT also has significant *personal utility* for prospective parents. Research suggests that women who use NIPT want to know this information without considering termination of pregnancy. For example, one study revealed that 38% of women want to know about non-preventable adult-onset conditions and only 3% indicated they would consider terminating their pregnancy on this basis.[105] Several studies revealed that the need for 'reassurance', 'security', and 'peace of mind' emerge as primary motivators for individuals who seek NIPT for conditions such as trisomy 21.[106] Similar assurance may be obtained from testing for other kinds of conditions, including adult-onset conditions. One study examining health professionals' opinions on NIPT for *BRCA1* mutations found that they perceived the test to be particularly useful for women with exceptional psychosocial trauma due to breast cancer (for example, through the deaths of affected family members).[107] Reassurance was highlighted as a significant benefit of undergoing the test. The ACMG has acknowledged the importance of parental reassurance in testing children for adult-onset conditions where there is a known family history.[108] Prospective parents with no known history of a particular condition may similarly find reassurance from NIPT for adult-onset conditions, although it may not be as significant as it is for those with a known history.[109]

[104] National Society of Genetic Counselors, *Reproductive Freedom* (Position Statement, 12 April 2018) <https://www.nsgc.org/Policy-Research-and-Publications/Position-Statements/Position-Statements/Post/reproductive-freedom>, archived at <https://perma.cc/SZA5-WS9U>. See also Hercher et al (n 47) 1140.

[105] Hilary Bowman-Smart et al, '"Is It Better Not to Know Certain Things?": Views of Women Who Have Undergone Non-Invasive Prenatal Testing on Its Possible Future Applications' (2019) 45(4) *Journal of Medical Ethics* 231, 234, 236 ('Is It Better Not to Know Certain Things?').

[106] Hilary Bowman-Smart et al, '"Small Cost to Pay for Peace of Mind": Women's Experiences with Non-Invasive Prenatal Testing' (2019) 59(5) *Australian and New Zealand Journal of Obstetrics and Gynaecology* 649, 650; Huso Yi et al, 'Motivations for Undertaking DNA Sequencing-Based Non-Invasive Prenatal Testing for Fetal Aneuploidy: A Qualitative Study with Early Adopter Patients in Hong Kong' (2013) 8(11) *PLOS ONE* e81794:1–11, 5. See also Bennett, Chitty and Lewis (n 100) 203.

[107] Bennett, Chitty and Lewis (n 100) 203.

[108] Ross et al (n 65) 238. For a more detailed analysis of the ACMG's recommendations, see above Part II(D).

[109] Women may not feel the need to seek reassurance until *after* they find out that a particular test exists. It could therefore be argued that the existence of the test generates its own justification. This highlights the importance of careful pre-test counselling.

Some commentators have argued that if a pregnant woman does not intend to terminate, it is unacceptable for her to test 'purely for information'.[110] However, as noted above, there may be benefits even if the pregnancy is not terminated. In addition, making a decision about termination of pregnancy in the abstract can be very different to making choices when faced with real results. A survey of Australian women who had undergone NIPT found that 43% indicated they would be willing to consider termination of pregnancy for trisomy 21.[111] However, the rate of termination of pregnancy for trisomy 21 in Australia is much higher, with one study from Victoria revealing that only 5% of pregnancies with a prenatal diagnosis of trisomy 21 were associated with a live birth.[112] This discrepancy may be explained by research that suggests that willingness to consider a termination of pregnancy may significantly increase with the certainty of the diagnosis.[113] The NSGC emphasised the importance of genuine choice based on real information in their position statement on prenatal testing for adult-onset conditions.[114] However, it is important to note that women may face additional pressure (perhaps familial, medical, or societal) to terminate once they have obtained the information about the genetic status of their child. Parents may be judged for choosing to continue the pregnancy, with the assumption being that they must not have 'found out in time'.[115] Thus, part of the tendency towards termination of pregnancy upon receipt of results may be due to external pressure on the woman or parent(s).

3 *Role of Law in Balancing the Interests of the Pregnant Woman and the Future Child*

The discussion above relates to medical decision-making by prospective parents and outlines how the law has responded to potential conflicts between the interests of parents and those of the future child. It is also worth briefly exploring how the law has dealt with non-medical decisions by pregnant women as this sheds light on the interrelatedness of the interests of a pregnant

[110] Deans, Clarke and Newson (n 40) 19.

[111] Bowman-Smart et al, 'Is It Better Not to Know Certain Things?' (n 105) 236.

[112] Veronica R Collins et al, 'Is Down Syndrome a Disappearing Birth Defect?' (2008) 152(1) *Journal of Pediatrics* 20, 23.

[113] Hyunkyung Choi, Marcia Van Riper and Suzanne Thoyre, 'Decision Making following a Prenatal Diagnosis of Down Syndrome: An Integrative Review' (2012) 57(2) *Journal of Midwifery & Women's Health* 156, 156. This raises an important question about the impact of using NIPT for broad screening purposes: see below Part III(D).

[114] Hercher et al (n 47) 1141.

[115] Chriselle L Hickerton et al, '"*Did You Find That Out in Time*?": New Life Trajectories of Parents Who Choose to Continue a Pregnancy Where a Genetic Disorder Is Diagnosed or Likely' (2012) 158A(2) *American Journal of Medical Genetics Part A* 373.

woman and the future child. The law's approach to potentially harmful antenatal behaviour and prenatal injury highlights the limitations of an overly restrictive or punitive approach to the activities of a pregnant woman.

(a) Antenatal Behaviour

Beyond prenatal testing, there are other antenatal activities[116] that may result in harm to the future child, including smoking, alcohol consumption and illicit drug use.[117] While there is some precedent in the US where the law has intervened to restrict the behaviour of pregnant women based on the welfare of the future child,[118] this has not occurred in Australia. Australia has instead focused on developing policies aimed at educating, supporting and counselling women to make informed decisions during pregnancy and minimising harmful antenatal behaviour.[119]

The scope for restricting antenatal behaviour that may cause harm to the future child was explored in detail in 1995 by a report prepared by John Seymour on behalf of the Australian medical profession ('*Seymour Report*').[120] The report concluded:

> There is no place for legal intervention designed to impose controls on the behaviour of a pregnant woman when this behaviour is potentially harmful to the fetus. … At best, the invocation of the law is ineffective and at worst counter-productive.[121]

[116] The term 'antenatal' is used here as this is the language used in these scenarios. We use the terms 'prenatal' and 'antenatal' interchangeably to describe the period after conception and before birth.

[117] See, eg, Anni Lehikoinen et al, 'The Effect of Maternal Alcohol and Drug Abuse on First Trimester Screening Analytes: A Retrospective Cohort Study' (2020) 20(1) *BMC Pregnancy and Childbirth* 562; Department of Health (Cth), *National Fetal Alcohol Spectrum Disorder (FASD) Strategic Action Plan 2018–2028* (Plan, November 2018) 8 <https://www.health.gov.au/sites/default/files/national-fasd-strategic-action-plan-2018-2028.pdf> ('*FASD Plan*'); 'Tobacco, Alcohol, Drugs, and Pregnancy', *American College of Obstetricians and Gynecologists* (Web Page, June 2020) <https://www.acog.org/womens-health/faqs/tobacco-alcohol-drugs-and-pregnancy>, archived at <https://perma.cc/X4QF-NQUF>.

[118] For a discussion of US case law, see John Seymour, *Fetal Welfare and the Law* (Australian Medical Association Inquiry Report, 1995) 116–17. See also Wilkinson et al (n 71) 430.

[119] See, eg, *FASD Plan* (n 117) which has the priority objective of '[i]ncreas[ing] community knowledge and awareness about the harms and consequences of drinking during pregnancy or when planning a pregnancy': at 19.

[120] Seymour (n 118).

