

Don't forget the legal framework: the public provision of non-invasive prenatal testing in England & Wales

Author: Jeffrey I Wale

Attribution: Department of Law, Bournemouth University, Talbot Campus, Christchurch House, Fern Barrow, Poole, Dorset, UK, BH12 5BB

Author/correspondence address: Jeffrey Wale, Department of Law, Bournemouth University, Talbot Campus, Christchurch House, Fern Barrow, Poole, Dorset, UK, BH12 5BB; Telephone 01202 962245; Email: jwale@bournemouth.ac.uk

Abstract

If the central purpose of non-invasive prenatal testing (NIPT) is to deliver improved reproductive autonomy - by facilitating enhanced choice between the continuation and termination of pregnancy - any public funded regime ought to be compatible with the legal grounds for decision-making in this context. It is problematic for a State to facilitate access to information that would or is likely to result in legal dilemmas and conceptual issues for those using these tests. Public funded testing should not be available for purposes that would be or are likely to be 'incompatible' with any framework for lawful abortion. In England and Wales, those incompatible purposes currently include the identification of: (1) gender in the absence of other genetic factors, and (2) specific abnormality that is unlikely to yield serious disability at birth. Consequently, expansion of the NIPT regime to include these purposes should entail changes in the abortion legal framework.

Introduction

Perhaps it is easiest to start by telling you what this paper is not about. It is not going to debate the ethical case for or against prenatal testing or diagnosis. Nor does it seek to examine the general ethical case for or against public/ private funding of any particular type of screening regime. What this paper seeks to do is examine the backdrop to prenatal screening and non-invasive prenatal testing (NIPT), and the particular purposes that a State might have in this context. It goes on to argue that there is a close connection between these testing regimes and the legal/ regulatory framework for abortion. As such, States ought to have regard to that connection when designing, facilitating and financing any prenatal testing regime.

This paper takes as its fulcrum the *UK RAPID* evaluation study. This study was created with input from the UK National Screening Committee (UK NSC) and funded by the National Institute for Health Research to evaluate the use of NIPT for Down's syndrome. NIPT is already available for this condition through the private sector in the UK¹ and a decision will have to be made shortly by the UK NSC on whether these tests should be funded publicly through the NHS. It is argued that NIPT for Downs is compatible with the current legal regime in England and Wales providing it is kept within defined limits. However, the conclusions caution against expansion of publicly funded NIPT without wider consideration and reform of the current abortion legal framework.² This short paper is written in five parts. Part 1 examines the UK model for prenatal screening and specifically non-invasive prenatal diagnosis (NIPD) and NIPT. Part 2 goes onto consider the aims and purposes of these regimes. Part 3 addresses why the

purposes and scope of any testing regime need to be consistent with the legal and regulatory framework for abortion. Part 4 considers what such a conclusion means in practice and, for simplicity, analysis is restricted to the legal and regulatory framework in England and Wales. This approach is taken because slightly different legal rules apply to abortion in other parts of the UK.³ And finally, part 5 concludes by looking at what this means for the future development and funding of pre natal testing in that jurisdiction.⁴

1 The UK Model for Non-Invasive Prenatal Testing

The UK already operates a fairly comprehensive prenatal screening programme. This includes a combination of maternal blood tests, ultrasound scans and invasive diagnostic testing (notably Chorionic Villus Sampling (CVS) and Amniocentesis). The discovery of cell-free fetal DNA (cffDNA) in maternal blood made it possible to undertake additional, and as we shall see, potentially more accurate and reliable tests that present minimal risk to mother and fetus.⁵ A distinction is made between the analysis of cell-free fetal DNA where the outcome can be diagnostic (NIPD)⁶ and where further tests are required to achieve this outcome (NIPT).⁷ However, NIPT is proving to be very accurate in the context of Down's syndrome and the detection of trisomy 21⁸. Some studies have shown a detection rate for this trisomy in excess of 99.5% and a false positive rate of 0.1%.⁹ Consequently, these tests are proving to be significantly more reliable than the current combined tests (maternal age, maternal serum biomarkers and ultrasound markers).¹⁰ NIPT also enables screening for other forms of aneuploidy (ie where there is an abnormal number of chromosomes) – specifically trisomy 13

(Patau's syndrome) and 18 (Edward's syndrome) - and can be undertaken earlier than conventional testing. This test combination should hopefully limit the number of women having to undertake unnecessary invasive diagnostic tests and the consequential exposure to the increased risk of miscarriage (0.5-1%)¹¹. In the private sector, NIPT is available to parents to screen for trisomies 13, 18 and 21 but with the option to test for gender.¹²

The *RAPID* study evaluated NIPT for Down's syndrome for the UK NSC before adoption within the NHS¹³ and was led by Professor Lyn Chitty and her team at Great Ormond Street Hospital. The study offered NIPT for Downs as a contingent test – pregnant women underwent conventional combined screening, and, only if their risk was identified as greater than 1: 1000, were they offered NIPT prior to invasive testing.¹⁴ The study used UK based laboratories to undertake the DNA analysis whereas many of the private sector providers continue to utilise overseas analytical services. The location of these services may be an important consideration because it might impact on the future access and security arrangements for this genetic data. The *RAPID* study reported to UK NSC in May 2015 and endorsed the contingent use of NIPT within the NHS Down's syndrome screening pathway.¹⁵ The UK NSC has still not made a final decision and evaluation of NIPT remains an ongoing project.¹⁶

Whilst the UK appears to be moving to a restricted and contingent based model for NIPT, there is no single world view on that model.¹⁷ Some academics have predicted universal population testing in the future¹⁸ - there is already proof of concept for the analysis and mapping of the whole human genome.¹⁹ There is and will be the scope to test for non-health related factors and trivial information²⁰ and the technical possibilities are only likely to increase over time.

However, it remains to be seen whether parents will be able to understand and process a much wider (and possibly uncertain) range of data about the fetus and any future child it might become.

A distinction has sometimes been made between testing for specific diseases or rhesus status (conditions) and testing for fetal anomalies.²¹ The argument is that detection of the former can improve outcomes for the pregnancy/fetus; whereas detection of the latter only results in a choice between the continuation and termination of the pregnancy. However, it is possible that detection of anomalies could provide parents with the opportunity to prepare psychologically and physically for parenthood of a child with disability. As De Jong & De Wert point out it may also be difficult to distinguish between these types of tests in practice²² as maternal blood may be subject to analysis for a number of distinct purposes (e.g. for the identification of Down's syndrome and fetal sex).

2 What is the aim and purpose of NIPT?

The RAPID study situated general prenatal screening and diagnosis as: '*a routine part of antenatal care*'.²³ A number of possible competing purposes have been highlighted for these tests depending on the information that is being looked for and the 'end' to which that information is to be put. So prenatal screening could involve looking for the serious or trivial, for the health related or other information. In terms of ultimate '*ends*', prenatal screening has tended to be categorised as either a process that improves public health or as one that facilitates/enhances reproductive autonomy. The question of '*means*' and '*ends*' are closely connected in this debate – for example, whether we are looking to act upon, ameliorate, cure or prevent a particular state of affairs. We can see this in action in the context of rhesus status. The rhesus status of a pregnant woman and her unborn baby are important because of the potential for

antibodies to cross the placenta and cause health problems for the developing child.²⁴ So early rhesus testing can guide clinical treatment and help to improve pregnancy outcomes. The difficulty comes when we look at screening for fetal abnormalities and non-health related factors.