[121] Ibid 135. There is arguably a difference between regulating behaviours that may directly harm the future child using coercive measures and restricting access to genetic information that may

The *Seymour Report* reinforced that pregnant women should be free to decide how to live their lives. However, when making decisions, it stressed the importance of providing pregnant women with access to information about the harm their behaviour might cause to the fetus, noting: 'It is on the provision of such information that a society which is genuinely concerned about ensuring the birth of healthy children should concentrate.'[122] Instead of using the law to control antenatal behaviour, the *Seymour Report* proposed education campaigns to alert women to the risks of harm to the fetus.[123]

(b) Prenatal Injury

The courts have similarly grappled with how the interests of a future child should be conceptualised and balanced against the current interests of a pregnant woman in prenatal injury cases. Although a fetus is not a legal person, the law recognises it has certain interests that crystallise at birth. Australian courts have found that a duty of care is owed to a fetus, although an action cannot be brought until the child is born. For example, in 1972 the Supreme Court of Victoria held that a child can sue a third party via a representative in the tort of negligence for prenatal injury.[124] In practice, a claim in negligence against the child's mother is unlikely to be in the best interest of the child as it would reduce the funds available to the family and create tension between family members.[125] Moreover, it is generally the mother who ultimately is the primary caregiver of the child once they are born.[126] Any punitive action against the mother would therefore be counterproductive. The law has made a limited exception to enable a child to sue their mother for prenatal injury caused by the mother's negligent driving of a car where compensation would be paid by an insurer.[127] There is also authority in the UK that a health worker owes a duty of care to the fetus.[128]

indirectly harm a future child. However, the underlying policy considerations around personal autonomy, self-determination and privacy apply in both scenarios.

122 Ibid.

123 Ibid.

124 *Watt v Rama* [1972] VR 353, 355, 361 (Winneke CJ and Pape J, Gillard J agreeing at 361). This case involved an infant who was injured as a result of a motor vehicle accident between her pregnant mother and another driver: at 354 (Winneke CJ and Pape J).

125 Wilkinson et al (n 71) 431. See also Seymour (n 118) 193.

126 Seymour (n 118) 196.

127 *Lynch v Lynch* (1991) 25 NSWLR 411, 415–16 (Clarke JA, Gleeson CJ agreeing at 413, Hope AJA agreeing at 421).

128 *Burton v Islington Health Authority* [1993] QB 204, 223, 227 (Dillon LJ, Balcombe LJ agreeing at 232, Leggatt LJ agreeing at 233).

In addition to practical considerations, there are fundamental questions about applying the tort of negligence to intra-familial relations. Seymour points out that this would involve the courts taking on 'the role of setting objective standards of care for parenting'.[129] The High Court of Australia has held that a parent's moral duty to feed, clothe, maintain, educate, and generally care for a child is not enforceable in tort: 'the moral duties of conscientious parenthood do not as such provide the child with any cause of action when they are not, or badly, performed or neglected'.[130]

Although parents have a moral duty to ensure the basic needs of their children are met, 'beyond this, parental decision-making is largely discretionary' subject to a threshold level of harm.[131] Feminist legal scholars have, however, highlighted the dangers of treating family as a 'private' sphere beyond the reach of direct legal intervention, namely that this does not adequately address violence against women in their homes.[132]

As particularly vulnerable members of a family unit, children require special protection under the law. Instead of facilitating civil action by children against parents, the law deals more appropriately with parental neglect, violence and abuse through its child welfare or criminal jurisdictions. The arguments against allowing civil action by children against their parents are even stronger in the context of antenatal behaviour of a pregnant woman, particularly if Isabel Karpin's 'Not-One-But-Not-Two' model of the maternal–fetal relationship is adopted.[133] The special and dependent nature of the relationship between a pregnant woman and her fetus precludes the fetus having rights 'which can be enforced at the expense of the woman'.[134] However, recognising that children are vulnerable and dependent, courts exercising welfare jurisdiction (once a child is born) should arguably take account of harmful antenatal behaviour in extreme cases in determining if some form of intervention is needed.[135] Any regulatory framework for NIPT should similarly explore a threshold level of protection for the future child.

[129] Seymour (n 118) 193.

[130] *Hahn v Conley* (1971) 126 CLR 276, 283 (Barwick CJ).

[131] Taylor-Sands, *Saviour Siblings* (n 29) 97.

[132] A seminal discussion of the feminist critique of the public–private dichotomy and its limitations can be found in Graycar and Morgan (n 82) ch 2.

[133] Isabel Karpin, 'Legislating the Female Body: Reproductive Technology and the Reconstructed Woman' (1992) 3(1) *Columbia Journal of Gender and Law* 325, 329. See also Elselijn Kingma, 'Were You a Part of Your Mother?' (2019) 128(511) *Mind* 609, which endorses the 'parthood view' (the fetus is considered a part of the gestating parent) over the 'containment view' (the fetus is merely contained within the gestating parent): at 609–10.

[134] Seymour (n 118) 194.

[135] Ibid 135.

C *Welfare and Interests of the Future Child*

Concerns raised about the impact of NIPT on the welfare of the future child generally fall within three categories: negative psychosocial outcomes; violating the autonomy of the future child; and breach of privacy or confidentiality leading to discrimination. These concerns have been raised in relation to predictive testing of minors, leading to clinical guidelines recommending testing be deferred until the child has capacity to consent or unless testing is in the child's best interests.[136] In this section, we briefly outline these three categories of concern, current empirical evidence in relation to these concerns, and the law's response to date.

The first concern is that predictive testing for adult-onset conditions prenatally can lead to negative psychosocial outcomes. It is argued the child may feel distress, anxiety, or decreased self-esteem as a consequence of knowing their genetic status.[137] Other concerns relate to the potential negative impact on the parent–child relationship, in particular that the child may suffer 'vulnerable child syndrome' whereby the parents may become unduly overprotective or restrictive of a child deemed 'at risk'.[138] Qualitative research suggests that, in the event of an abnormal screening result, parents may medicalise their child and implement onerous health regimes (eg diets).[139] Although there is little data in the context of prenatal testing, it is helpful to examine empirical evidence addressing the issue of predictive testing in minors. Significantly, a 2016 meta-review of existing studies found little conclusive evidence for negative psychological harms from predictive testing in minors.[140] It is important to note, however, that there was less data for non-treatable adult-onset conditions and few long-term studies.[141] In the absence of longitudinal data, concerns

[136] Ross et al (n 65) 238; Botkin et al (n 63) 6–7, citing The American Society of Human Genetics Board of Directors and The American College of Medical Genetics Board of Directors, 'Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents' (1995) 57(5) *American Journal of Human Genetics* 1233.

[137] Cara Mand et al, 'Predictive Genetic Testing in Minors for Late-Onset Conditions: A Chronological and Analytical Review of the Ethical Arguments' (2012) 38(9) *Journal of Medical Ethics* 519, 522 figure 1 ('Predictive Genetic Testing in Minors').

[138] Morris Green and Albert J Solnit, 'Reactions to the Threatened Loss of a Child: A Vulnerable Child Syndrome' (1964) 34(1) *Pediatrics* 58, 58, 62–3. The American Society of Human Genetics Board of Directors and The American College of Medical Genetics Board of Directors (n 136) prepared a detailed list of potential harms in 1995: at 1235 table 1. See also Hercher et al (n 47) 1142.

[139] Rachel Grob, 'Qualitative Research on Expanded Prenatal and Newborn Screening: Robust but Marginalized' (2019) 49(S1) *Hastings Center Report* S72, S74–5.

[140] Claire E Wakefield et al, 'The Psychological Impact of Genetic Information on Children: A Systematic Review' (2016) 18(8) *Genetics in Medicine* 755, 755.

[141] Ibid.

about negative psychosocial outcomes are largely theoretical at this stage but warrant continued monitoring. It is also possible children may receive psychological benefits from testing, even if they are gene-positive for a non-treatable condition such as Huntington's disease.[142]

The second concern relates to the future child's autonomy. Testing a fetus for an adult-onset condition would reveal the genetic status of the future child without their consent. According to Bernard Dickens, disclosure to children about their genetic status may inhibit their ability to flourish and express independent personalities.[143] Some frame this concern as infringing what Joel Feinberg describes as the child's 'right to an open future'.[144] According to Feinberg, a child's future interests or options should be kept open until that child has the capacity for self-determination. The notion of a child's right to an open future has, however, been criticised as 'self-contradictory' and 'problematic'[145] as 'parental decisions inevitably shape the course of their children's lives'.[146] The emphasis on a child's future autonomy has also resulted in an individualistic, child-centred approach to genetic testing of minors[147] and neglects the interconnectedness of the child's interests with those of their family.[148] Moreover, some argue that predictive testing in children actually has the potential to *enhance* their autonomy, because it allows for children to make more informed decisions about their life plans as they mature.[149]

The third concern is that a child's privacy or confidentiality is breached if their genetic information is revealed to their parents. This in turn raises

[142] Cara Mand et al, '"It Was the Missing Piece": Adolescent Experiences of Predictive Genetic Testing for Adult-Onset Conditions' (2013) 15(8) *Genetics in Medicine* 643, 645–6.