The stated aim of the RAPID evaluation study on Down's syndrome was:

*'to develop better and safer ways of detecting Down's syndrome and some other chromosomal conditions in pregnancy.'*²⁵

Clearly detection of these syndromes has a purpose. The NHS website emphasizes that Down's screening is about enhancing and facilitating parental choice²⁶ albeit with limited options:

*'A small number of women who have a diagnostic test will find out their baby has Down's, Edwards' or Patau's syndrome. They then have **two** options.*

*Some women decide to continue with the pregnancy and prepare for their child with the condition; others decide they do not want to continue with the pregnancy and have a termination (abortion).'*²⁷

There appears to have been greater emphasis on the role of enhancing and/or enabling maternal and general parental autonomy in relation to anomaly screening. In this context, De Jong and De Wert comment:

*'The traditional aim of population screening (health gains won through timely treatment or prevention) does not readily apply to this type of prenatal screening...instead, prenatal screening for foetal abnormalities is generally understood as aiming at offering pregnant women (and their partners) options for reproductive choice.'*²⁸

The focus on autonomy is understandable particularly where there are no remedial or preventative options available to the parents and clinicians. There is also a patent desire to avoid any discussion about eugenics and the explicit improvement of public health through the removal of unwanted disability.²⁹ However there are also reasons why we may want to qualify the autonomy view of this form of prenatal testing.³⁰ A State may have legitimate reasons not to support³¹ unfettered reproductive choice if it could send an explicit discriminatory message about disability. Wilkinson argues there has to be limits placed on the role of autonomy because otherwise it would be untenable for the pro-choice camp to argue for any restrictions on testing.³² Further, when it comes to the issue of public funding, unrestricted parental reproductive autonomy does not easily fit with the concept of distributive justice.³³ Christian Munthe highlights the significant economic consequences that could flow from the public funding of open choice, and in particular, the expense of providing an adaptable test that meets patient wishes and at the same time ensures sufficient standards of counseling.³⁴

Public health considerations may also have an explicit role to play in the context of prenatal testing for fetal abnormality.³⁵ Reliable non-invasive testing methods can reduce adverse pregnancy outcomes by limiting the number of unnecessary invasive tests and the accompanying risks and parental anxiety associated with these tests. Advance knowledge may help prepare families psychologically and physically for the birth of a child with disability.³⁶ The ability to identify disability or conditions accurately could have a role to play in public health planning and the distribution of future State resources. The potential benefits of a publicly funded scheme might include central co-ordination and greater quality control over testing. Specific public funding of NIPT would also remove the current inequity for those who cannot afford the expense of private testing.³⁷ However, when it comes to trivial and/or non-medical reasons for NIPT,

there seems to be a much weaker public health rationale.³⁸ Even if it can be argued that there is a moral right to access this type of information about a future child, it does not follow that the State should fund all stages of a process that would convert this into a meaningful legal right.³⁹

3 Why aims and purposes matter

The central argument of this paper is that the purposes/aims of any prenatal testing regime need to be consistent with and correlate to the wider regulatory/legal framework in which that regime operates. A State should act coherently and consistently in the promotion of health and reproductive autonomy. Inconsistency would send confusing messages to the public and prenatal testing arrangements need to operate within the formal legal frameworks that have been agreed upon and not simply constructed by the Executive as they see fit. If reproductive freedom is the central aim of a publicly funded NIPT regime for fetal abnormality, the State has to be clear and transparent about how these ends are to be realized. If the only plausible way to exercise choice in the event of detection is to terminate or continue the pregnancy, that needs to be made clear and a State can only plausibly facilitate reproductive choice if there are lawful options for the parents. To be even more explicit, there ought to be consistency between the choice presented or highlighted by NIPT and the legal/ regulatory framework in which reproductive choices and related clinical decisions are made.⁴⁰ Further, public funding of information should not facilitate or encourage, either directly or indirectly, choices that are inconsistent with other public goals, policies and laws.⁴¹ Wilkinson argues that supporters of prenatal screening must apply their view consistently:

'If we are talking about state funding and support then consistency requires proponents of the Pure Choice view to go further and to advocate not merely the legal permissibility of sex-selective abortion, but the state funding of this as well'.⁴²

It also means that pro-choice supporters need to align any publicly funded NIPT regime with the legal framework for abortion in that jurisdiction. If the law restricts (as it does) certain types of abortion and reproductive choice,⁴³ consistency demands that there should be some limitation on publicly funded prenatal screening where the stated purpose is enabling/ enhancing particular reproductive choices. Of course, this argument rests on a stark presentation of choice,⁴⁴ and as discussed, the provision of information about anomaly or disability may very well provide parents with an opportunity to prepare for their future child and for any diagnosed conditions upon birth.⁴⁵ It also assumes that a choice exists at all – for some, the legal option to terminate may not provide an acceptable moral option because of their own personal values and beliefs. Despite these qualifications, it seems to me that there still ought to be some degree of convergence between the two frameworks if reproductive choice is the central public rationale for NIPT. There may be those that argue that the State should facilitate and fund reproductive freedom in relation to lawful options (eg to terminate) available in other jurisdictions.⁴⁶ Again there appears to be the risk of incoherent public narratives and State complicity in what would otherwise be an unlawful procedure in the home jurisdiction.

It has already been argued that the aim of reproductive choice may have to be qualified and there may be plausible public health aims for NIPT. However, the aim of improving public health sits uncomfortably alongside non-health related testing and a legal abortion framework that explicitly works on a medical model. It has been argued that the reality of the Abortion Act 1967 is that abortions are available on demand, at least until the 24th week of pregnancy.⁴⁷ This is because

Section 1(1) (a) of that Act includes a social ground for abortion that concentrates on the comparative risk to the physical or mental health of the pregnant woman or any existing children of her family. However, whilst the legal provisions explicitly adopt a medical model for abortion, public perception must surely play a role. Again, is there not a risk that State funded testing for trivial and non health related information could confuse public understanding and damage respect for the law? To be explicit, it might send mixed messages if a State were to fund testing with the purpose of facilitating a choice that it otherwise restricts through other legal means (eg facilitating a choice to terminate a pregnancy for trivial or non-health related reasons). A State should not seek to normalize choices that it does not legislate for and a better solution seems to be to work towards some convergence in the two frameworks. In practical terms, that may only be realized through amended or new legislation. Further, even if a State provides lawful options to terminate pregnancy, it does not follow that those options should be encouraged via prenatal testing or otherwise through unlimited public funding.⁴⁸ Once again we come back to the need for clear public messages and understanding about the central purposes of any testing regime.

Finally we come to the argument that parents should have the right to know.⁴⁹ Even if there should be an unfettered parental right to access full genetic information about their future child, it does not automatically follow that the State has an obligation to fund that right. If the purpose of testing is to enable access to information in furtherance of such a right, and not to facilitate choice, why should the State fund that provision without clear evidence of public benefit? The evidence of public benefit in relation to non-health related information appears to be weak and far from compelling.

4 The practical impact in England & Wales⁵⁰

The Abortion Act 1967 (AA 1967) does not explicitly acknowledge or provide a parental/maternal right to terminate a pregnancy – there remains medical control over the decision even if the public perception is of abortion on demand.⁵¹ So the starting point is that the law does not provide unfettered parental choice to terminate a pregnancy in England & Wales. In the context of NIPT, the most relevant legal provisions are sections 1(1)(a) and 1(1)(d) of the Abortion Act. The former section⁵² provides the following lawful ground for termination:

‘That the pregnancy has not exceeded its 24th week and that the continuance of the pregnancy would involve risk, greater than if the pregnancy were terminated, of injury to the physical or mental health of the pregnant woman or any existing children of her family’.