[143] Bernard M Dickens, 'Ethical and Legal Aspects of Noninvasive Prenatal Genetic Diagnosis' (2014) 124(2) *International Journal of Gynecology and Obstetrics* 181, 182, cited in Bennett, Chitty and Lewis (n 100) 199.

[144] Joel Feinberg, 'The Child's Right to an Open Future' in William Aiken and Hugh LaFollette (eds), *Whose Child? Children's Rights, Parental Authority, and State Power* (Rowman and Littlefield, 1980) 124, 125–6. For a discussion of the child's right to an open future in the context of genetic testing, see Annelien L Bredenoord, Martine C de Vries and Hans van Delden, 'The Right to an Open Future concerning Genetic Information' (2014) 14(3) *American Journal of Bioethics* 21.

[145] See, eg, Stephen Wilkinson, *Choosing Tomorrow's Children: The Ethics of Selective Reproduction* (Clarendon Press, 2010) 46; Lainie Friedman Ross, *Children, Families, and Health Care Decision Making* (Clarendon Press, 1998) 48.

[146] Nuffield Council on Bioethics (n 37) 108 [4.56].

[147] Bredenoord, de Vries and van Delden (n 144) 22.

[148] For a discussion of a relational approach to the welfare of the child in the context of donor conception, see Taylor-Sands, *Saviour Siblings* (n 29) ch 4.

[149] Stephen Robertson and Julian Savulescu, 'Is There a Case in Favour of Predictive Genetic Testing in Young Children?' (2001) 15(1) *Bioethics* 26, 42, 47–9.

concerns about potential discrimination in the areas of insurance, employment, education and housing, as well as general social stigmatisation.[150] For example, if insurance companies or employers were able to access genetic information obtained through NIPT, this may impact an individual's ability to obtain insurance or secure employment.[151] In Australia, insurance companies may not use genetic test results to affect provision of health insurance, but they can be used in the context of life, income protection, and travel insurance.[152] Evidence suggests that competent adults often decide not to find out about predictive information available from genetic testing based on concern about discrimination or stigma.[153] For example, uptake on the test for Huntington's disease when it became available was lower than the hypothetical demand.[154] This suggests many at-risk individuals preferred not to know when presented with the option for testing.

There is no clear consensus on whether informing prospective parents about genetic characteristics of their unborn child violates any ethical duty of confidentiality.[155] Parents frequently obtain and hold personal and sensitive information about a child which has the potential to significantly impact the child's welfare, such as the child's biological parentage. Research into adoption and donor-conception reveals that the timing and method of delivering sensitive information to children can be critical to their wellbeing.[156]

[150] See Elizabeth Penziner et al, 'Perceptions of Discrimination among Persons Who Have Undergone Predictive Testing for Huntington's Disease' (2008) 147(3) *American Journal of Medical Genetics Part B* 320, 322–3; RE Duncan and MB Delatycki, 'Predictive Genetic Testing in Young People for Adult-Onset Conditions: Where Is the Empirical Evidence?' (2006) 69(1) *Clinical Genetics* 8, 10.

[151] This concern was raised by the Nuffield Council: Nuffield Council on Bioethics (n 37) 108 [4.57].

[152] Jane Tiller et al, 'Genetic Discrimination by Australian Insurance Companies: A Survey of Consumer Experiences' (2020) 28(1) *European Journal of Human Genetics* 108, 108.

[153] Hercher et al (n 47) 1141–2. See also Pascal Borry, Mahsa Shabani and Heidi Carmen Howard, 'Is There a Right Time to Know? The Right Not to Know and Genetic Testing in Children' (2014) 42(1) *Journal of Law, Medicine & Ethics* 19; Bartha Maria Knoppers, 'From the Right to Know to the Right Not to Know' (2014) 42(1) *Journal of Law, Medicine & Ethics* 6.

[154] Hercher et al (n 47) 1142, citing Christiane Bernhardt et al, 'Decreasing Uptake of Predictive Testing for Huntington's Disease in a German Centre: 12 Years' Experience (1993–2004)' (2009) 17(3) *European Journal of Human Genetics* 295; S Creighton et al, 'Predictive, Pre-Natal and Diagnostic Genetic Testing for Huntington's Disease: The Experience in Canada from 1987 to 2000' (2003) 63(6) *Clinical Genetics* 462; Roslyn J Tassicker et al, 'Problems Assessing Uptake of Huntington Disease Predictive Testing and a Proposed Solution' (2009) 17(1) *European Journal of Human Genetics* 66.

[155] Duncan, Foddy and Delatycki (n 54) 1068. Cf Wadrup et al (n 53) 1223.

[156] See, eg, AJ Turner and A Coyle, 'What Does It Mean to Be a Donor Offspring? The Identity Experiences of Adults Conceived by Donor Insemination and the Implications for Counselling and Therapy' (2000) 15(9) *Human Reproduction* 2041, 2049.

Prospective parents who learn their future child will develop a severe genetic condition later in life face similarly difficult decisions about when and how to share this information with their child. In particular, they should be made aware of the potential for future discrimination and stigma should they disseminate this information more broadly. Laura Hercher et al suggest that prospective parents should receive genetic counselling before and after testing to help them understand the 'responsibilities associated with becoming custodians of their future child's genetic information'.[157] To minimise any breach of confidentiality, Felicity Wadrup et al suggest prospective parents be counselled to not disclose their child's genetic information to relatives or friends.[158]

As an aside, when we are considering the issues of privacy and confidentiality, we might also consider the interests of the parents in addition to the fetus, in particular the other genetic parent. For example, when we consider Huntington's disease, scenarios can occur when the pregnant woman may want to test the fetus, but the at-risk parent (the father) does not want to know his status.[159] In this case, the mother testing the fetus may reveal information about the father that he may not wish to know. This can be avoided by engaging in exclusion testing, where the fetus is tested to see if it has inherited a chromosome from the affected grandparent or not.[160] This allows the prospective parents to determine if the fetus is at 50% chance of having Huntington's disease, without conclusively determining the status of the at-risk parent. However, this has the disadvantage of a 50% chance of a non-affected fetus being terminated (if the decision is made to terminate), as well as requiring co-operation from other family members. If a pregnant woman wishes to proceed to conclusive genetic testing, then the issues of privacy and confidentiality may be raised for one of the parents as well as the fetus. This is because of the inherent nature of genetic information as shared, familial information.[161]

[157] Hercher et al (n 47) 1143.

[158] Wadrup et al (n 53) 1223.

[159] See generally A Erez et al, 'The Right to Ignore Genetic Status of Late Onset Genetic Disease in the Genomic Era: Prenatal Testing for Huntington Disease as a Paradigm' (2010) 152A(7) *American Journal of Medical Genetics Part A* 1774; Ros Tassicker et al, 'Prenatal Diagnosis Requests for Huntington's Disease when the Father Is at Risk and Does Not Want to Know His Genetic Status: Clinical, Legal, and Ethical Viewpoints' (2003) 326(7384) *BMJ* 331.

[160] A Tyler et al, 'Exclusion Testing in Pregnancy for Huntington's Disease' (1990) 27(8) *Journal of Medical Genetics* 488, 488.

[161] Australian Law Reform Commission, *Essentially Yours: The Protection of Human Genetic Information in Australia* (Report No 96, March 2003) 237 [7.12].

D *Interests of Broader Society*

Expanding the scope of prenatal testing raises broader societal concerns about what constitutes a 'healthy pregnancy', parental expectations of future children, equality, inclusivity and diversity. Frequently this concern is described in terms of a slippery slope towards 'designer babies'.[162] Beyond the hyperbole lies a genuine fear about our future humanity in a world where pregnancies are terminated (or women feel pressure to terminate)[163] for potentially trivial or eugenic reasons.[164] As foreshadowed by Isabel Karpin and Kristin Savell, governments may consider limiting access to prenatal testing technologies for conditions that are 'either insufficiently serious or inappropriate'.[165] For example, the governments of some countries, such as India and China, have specifically introduced regulations limiting access to prenatal sex determination (primarily ultrasound, although NIPT is also covered) in order to prevent sex-selective terminations of pregnancy.[166] The motivations for such legislation may be in part to prevent individual-level harm (certain gender role expectations placed on the future child), but more primarily societal-level harms (imbalanced sex ratios; reinforcing harmful gender norms). There is therefore a precedent for limiting access to prenatal testing based on the interests of those beyond the individual parent.