It has been argued that this provides the lawful basis for termination for inconvenience or sex selection on the basis that the continuation of the pregnancy always presents a greater risk than termination.⁵³ However, that view seems flawed – there may be circumstances where a termination could present a greater risk than continuing the pregnancy,⁵⁴ and in any event, the professional regulators and the Department of Health (DOH) stipulate individual medical assessment in every case.⁵⁵ Further, the DOH has made it abundantly clear that termination on the sole grounds of gender is unlawful:

‘Abortion on the grounds of gender alone is illegal. Gender is not itself a lawful ground under the Abortion Act (see Annex A for the lawful grounds under Section 1(1)). However, it is lawful to abort a fetus where two RMPs⁵⁶ are of the opinion, formed in good faith, “that there is a substantial risk that if the child were born it would suffer from such physical or mental

*abnormalities as to be seriously handicapped”, and some serious conditions are known to be gender-related.*⁵⁷

That takes us neatly onto the lawful ground in section 1(1)(d):

*‘that there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped’.*⁵⁸

Now this provision has considerable scope for uncertainty both in terms of assessing the likelihood of risk and establishing its core purpose.⁵⁹ Indeed, Sheelagh McGuinness has suggested that in practice this section may be ‘operating beyond any plausible legitimate interpretation of the ground’.⁶⁰ She also highlights the presumptive effect that this section could have on parental choices following prenatal screening and this is important if the options presented are not lawful ones.⁶¹ ‘Substantial risk’ is not defined in this section - plainly it does not require a certainty but the degree of probability required is not clear. The phrase *‘would suffer from such physical or mental abnormalities’* is essentially a medical question. Down’s syndrome appears to fit this test but what about a HIV positive fetus that may be asymptomatic at birth?⁶² Andrew Grubb concludes that HIV infection could be construed as a physical abnormality under this subsection⁶³ but questions whether any future child with that condition would suffer from abnormalities so *‘as to be seriously handicapped’*. The section links abnormality with a certain (serious) degree of handicap/ disability. Down’s syndrome may fit this requirement but there must be uncertainty whether the subsection applies to a fetus that only has a high likelihood of the future risk of handicap (eg Huntington’s Chorea) or where there is only a possible future risk of handicap (as in the HIV example).⁶⁴

Let us put to one side the lengthy discussion we could have about the nature and assessment of disability.⁶⁵ What is reasonably clear is that the regulatory framework in England and Wales does not explicitly authorise the termination of pregnancy where gender or trivial non-medical reasons are the sole criterion for decision-making. There may be some equivocation because of the effects of section 1(1)(a) but again there is no explicit State endorsement of terminations based solely on minor abnormality or possible future disability. Whilst these legal restrictions remain in place it would be illogical and inconsistent for the State to fund a system of explicit prenatal testing for gender, minor abnormality or uncertain/future disability. Nor is it clear that an advance system of legal assurances from pregnant women *'that they will not seek to use the information gained from the test for terminating pregnancy on the grounds of fetal sex'*⁶⁶ would obviate that concern and indeed, may prove unworkable and unenforceable in practice in any event.

The *RAPID* evaluation study on Downs appears to align NIPT with the current regulatory framework for abortion and so the difficulty will only arise if we look to expand the categories for genetic analysis. As De Jong et al. have noted:

'there is a tendency to widen the scope of testing in the context of prenatal screening for foetal abnormalities....As soon as NIPT can be affordably and reliably used for screening beyond the major trisomies, a further widening can be expected'.⁶⁷

Of course, there could be other reasons to resist expansion of any testing regime both in terms of scope and category of condition. For example, there would need to be careful consideration as to whether expansion would increase the availability of uncertain information and unnecessarily

complicate counseling and decision-making in this context.⁶⁸ Thought would also have to be given as to the possible rights of the future child and whether it ought to encompass a right ‘not to know’ about genetic data obtained from such testing.⁶⁹

Finally and for completeness, I should also mention the legal framework governing the use of pre-implantation genetic screening (PGS) and pre-implantation genetic diagnosis (PGD). In summary, PGS is pre-implantation embryo testing for aneuploidies and PGD is pre-implantation embryo screening for specific gene mutations and diseases. The Human Fertilisation and Embryology Act 1990⁷⁰ makes it permissible (subject to licence) to test the ex vivo embryo for certain abnormalities that might affect its capacity to result in a live birth and to avoid serious medical conditions.⁷¹ Importantly, the legal framework does not explicitly permit embryo testing/selection on the basis of gender alone – instead the emphasis is on avoiding the risk of a genetic condition related to sex.⁷² In any event, the choice is technically different to that presented to parents post NIPT - with PGS/PGD the option is whether to implant the embryo or not.

5 Conclusions

Although NIPT appears to bring many benefits, there does need to be clear and consistent public narrative in relation to the promotion and facilitation of parental choice in relation to any testing regime. If the facilitation of parental choice is a core purpose of testing, States should make that explicit and exercise caution before expanding the public funding and provision of NIPT without wider consideration of the connected regulatory frameworks. Indeed, this paper goes further

and argues that publicly funded provision of NIPT ought not to be made available for any purposes that would be or are likely to be incompatible with any framework for lawful abortion. In England and Wales, those ‘incompatible purposes’ currently include the primary identification of (1) gender in the absence of related genetic factors and (2) specific abnormalities that are unlikely to result in serious disability at birth.⁷³ Of course, Parliament is at liberty to change the abortion framework and expand NIPT but it is important that changes are not made in isolation. Further, if enhancement of parental choice is a core aim of screening, Parliament should also be explicit about the priority of and mechanism for ensuring choice in the abortion framework.

That still leaves two issues for consideration. First, what should a State do about the incidental availability of genetic data following the collection of blood samples in the public health system?⁷⁴ Even if testing is carried out for a specific funded purpose, the blood samples may be capable of yielding other genetic data (including trivial information) outside that purpose. Maternal access to incidental blood data probably cannot be resisted on legal grounds unless there are strong therapeutic reasons for withholding that information. However, there appear to be stronger arguments for not facilitating analysis of incidental data via additional public funding.⁷⁵ Certainly the public facilitation of such analysis (before and after birth) merits further consideration.

Secondly, even if public funding is limited to contingent and restricted options,⁷⁶ consideration ought to be given to the potential impact on abortion services. So for example, will NIPT have any material effect on the demand for abortions and the balance between private and public sector abortion provision? How might the availability of new or different data impact on the complexity of parental decision-making during the pregnancy? It does not automatically follow that the provision of more information to parents will necessarily result in better choices and

pregnancy outcomes. Careful consideration will need to be given as to how parents are supported and whether the State should be funding arrangements for this process.

Funding: This research received no specific grant from any funding agency in the public, commercial or not-for-profit sectors.

Declaration of conflicting interests: The author declares that there is no conflict of interest.

References and Notes

¹ And has been available since 2012

² I do not specifically explore the need for compatibility with other legal frameworks (eg the Human Fertilisation & Embryology Act 1990 (as amended) or the Equality Act 2010).

³ The Abortion Act 1967 does apply in Scotland but the criminal law framework is different from England and Wales. The Abortion Act 1967 does not apply to Northern Ireland.

⁴ This paper does not attempt to endorse the current or any particular regulatory structure for abortion. Rather it is argued that States should act in a coherent and compatible fashion in the context of abortion and prenatal testing. This includes a requirement to disseminate the aims of any testing regime in a transparent, coherent and understandable form

⁵ A De Jong, I Maya & J M.M. Van Lith, 'Prenatal Screening: Current Practice, New Developments, Ethical Challenges', *Bioethics*, XXIX No1 (2015) pp 3-6

⁶ Currently NIPD is available in the NHS for fetal sex determination where there are clinical indications for a sex related genetic condition & for certain single gene disorders. See <http://www.labs.gosh.nhs.uk/laboratory-services/genetics> (accessed 23 March 2016)

⁷ As per Down's, Edward's & Patau's syndromes

⁸ Down's syndrome is associated with physical and intellectual disability and trisomy 21 denotes the presence of a 3rd copy (or part thereof) of chromosome 21.