Routinely testing for adult-onset conditions could potentially be criticised as actively promoting a culture of selective reproduction that devalues the lives of those living with disability. The Nuffield Council highlighted the importance of considering the wider societal environment when developing policy in relation to NIPT by promoting the values of equality, fairness and inclusion.[167] In particular, the Nuffield Council expressed

[162] See, eg, Sally Sheldon and Stephen Wilkinson, 'Hashmi and Whitaker: An Unjustifiable and Misguided Distinction?' (2004) 12(2) *Medical Law Review* 137, 148–9; Brownsword and Wale (n 41) 655.

[163] For a discussion of the maternal pressure to terminate see Karpin and Savell (n 25) 3. This is raised in Bennett, Chitty and Lewis (n 100) 201.

[164] For a detailed discussion of concerns about the impact of increased genetic selection on human dignity, see generally Leon R Kass, *Life, Liberty and the Defense of Dignity: The Challenge for Bioethics* (Encounter Books, 2002); Francis Fukuyama, *Our Posthuman Future: Consequences of the Biotechnology Revolution* (Strauss & Giroux, 2002) ch 5; Bill McKibben, *Enough: Genetic Engineering and the End of Human Nature* (Bloomsbury, 2003). By way of contrast, some evidence suggests that experience with certain conditions makes it more likely that parents will make selective reproductive choices: Felicity Kate Boardman, 'The Expressivist Objection to Prenatal Testing: The Experiences of Families Living with Genetic Disease' (2014) 107 *Social Science & Medicine* 18, 18, 23.

[165] Karpin and Savell (n 25) 301.

[166] Bowman-Smart et al, 'Sex Selection and Non-Invasive Prenatal Testing' (n 8) 400.

[167] Nuffield Council on Bioethics (n 37) 121–2 [5.25]–[5.28].

> concerns about the potential future uses of NIPT for less significant medical conditions and impairments, non-medical traits and whole genome and exome sequencing that relate to the potential long-term societal implications of such uses.[168]

Good regulation should be supported by robust ethical analysis and empirical evidence, particularly when it impacts on reproductive choice. According to John Harris, reproductive choices should 'not simply be dismissed wherever and whenever a voting majority can be assembled against them'.[169] Concerns about the impact of selective reproduction on society at large or particular groups within it have been discussed in detail in the broader debate around selecting out 'less significant' medical impairments[170] and non-medical sex selection.[171] These concerns, which arise when parents choose to terminate a pregnancy upon finding out genetic information about the fetus, raise questions about whether a threshold of 'seriousness' ought to be applied to conditions being tested.[172] As discussed in Part II(A) above, a flexible approach is needed in determining an appropriate 'threshold' for NIPT in particular contexts.

Some commentators argue that to restrict access to prenatal testing for *any* condition on the basis of broader societal concerns would be unduly paternalistic.[173] Certain adult-onset conditions, such as Huntington's disease, are considered sufficiently serious to warrant prenatal testing for the purposes

168 Ibid 122 [5.28].

169 John Harris, 'Sex Selection and Regulated Hatred' (2005) 31(5) *Journal of Medical Ethics* 291, 293. Sally Sheldon and Stephen Wilkinson argue that a 'slide down the slope' toward 'designer babies' can be averted by careful regulation: Sally Sheldon and Stephen Wilkinson (n 162) 148.

170 Nuffield Council on Bioethics (n 37) 106–9 [4.52]–[4.59], 130 [6.12]–[6.13]. The authors of this article acknowledge the loaded and problematic nature of this terminology.

171 See, eg, Michelle Taylor-Sands, 'Non-Medical Sex Selection: Sliding Down the Slippery Slope?' in Ian Freckelton and Kerry Petersen (eds), *Tensions and Traumas in Health Care* (Federation Press, 2017) 317 ('Non-Medical Sex Selection'). A detailed analysis of these objections is beyond the scope of this article.

172 The Nuffield Council recommended that NIPT should not normally be used to test for 'a less significant medical condition or impairment': Nuffield Council on Bioethics (n 37) 130 [6.13]. The Nuffield Council report contains a discussion of the concerns on which this is based: at 106–9 [4.52]–[4.59]. For an in-depth analysis of the concept of 'serious disability', see Karpin and Savell (n 25) ch 3.

173 Rosamond Rhodes, 'Resisting Paternalism in Prenatal Whole-Genome Sequencing' (2017) 17(1) *American Journal of Bioethics* 35, 36. See also Stephanie C Chen and David T Wasserman, 'A Framework for Unrestricted Prenatal Whole-Genome Sequencing: Respecting and Enhancing the Autonomy of Prospective Parents' (2017) 17(1) *American Journal of Bioethics* 3.

of facilitating early termination.[174] Furthermore, as noted above, decisions about prenatal testing are not necessarily linked to termination of pregnancy. Roger Brownsword and Jeff Wale highlight the importance of the 'distinction between a woman simply wanting to know and her wanting to know in order to decide whether to terminate her pregnancy'.[175] However, we would question whether these motivations can be so neatly and clearly demarcated in actual practice. A key issue arising out of testing for adult-onset conditions where termination may not ultimately be carried out is how to manage both the interests of parents in wanting to know and those of the future child outlined above. We discuss how this might be achieved through regulation in the next Part.

IV A Facilitative Approach to Regulating NIPT

A *Should NIPT Be Regulated by the State?*

The perceived tensions around NIPT between the interests of prospective parents, the future child, and society, raise important questions about what support prospective parents require to make informed reproductive decisions and when (if ever) the state should intervene in those decisions. Given the myriad of complex and interrelated interests outlined in Part III, there is an important role for regulation that supports prospective parents in prenatal decision-making and adequately protects the welfare of the future child.

The term 'regulation' has a 'bewildering variety of meanings'[176] and can denote a vast array of different controls. As noted in Part II, reproductive medicine is regulated over and above the legal and ethical restraints that apply generally to medical practitioners. Brownsword argues there is 'little virtue in leaving genetics to the play of subjective preference and the market'.[177] Various justifications are cited for directly regulating certain areas of reproductive medicine. It is helpful to briefly explore the justifications for regulating ART as they are potentially relevant to NIPT. According to Helen Szoke, the state plays an important role in resolving any conflicts which may exist over reproductive technologies and 'formalising an expression of the public interest'.[178] Leslie

[174] Nuffield Council on Bioethics (n 37) 84 [3.33].

[175] Brownsword and Wale (n 41) 659.

[176] Anthony Ogus, *Regulation: Legal Form and Economic Theory* (Clarendon Press, 1994) 1.

[177] Roger Brownsword, 'Regulating Human Genetics: New Dilemmas for a New Millennium' (2004) 12(1) *Medical Law Review* 14, 15.

[178] Helen Szoke, 'The Nanny State or Responsible Government?' (2002) 9(4) *Journal of Law and Medicine* 470, 481. Szoke recognises that resolution by the state will not satisfy all parties in a pluralist society.

Cannold and Lynn Gillam similarly argue that 'the state has an obligation to protect the interests of its citizens and regulation is a legitimate method of achieving this'.[179] Applying a harm-based approach to regulation as proposed by political liberalist John Stuart Mill,[180] Cannold and Gillam argue:

> It is possible that ART can be practised in ways that threaten the interests of at least some citizens and so, in principle, it is ethically permissible for the state to regulate in such situations.[181]

The rationale for regulating ART to prevent harm to others also applies to current and future applications of NIPT. As discussed in Part III, various commentators have raised concerns about potential harms to the future child where NIPT is used to detect adult-onset conditions and the pregnancy is continued.[182] The Nuffield Council adopted a harm-based approach in its recommendations for regulating NIPT, arguing that

> restricting freedoms in order to prevent harm to others may mean limiting the freedom to access NIPT in some circumstances in order to protect the fetus, or to prevent harm to wider society.[183]

However, as Colin Gavaghan points out, 'the concept of "harm" is not as straightforward as Mill … perhaps assumed, and it is especially problematic in relation to "genesis questions"'.[184] In the context of NIPT, the potential for a termination of pregnancy means the interests of the future child are conditional on that child coming into existence. This distinguishes the future child's interests from the current existing interests of the prospective parents. According to Emily Jackson, the welfare of the future child should not be relied on as a justification for interfering with an individual's reproductive choices.[185] Given the potential for regulation of NIPT to interfere with reproductive

[179] Leslie Cannold and Lynn Gillam, 'Regulation, Consultation and Divergent Community Views: The Case of Access to ART by Lesbian and Single Women' (2002) 9(4) *Journal of Law and Medicine* 498, 501.