⁹ http://obstetricsgynecology.eu/nipt-fetal-dna-maternal-blood?utm_source=email&utm_medium=email&utm_content=button&utm_campaign=May%20update (accessed 23 March 2016). It should be noted that the UK RAPID study originally evaluated the reliability factor at 99% and the risk of false positives at 0.5-1% (see <http://www.rapid.nhs.uk/wp-content/uploads/2014/01/NIPT-study-Participant-information.pdf> (accessed 23 March 2016)

¹⁰ De Jong et al, 'Prenatal Screening', pp3 & 6

¹¹ R Akolekar et al., 2014, 'Procedure-Related Risk of Miscarriage Following Amniocentesis and Chorionic Villus Sampling: A Systematic Review and Meta-Analysis', *Ultrasound Obstet Gynecol*, (2014) DOI 10.1002/uog.14636

¹² See for example: <http://www.ultrasound-direct.com/babybond-pregnancy-scans/nipt-scan/> (accessed 23 March 2016)

¹³ <http://www.rapid.nhs.uk/about-rapid/evaluation-study-nipt-for-down-syndrome/> (accessed 23 March 2016)

¹⁴ See <http://www.rapid.nhs.uk/about-rapid/evaluation-study-nipt-for-down-syndrome/> (accessed 23 March 2016).

¹⁵ L Chitty et al., 'RAPID Non-Invasive Prenatal Testing (NIPT) Evaluation Study: Executive Summary', May 2015 (ISBN 978-1-907198-17-5)

¹⁶ <http://legacy.screening.nhs.uk/fetalanomalies> (accessed 23 March 2016)

¹⁷ A recent summary of the German regime can be found in R Wegner et al., 'Non-invasive Prenatal Diagnosis Using Massively Parallel Sequencing – First Experience in Germany', *Molecular Cytogenetics*, VII (Supp1) (2014), I14

¹⁸ De Jong et al, 'Prenatal Screening', p 8

¹⁹ J O Kitzman et al, 2012, 'Noninvasive Whole-Genome Sequencing of a Human Foetus', *Science Transl Med*, IV (2012), pp137ra 76

²⁰ For example, hair colour

²¹ De Jong et al, 'Prenatal Screening', p 2

²² A De Jong & G De Wert, 'Prenatal Screening: An Ethical Agenda For The Near Future', *Bioethics*, XXIX no1 (2015), p 47

²³ <http://www.rapid.nhs.uk/about-rapid/background> (accessed 23 March 2016)

²⁴ <http://www.nhs.uk/Conditions/Rhesus-disease/Pages/Causes.aspx> (accessed 11 March 2016)

²⁵ <http://www.rapid.nhs.uk/wp-content/uploads/2014/01/NIPT-study-Participant-information.pdf> (accessed 23 March 2016)

²⁶ <http://www.nhs.uk/conditions/pregnancy-and-baby/pages/screening-amniocentesis-downs-syndrome.aspx> (accessed 23 March 2016)