[180] Ibid 500; John Stuart Mill, 'On Liberty' in Mary Warnock (ed), *Utilitarianism and on Liberty* (Blackwell Publishing, 2nd ed, 2003) 88, 94.

[181] Cannold and Gillam (n 179) 501–2.

[182] A similar argument may be made about limiting access to NIPT for non-medical sex selection as is currently the case with PGD: Karpin and Savell (n 25) 300–1.

[183] Nuffield Council on Bioethics (n 37) 29 [1.67].

[184] Colin Gavaghan, *Defending the Genetic Supermarket: Law and Ethics of Selecting the Next Generation* (Routledge-Cavendish, 2007) 37–8.

[185] Emily Jackson, 'Conception and the Irrelevance of the Welfare Principle' (2002) 65(2) *Modern Law Review* 176, 176.

choice, any intervention 'must have a sound ethical basis and also take into account evidence of harm to children or to society'.[186]

Martin Johnson extends the rationale for direct regulation of ART beyond the harm principle by proposing that regulation should *support* 'those who should be capable of making an informed decision' and *protect* 'those incapable of protecting themselves'.[187] Johnson's regulatory approach goes beyond Mill's harm principle to suggest that, in addition to protecting the interests of those who may be born, regulation also plays an important role in supporting patients seeking treatment to make informed decisions. This lays the path for a more facilitative approach to regulation as opposed to one focused primarily on restricting access to services. The interests of broader society are notably missing from Johnson's formulation. While the interests of broader society outlined in Part III are important considerations in a regulatory framework for NIPT, they should not be overstated at the expense of reproductive choice. As Brownsword and Wale point out:

> While it is one thing to restrict reproductive autonomy and choice in order to prevent catastrophic harm to the conditions for human existence, it is quite another to restrict it because this is the majority's preference in a particular community.[188]

Although routinely testing for adult-onset conditions might be seen by some as devaluing the lives of those living with disability, more evidence is needed to substantiate this conclusion in order to justify a restrictive approach to NIPT that curtails reproductive choice. Where the boundaries should lie for NIPT more generally is an important issue requiring rigorous ethical debate based on strong empirical foundations.[189] Although this article focuses on the potential impact on the future child of using NIPT to detect adult-onset conditions, it is important to acknowledge concerns about the impact of generally expanding

[186] Science and Technology Committee, House of Commons, *Human Reproductive Technologies and the Law* (House of Commons Paper No 7, Session 2004–05) 169 [390] <https://publications.parliament.uk/pa/cm200405/cmselect/cmsctech/7/7i.pdf>.

[187] Martin H Johnson, 'The Art of Regulation and the Regulation of ART: The Impact of Regulation on Research and Clinical Practice' (2002) 9(4) *Journal of Law and Medicine* 399, 405. Johnson envisages that the first category would include patients and possibly doctors. He suggests that the second category 'would include children (desired and existing), the embryo', and gamete donors.

[188] Brownsword and Wale (n 41) 652–3.

[189] For example, an argument for restricting the use of NIPT for non-medical sex selection may be based on empirical evidence of sex-ratio imbalances in certain societies with a preference for a particular sex: see, eg, Kristina Edvardsson et al, 'Male-Biased Sex Ratios in Australian Migrant Populations: A Population-Based Study of 1 191 250 Births 1999–2015' (2018) 47(6) *International Journal of Epidemiology* 2025, 2027–36.

the conditions for which NIPT is offered on broader society (or particular groups within it). These concerns have been raised in the context of PGT for which a threshold of 'seriousness' notionally limits the conditions for which testing is available. PGT therefore provides a useful comparator for how NIPT might be regulated.

B *A Comparator: Regulation of PGT*

Access to PGT, which involves genetic screening of embryos prior to implantation, is limited by national guidelines and legislation dealing with ART in certain states.[190] Prospective parents in Australia can use PGT to detect serious genetic conditions and improve ART outcomes[191] but are expressly prohibited from using PGT to select in favour of a serious genetic condition[192] or for non-medical sex selection,[193] on the basis that this may be harmful to the future child.[194] This is similar to the situation in the UK, where PGT is available subject to authorisation by the Human Fertilisation and Embryology Authority.[195] The use of PGT to screen out certain genetic conditions has attracted considerable controversy about what constitutes a 'serious' genetic condition or disability.[196] As with NIPT, the scope of PGT has raised similar concerns about eugenics and the slippery slope toward 'designer babies'.[197] The

[190] *ART Act (Vic)* (n 30) s 28; *HRT Act (WA)* (n 30) s 14(2b); *NHMRC Guidelines* (n 30) 73–7 [8.15]–[8.19]. See also *ART Act (SA)* (n 30) s 9(1)(c).

[191] *NHMRC Guidelines* (n 30) 73 [8.15.1]. The guidelines refer to 'conditions, diseases or abnormalities that would severely limit the quality of life' of the future child.

[192] Ibid 73 [8.15.2].

[193] Ibid 72 [8.14.1]. See also *ART Act (Vic)* (n 30) s 28; *Human Fertilisation and Embryology Act 1990* (UK) sch 2 paras 1ZA(1)(c), 1ZB ('*HFE Act*').

[194] This raises a question about the acceptability of parents choosing to transfer an embryo with a genetic condition in cases where PGT identifies all available embryos as having the condition. Some commentators argue that transferring an 'affected' embryo in such situations would not constitute a 'preference' for disability: see Isabel Karpin and Roxanne Mykitiuk, 'Reimagining Disability: The Screening of Donor Gametes and Embryos in IVF' (2021) 8(2) *Journal of Law and the Biosciences* 1, 6. See also Andrew Joseph, 'A Baby with a Disease Gene or No Baby at All: Genetic Testing of Embryos Creates an Ethical Morass', *STAT* (Web Page, 23 October 2017) <https://www.statnews.com/2017/10/23/ivf-embryo-genetic-testing>, archived at <https://perma.cc/8QKV-DH6P>.

[195] *HFE Act* (n 193) sch 2 paras 1(1)(b)–(c), 1ZA(1)(c).

[196] For an in-depth analysis of the concept of 'serious disability', see Karpin and Savell (n 25) ch 3. See, more recently, Karpin and Mykitiuk (n 194). Scholarship over the last 25 years highlights both the medical and social aspects of disability: see, eg, Jonathan Glover, *Choosing Children: Genes, Disability, and Design* (Clarendon Press, 2006) 6–8; Wilkinson (n 145) 164–6.

[197] For an in-depth discussion of the ethical concerns associated with selective reproduction and PGT, see Taylor-Sands, *Saviour Siblings* (n 29) 6–20.

extent to which PGT is available to screen out susceptibility to conditions (such as *BRCA1* and *BRCA2* susceptibility for breast cancer) or adult-onset conditions (such as Huntington's disease) has, for example, led to concerns about where the line should be drawn in selective reproduction.[198] The therapeutic benefit in selecting out adult-onset and susceptibility to conditions is more uncertain as the child may have a healthy 30–40 years before the disease manifests itself. Moreover, in the case of susceptibility to conditions, the child may never develop the disease or, if they do, may have it effectively treated. As Isabel Karpin and Roxanne Mykitiuk note, the scope of testing 'is in part determined by what conditions it is possible to test for',[199] which inevitably changes over time.

Recognising that the ethical acceptability of selecting out specific genetic conditions depends on context and may change over time, national guidelines in Australia provide clinics with factors to consider when assessing whether or not to provide PGT.[200] These include: current evidence about the impact of the condition on the quality of life of the future child; parental concerns about their ability to care for the child; the availability of treatments for the condition; the limitations of technology (including false positive and false negative results); the experiences of individuals and families living with the condition and the stigma associated with it; and the extent of social supports available to the prospective parents.[201] Prospective parents seeking access to PGT should also be given access to a geneticist and genetic counsellor and provided relevant information to enable an informed treatment decision.[202] This approach to regulating PGT reflects a facilitative approach that focuses on supporting prospective parents in making treatment decisions and accommodates the different contexts in which these decisions take place. This creates a flexible threshold for limiting access to PGT for serious medical conditions that may vary between individual cases.[203]

There is arguably less justification for limiting access to NIPT than PGT. Unlike PGT, where decisions about testing are made prior to implantation of an embryo, limiting access to NIPT has significant implications for a pregnant woman, in particular her bodily integrity. Although the outcomes of prenatal

198 See, eg, Julia Medew, 'Couples Use IVF to Pick Genes', *The Age* (Melbourne, 3 July 2012) 1.

199 Karpin and Mykitiuk (n 194) 10.

200 *NHMRC Guidelines* (n 30) 73–4 [8.16].

201 Ibid 74 [8.16.1].

202 Ibid 75–6 [8.18].

203 Some commentators argue, however, that not setting clear limits runs the risk of creating a 'closed future' in which a 'particular imaginary of disability avoidance is enabled': Karpin and Mykitiuk (n 194) 10, 12.

and pre-implantation testing at an individual and societal level may be similar,[204] NIPT occurs at a time when the woman and fetus are inextricably interconnected. This interrelationship between a pregnant woman and her fetus requires the law to traverse difficult territory and demands a nuanced approach to support and protect not only the interests of all relevant parties, but the relationship that connects them. The potential pressure on women to test for a broader array of conditions as NIPT becomes more accessible is arguably best addressed through a facilitative regulatory regime similar to the national guidelines on PGT which aims to educate and support prospective parents in deciding the scope of any available testing.[205]

Broader societal concerns about the impact of testing for adult-onset and susceptibility to conditions, and the slippery slope towards 'designer babies', require ongoing debate to determine where, if at all, any clear limits on NIPT should be placed in relation to specific conditions as new applications for NIPT arise.[206] For example, the value and ethical acceptability of testing for autism spectrum disorder is disputable.[207] The current prohibition on using PGT for non-medical sex selection represents a clear limit that suggests an ethical distinction between therapeutic outcomes[208] and parental preferences for a particular 'type' of child.[209] It may be appropriate to consider a similar limit on the scope of NIPT for non-therapeutic or 'trivial' reasons, such as selecting eye

[204] For example, allowing PGT and NIPT to detect the sex of an embryo or fetus may enable parents to select the sex of their child and has potential to create an imbalance of male–female live birth ratios within a society.

[205] A facilitative approach that educates and supports is preferable to a restrictive one, given the potential availability of NIPT through DTC testing: Brownsword and Wale (n 41) 660.

[206] As noted elsewhere, restrictions on access to reproductive testing based on concerns about the welfare of the future child should be supported by robust empirical evidence: Taylor-Sands, 'Non-Medical Sex Selection' (n 171) 321.

[207] Although there is currently no genetic test for autism, which is a multi-factorial condition, it has been proposed that screening out male embryos will potentially reduce likelihood of having a child with this condition as the condition appears to be more prevalent in males: Kristien Hens, Hilde Peeters and Kris Dierickx, 'The Ethics of Complexity: Genetics and Autism, a Literature Review' (2016) 171B(3) *American Journal of Medical Genetics Part B* 305, 311. The assumption that autism is more prevalent in males may, however, be incorrect as more recent studies suggest autism in females is under-diagnosed due to 'masking' of symptoms: Laura Hull, KV Petrides and William Mandy, 'The Female Autism Phenotype and Camouflaging: A Narrative Review' (2020) 7(4) *Review Journal of Autism and Developmental Disorders* 306.

[208] In this article, we take an expansive view of 'therapeutic' outcomes to include both personal and clinical utility.

[209] Whilst the distinction between therapeutic and non-therapeutic outcomes is an important one, the concerns about slippery slopes are arguably not well founded, at least in the case of non-medical sex selection: Taylor-Sands, 'Non-Medical Sex Selection' (n 171) 325–9.

colour, athleticism and intelligence. This raises a broader question about the ethics of 'enhancement' and how society wishes to reproduce in the future, which is beyond the scope of this article.[210]

In the next section, we argue that regulation of NIPT for adult-onset conditions should focus on supporting prospective parents seeking testing and protecting any future child who may be born. As with PGT, any threshold for limiting access to NIPT should be approached in a flexible way that may vary depending on various factors and the individual circumstances of the prospective parents.

C *A Relational Approach to Reconciling Interests*

In this section, we outline a facilitative approach to regulating NIPT for adult-onset conditions that acknowledges and attempts to accommodate the sometimes-conflicting yet interconnected interests of prospective parents and the future child by viewing them through a relational lens. The discussion in Part III highlighted a tension between the individual interests of the prospective parents and those of the future child in cases where a pregnancy is continued after NIPT testing is positive for an adult-onset condition. Much of the debate to date has focused on this tension, with minimal recognition of the interrelated nature of these interests.[211] The interests of the future child are, however, inextricably linked with the interests of the prospective parents, and the law has acknowledged the inevitability that prospective parents ultimately carry the primary responsibility of caring for their child once born. Courts and policymakers have generally upheld the principles of bodily integrity and reproductive choice to support prospective parents rather than impose restrictions or punitive measures on antenatal behaviour, which might cause harm to a future child.

Careful thought is required in framing the various interests at stake in NIPT to acknowledge the 'conflict, confluence, and confusion of interests' within families.[212] By adopting the language of individual and competing rights, legal

[210] There is considerable disagreement about the ethics of 'enhancement': see, eg, Julian Savulescu, 'Procreative Beneficence: Why We Should Select the Best Children' (2001) 15(5–6) *Bioethics* 413, 414; Selina Metternick-Jones, 'Choosing Impairment: Conflicting Interests' (2018) 18(2) *QUT Law Review* 229, 244–5. Cf Michael J Sandel, 'The Case against Perfection: What's Wrong with Designer Children, Bionic Athletes, and Genetic Engineering' (April 2004) *The Atlantic Monthly* 51, 52.

[211] As noted in Part II(D), there is some recognition of the interrelated nature of interests within families in the HGSA, ASHG and ACMG clinical guidelines on predictive testing of minors.

[212] Robert A Crouch and Carl Elliott, 'Moral Agency and the Family: The Case of Living Related Organ Transplantation' (1999) 8(3) *Cambridge Quarterly of Healthcare Ethics* 275, 284.

analysis has often struggled to manage the tension between the rights of the future child to decisional autonomy and an open future, and those of the prospective parents to reproductive choice. A shift in language to 'interests' rather than 'rights' promotes recognition of the interconnected nature of these interests by reconceptualising both the individual and collective interests at stake. In the face of new challenges arising out of technological developments in genetic testing, the law has been forced to revisit individualistic legal paradigms to address the complex and overlapping interests involved. For example, in the recent case of *ABC v St George's Healthcare NHS Trust*, the Court of Appeal of England and Wales endorsed a relational approach to patient autonomy by acknowledging that a decision made by one individual about genetic testing has implications for other family members in making their own informed decisions.[213]

In the context of NIPT, we propose a relational approach for evaluating and promoting the interests of the future child *in connection with* the interests of prospective parents and any existing children.[214] A relational approach views the future child within the social context of the family into which they will be born and acknowledges that parents, as primary carers, have a broad discretion in making (often significant) decisions on behalf of their children.[215] Relational theory has evolved over the last 20 years as a result of attempts by feminist[216]

[213] [2017] EWCA Civ 336, [44] (Irwin LJ, Underhill LJ agreeing at [67], Gloster LJ agreeing at [68]). Based on this reasoning, the Court of Appeal determined the defendant's case should not be struck out: at [42]–[45] (Irwin LJ, Underhill LJ agreeing at [67], Gloster LJ agreeing at [68]). The matter was remitted to the High Court to be determined on the evidence, and in 2020 Yip J left open the possibility that a clinician could owe a duty of care to a family member: *ABC v St George's Healthcare NHS Trust* [2020] EWHC 455 (QB), [259]–[261]. For a discussion of the relational approach applied by the Court of Appeal, see Roy Gilbar and Charles Foster, 'It's Arrived! Relational Autonomy Comes to Court: *ABC v St George's Healthcare NHS Trust* [2017] EWCA 336' (2018) 26(1) *Medical Law Review* 125, 132–3. For a discussion of relational approaches generally, see Catriona Mackenzie and Natalie Stoljar's account of 'relational autonomy': Catriona Mackenzie and Natalie Stoljar, 'Introduction: Autonomy Refigured' in Catriona Mackenzie and Natalie Stoljar (eds), *Relational Autonomy: Feminist Perspectives on Autonomy, Agency, and the Social Self* (Oxford University Press, 2000) 3. See also Virginia Held, *The Ethics of Care: Personal, Political, and Global* (Oxford University Press, 2006).

[214] For a discussion of a relational approach to the welfare of the future child in the context of ART, see Taylor-Sands, *Saviour Siblings* (n 29) 26–9.

[215] Ibid 26.

[216] For example, Mackenzie and Stoljar's account of 'relational autonomy' reconceptualises autonomy from a feminist perspective: Mackenzie and Stoljar (n 213) 3–4. Virginia Held has developed an alternative moral paradigm, based on Carol Gilligan's work on moral psychology and the ethics of care: Held (n 213) 70–1. See also Carol Gilligan, *In a Different Voice: Psychological Theory and Women's Development* (Harvard University Press, 1993).

and communitarian[217] scholars to put the 'relational' into the moral realm. There are, however, limits on parental discretion, and both criminal and child welfare laws are designed to protect children (once born) from harm and neglect. Various commentators have offered different perspectives on when state intervention into parental healthcare decision-making is justified to prevent unacceptable risk of harm to a child. According to Gillam's idea of a 'zone of parental discretion', parents generally have significant discretion to make decisions for their children, subject to 'probable significant' harm.[218] Lainie Friedman Ross proposes a similar notion of 'constrained parental autonomy', which focuses on the likelihood and degree of risk of harm to a child.[219] Other commentators have proposed, in the context of selective reproduction, approaches that attempt to weigh risks against benefits to the future child.[220] Commentary on the ethics of intra-familial donations imposes what is arguably a lower threshold for state intervention of 'significant risk of serious harm'.[221]

The point at which the state should intervene in parental decision-making is a complex question and will vary in different contexts and different stages of a child's life. We argue that, in the context of NIPT and based on current empirical evidence, there is insufficient basis for the law to restrict prospective parents from accessing prenatal testing for adult-onset conditions where this information has clinical or personal utility. Whilst the future child's interests ought to be protected, there is currently insufficient and inconclusive evidence of harm to the child to justify restricting the prospective parents' reproductive choice to find out information about such conditions. As discussed in Part III,

217 Communitarian theory has also influenced the debate over how the welfare of the child is conceptualised. For example, Nancy Sherman emphasises the importance of collective endeavour or 'affiliation' to human flourishing: Nancy Sherman, 'The Virtues of Common Pursuit' (1993) 53(2) *Philosophy and Phenomenological Research* 277, 278.

218 Lynn Gillam, 'The Zone of Parental Discretion: An Ethical Tool for Dealing with Disagreement between Parents and Doctors about Medical Treatment for a Child' (2016) 11(1) *Clinical Ethics* 1, 4. See also Rosalind McDougall, Clare Delany and Lynn Gillam (eds), *When Doctors and Parents Disagree: Ethics, Paediatrics and the Zone of Parental Discretion* (Federation Press, 2016).

219 Ross (n 145) 50–2.

220 S Sheldon and S Wilkinson, 'Should Selecting Saviour Siblings Be Banned?' (2004) 30(6) *Journal of Medical Ethics* 533, 535–6. See also G Pennings, R Schots and I Liebaers, 'Ethical Considerations on Preimplantation Genetic Diagnosis for HLA Typing to Match a Future Child as a Donor of Haematopoietic Stem Cells to a Sibling' (2002) 17(3) *Human Reproduction* 534, 537.

221 Sarah Elliston, *The Best Interests of the Child in Healthcare* (Routledge-Cavendish, 2007) 257. See also Ferdinand Schoeman, 'Parental Discretion and Children's Rights: Background and Implications for Medical Decision-Making' (1985) 10(1) *Journal of Medicine and Philosophy* 45, 60.

evidence of harm to the future child is contingent, speculative and largely theoretical at this stage.[222] Karpin highlights that state intervention based on speculative harm to the welfare of children 'is prone to invidious bias and distortion'.[223] There is also some evidence to suggest that the future child could potentially benefit from this information being made available early on.[224] The potential harms identified in Part III can be largely mitigated with careful regulation in this area. We propose a facilitative approach to regulating NIPT that supports and educates parents seeking prenatal testing and thereby promotes the welfare of the future child. A facilitative approach to NIPT is consistent with the current clinical guidelines on prenatal screening discussed in Part II, which recognise both the clinical and personal utility of testing for prospective parents.

Although guidelines on predictive testing of minors prioritise the welfare of the child over the interests of parents, there are stronger arguments for allowing access to prenatal testing than testing of minors. First, prenatal testing involves testing the woman's blood and, at the time of testing, the fetus is contained within (and dependent on) the woman. An approach that prioritises the welfare of the child at the expense of the mother's bodily integrity is not only inconsistent with common law principles[225] but also nonsensical as the child's welfare, and very existence, are dependent on the mother and the choices that she makes. Moreover, there are important practical considerations if access to NIPT is made contingent on certain decisions, such as an intention to terminate the pregnancy. Counselling around NIPT is more likely to be directive rather than supportive if access is limited to certain scenarios. Limiting access may also encourage parents to lie about their intentions in order to access NIPT, thereby eroding the patient–doctor therapeutic relationship. Finally, prospective parents may simply resort to an unregulated DTC sector for prenatal testing with a corresponding lack of protection for all parties. As Brownsword and Wale note:

> [W]hile we might endlessly debate the rights and wrongs of particular uses of NIPT, we should lower our expectations about the effective implementation of any policy that impinges on the reproductive choices that pregnant women —

[222] See Wakefield et al (n 140) 760.

[223] Isabel Karpin, 'The Legal and Relational Identity of the "Not-Yet" Generation' (2012) 4(2) *Law, Innovation and Technology* 122, 128–9.

[224] See Mand et al, 'Predictive Genetic Testing in Minors' (n 137) 522.

[225] See, eg, *Re MB* (n 79) 436–7 (Butler-Sloss LJ).

> or, at any rate, pregnant women with sufficient resources — might want to have.[226]

A facilitative approach to NIPT accords with the common law principles of bodily integrity and reproductive autonomy applied by courts in Australia and the UK.[227] In addition, policymakers are reluctant to intervene with a woman's antenatal behaviour even when it may cause harm to the future child.[228] Whilst regulation should aim to protect the welfare of the future child, there is currently insufficient evidence of harm to the future child to restrict prospective parents' reproductive choice to test for adult-onset conditions. Concerns about negative psychosocial outcomes and the future child's autonomy as a result of NIPT are, so far, largely theoretical and rely on an individualised account of the child, which neglects the collective interests a child shares with their family. Moreover, some commentators suggest a child may in fact benefit from their genetic information being obtained by NIPT. Although potential discrimination and stigmatisation are legitimate concerns, any harm that might result from a breach of the future child's confidentiality may be mitigated by providing prospective parents with appropriate support and genetic counselling before and after testing.

D *Recommendations for Future Reform*

The two key goals for regulating NIPT for adult-onset conditions should be to support parents in making ethically and emotionally complex decisions about testing, and to protect the welfare of the future child from potential harms — particularly where the child is born following a diagnosis. Given that the interests of the future child are intertwined with, and dependent on, the interests of their parents, any regulation around NIPT should enable prospective parents to access relevant information so they can make informed choices that equip them to care for their future child.[229] Although a detailed regulatory framework is beyond the scope of this article, we make the following

[226] Brownsword and Wale (n 41) 660.

[227] *JWB* (n 73) 233–4 (Mason CJ, Dawson, Toohey and Gaudron JJ); *Re T* (n 74) 102 (Lord Donaldson MR).

[228] See, eg, *Prenatal Screening and Diagnostic Testing* (n 35) 6 recommendation 2.

[229] As Emily Jackson and Shelley Day Sclater point out, protecting autonomous choice 'may not only involve simply an *absence* of state interference, but could require the positive provision of resources to enable someone to have a meaningful set of options': Emily Jackson and Shelley Day Sclater, 'Introduction: Autonomy and Private Life' in Shelley Day Sclater et al (eds), *Regulating Autonomy: Sex, Reproduction and Family* (Hart Publishing, 2009) 1, 2 (emphasis in original). See also Mills (n 92) 640.

preliminary recommendations about how regulation may support a facilitative approach to NIPT for adult-onset conditions. The framework we map out could also be adapted and applied more broadly to NIPT in general, as clinical capabilities for testing are further developed in the future.[230]

A range of models on the regulatory spectrum operate in the area of ART in the UK and Australia, some of which are more prescriptive than others.[231] Our proposal for a facilitative regulatory framework is based on a permissive regulatory model, which offers greater flexibility than a prescriptive framework and is intended to guide behaviour through clinical practice guidelines. Guidelines are sometimes described as 'soft law' as they lack formal monitoring mechanisms of 'hard law' legislation.[232] However, soft law regulatory regimes can have a significant influence on behaviour and can evolve more easily than hard law regimes in response to technological developments and changing community concerns.[233] To ensure consistency, we propose national guidelines similar to those that currently regulate PGT. As noted above, these guidelines acknowledge that the scope of conditions for which screening should be offered is context-specific and likely to change over time. Although the national guidelines on PGT do not have the force of law, compliance with them is a key element in the national accreditation system for state and territory-based ART clinics in Australia and is linked to federal funding.[234] National guidelines are easier to implement than statute, given the constitutional limitations on the legislative power of the Commonwealth Parliament to legislate nationally in the area of health.[235] Arguably, national guidelines are more appropriate for regulating prenatal screening in Australia than legislation, as a national

[230] For a discussion of the timing for introducing NIPT in antenatal pathways, see Zuzana Deans and Ainsley Janelle Newson, 'Ethical Considerations for Choosing between Possible Models for Using NIPD for Aneuploidy Detection' (2012) 38(10) *Journal of Medical Ethics* 614, 615–17.

[231] For a detailed discussion of different regulatory models in reproductive medicine within Australia and the UK and their relative merits, see Taylor-Sands, *Saviour Siblings* (n 29) 135–7.

[232] Brownsword (n 177) 18.

[233] Helen Szoke, 'Australia: A Federated Structure of Statutory Regulation of ART' in Jennifer Gunning and Helen Szoke (eds), *The Regulation of Assisted Reproductive Technology* (Ashgate, 2003) 75, 81.

[234] Reproductive Technology Accreditation Committee, Fertility Society of Australia, *Code of Practice for Assisted Reproductive Technology Units* (as at October 2017) 5. Accreditation is now mandatory under Commonwealth legislation regulating embryo research and human cloning: *Research involving Human Embryos Act 2002* (Cth) ss 8 (definition of 'accredited ART centre') 11.

[235] The Commonwealth Parliament does not have express power under the *Constitution* to legislate in relation to the provision of ART services or health and medicine generally.

approach reduces regulatory duplication, ensures common ethical standards, prevents 'reproductive tourism' between states,[236] and can support national data collection for any longitudinal studies on the welfare of the future child.[237] In the remainder of this section, we outline some key aspects of a regulatory model for NIPT based on the ethical concerns raised in relation to screening for adult-onset conditions.

First, clear guidance is needed on the scope and availability of NIPT. Ideally, NIPT should be available to all women through the public health sector, which would require its inclusion within Australia's Medicare health insurance scheme. Any threshold for limiting access to NIPT in specific cases should be approached in a flexible way that takes account of the treatability, penetrance and seriousness of the condition, any limitations of the technology, current evidence about any negative impacts on the future child, and the individual circumstances and risk appetites of prospective parents. Ongoing discussion about whether a clear threshold of seriousness should apply to specific conditions for which testing is sought will determine any limits on the scope of NIPT in the future. Arguably, access to NIPT should be limited to therapeutic conditions rather than parental preferences for a particular type of child, at least where NIPT is publicly funded.

Secondly, women should have access to NIPT to test for adult-onset conditions, irrespective of any plans to terminate the pregnancy. This may extend to obtaining information for personal as well as clinical utility. Whether this will be a matter of provision upon request, or offered as part of a panel that can be selected from, requires further empirical research to inform guidelines as there are additional considerations, particularly in the public sector, such as resource allocation.

Thirdly, guidelines should support parents seeking NIPT to make informed reproductive choices by ensuring pregnant women receive adequate care, treatment, information and counselling before, during and after testing. Guidelines crafted in this way would address the concerns raised about harm to the future child through a supportive and informative framework rather than a restrictive approach. Relevant information includes the scope of testing

[236] In the context of ART, the Victorian Law Reform Commission reported that the absence of uniform ART legislation has led to 'reproductive tourism' within Australia, whereby people who are ineligible for treatment in one state travel to unregulated states to undergo treatment: Victorian Law Reform Commission, *Assisted Reproductive Technology & Adoption* (Final Report, March 2007) 44, 55.

[237] As Joyce Harper et al point out in the context of IVF, clinics have a duty to evaluate data surrounding the efficacy of interventions and collect long-term data on the health of any future child: Joyce Harper et al, 'Adjuncts in the IVF Laboratory: Where Is the Evidence for "Add-On" Interventions?' (2017) 32(3) *Human Reproduction* 485, 489.

available, implications of a positive or negative result including any medical options available, and the potential need for further testing. Specifically, clinicians and healthcare practitioners should ensure that women making decisions about prenatal testing understand that the future child may have future interests in relation to knowledge of the results of the testing process. This requires NIPT to be provided in a supportive clinical context, as opposed to a DTC context where there may not be sufficient regulation to ensure this clinical obligation is discharged. Careful consideration should be given to whether or not testing for adult-onset conditions is offered to all pregnant women or provided upon request. Whilst the latter may alleviate concerns about 'testing by default', it may lead to testing inequalities based on education and access to information.

Fourthly, a supportive regulatory framework would require further education of clinicians and healthcare practitioners to provide appropriate and timely counselling to prospective parents seeking NIPT. Counselling should be non-directive and inform parents of the implications of testing and termination or continuing with a pregnancy. Counselling should also highlight ways in which harm to a future child may be mitigated, for example, by encouraging parents to not disclose results of a test to third parties, so as to protect the privacy and autonomy of the future child. Further empirical evidence would be required to determine the best approach for parents to take in terms of disclosing information to the child.

Fifthly, given the lack of empirical evidence around harms to the future child, reporting and recording requirements should be imposed to enable further examination of patient outcomes longitudinally. It would be valuable to capture data on how many people choose to test and the reasons for testing as well as the long-term implications of testing for the patient and child. This data would assist in developing an evidence base in response to concerns raised about harms to the future child.

Finally, in conjunction with specific guidelines on NIPT, an independent review of current insurance and employment laws could shed light on the reforms necessary to prevent discrimination against individuals based on genetic information.

V Conclusion

With whole-genome sequencing of a fetus on the horizon, the potential impact of NIPT on the future child must be evaluated and addressed. Convenient and accurate prenatal testing has the potential to enhance reproductive choice by providing parents with genetic information about their future child. Although

NIPT is currently routinely used in private healthcare settings to test for certain genetic conditions, such as Down Syndrome, the scope for broader testing raises new and challenging issues. Testing for adult-onset conditions raises specific concerns about the welfare of the future child, potentially interfering with the child's future autonomy and opportunities. Extending the scope of prenatal testing also has broader implications for society as a whole. This article highlights the complex interrelationship between the interests of prospective parents and those of their future children and proposes a facilitative approach to regulating NIPT for adult-onset conditions. It proposes a flexible approach for determining an appropriate 'threshold' for NIPT in particular contexts. Subject to any clearly substantiated threshold of 'seriousness', the regulatory framework mapped out in this article could be applied more broadly and tailored to other controversial applications of NIPT in the future, including non-medical sex selection and polygenic disease susceptibilities to diabetes and mental illness.