²⁷ NHS, Op.cit

²⁸ De Jong & De Wert, 'An Ethical Agenda', p 48

-
- ²⁹ See the discussion by De Jong & De Wert (Op. cit, pp 48-49) on the disability rights critique of pre natal screening and their view that this can only be defended 'if it is clear that such screening is not offered in order to achieve population-level health and economic benefits'. See also Stephen Wilkinson, 'Prenatal Screening, Reproductive Choice, And Public Health', *Bioethics*, XXIX No1 (2015), pp27-29
- ³⁰ De Jong & De Wert, 'An Ethical Agenda' p 48
- ³¹ Directly or indirectly
- ³² De Jong & De Wert, 'An Ethical Agenda', pp 48-49 and Wilkinson 'Prenatal Screening', pp 30-31
- ³³ De Jong & De Wert, 'An Ethical Agenda', p 49
- ³⁴ C Munthe, 'A New Ethical Landscape Of Prenatal Testing: Individualizing Choice To Serve Autonomy And Promote Public Health: A Radical Proposal', *Bioethics*, XXIX No1 (2015), p43
- ³⁵ De Jong & De Wert argue that screening for anomaly 'should be regarded as belonging to the responsibility of public health authorities' ('An Ethical Agenda', p50). Compare Munthe who argues that the private health/commercial sector has a role to play in this context ('A New Ethical Landscape', p 44). See also Wilkinson's discussion on the role of public health pluralism in prenatal screening ('Prenatal Screening', pp26-35).
- ³⁶ B Dickens, 'Ethical and Legal Aspects of Noninvasive Prenatal Genetic Diagnosis', *J of Gyne & Obste C*, XXIV (2014), p184
- ³⁷ I am grateful to an anonymous reviewer for highlighting some of the potential benefits of a publicly funded testing regime.
- ³⁸ For example, the detection of fetal gender or hair colour unrelated to disease/abnormality
- ³⁹ Including access, analysis and provision of that information to the parents.
- ⁴⁰ See Caroline Wright, *Cell-Free Fetal Nucleic Acids for Non-invasive Prenatal Diagnosis: Report of the UK Expert Group* (Cambridge, PHG Foundation, 2009), at p19 where she highlights the difficulty for an individual that wants to terminate a pregnancy on the basis of test results but cannot do so lawfully.
- ⁴¹ Particularly if there is a potential danger that NIPT might be used to justify abortions for which there are no independent lawful grounds (See Alison Hall, Adam Bostanci & Stephen John, 'Ethical, Legal & Social Issues Arising from Cell-Free Fetal DNA Technologies' (Cambridge, PHG Foundation, 2008) in Wright, Op.cit, appendix III)
- ⁴² Wilkinson, 'Prenatal Screening', p 31
- ⁴³ For example, abortion selection solely on the grounds of trivial abnormality or gender
- ⁴⁴ Between termination or continuation of pregnancy
- ⁴⁵ Dickens, 'Ethical & Legal Aspects', p184
- ⁴⁶ Cf the recent decision in *A & B v Secretary of State for Health* [2015] EWCA Civ 771
- ⁴⁷ By virtue of S1(1)(a) Abortion Act 1967
- ⁴⁸ It may be lawful to have cosmetic surgery but public funding is frequently restricted in relation to such treatments.
- ⁴⁹ For a discussion see Z Deans, A J Clarke & A J. Newson, 'For Your Interest? The Ethical Acceptability of Using Non-invasive Prenatal Testing to Test "Purely for Information"', *Bioethics*, XXIX (2015), pp 19-25
- ⁵⁰ For the reasons already outlined it easier to address the legal framework in England and Wales than across the whole of the UK.
- ⁵¹ Jonathan Montgomery, *Health Care Law* (Oxford, OUP, 2002), part 15. See also E Jackson, 'Abortion, Autonomy and Prenatal Diagnosis', *Social Legal Studies*, XXIX (December 2000), pp 470-471
- ⁵² Often described as the social ground
- ⁵³ See for example Jackson, 'Abortion, Autonomy and Prenatal Diagnosis', p470
- ⁵⁴ Marc Stauch & Kay Wheat, *Text, Cases & Materials on Medical Law & Ethics* (London, Routledge, 2015), p396
- ⁵⁵ Department of Health, *Guidance in Relation to the Requirements of the Abortion Act 1967* (London, HMSO, 2014); para 11 at p6; see also Jackson, 'Abortion, Autonomy and Prenatal Diagnosis', p 471
- ⁵⁶ Registered Medical Practitioners (ie Doctors)
- ⁵⁷ DOH 2014, Op, cit, para 25 at p10. CF the provisions on in-vitro embryo selection (see schedule 2, para 1ZA of the Human Fertilisation & Embryology Act 1990 (as amended))
- ⁵⁸ Abortion Act 1967
- ⁵⁹ Is this provision for the benefit of the mother/ her family, the fetus or the future child it might become?
- ⁶⁰ S McGuiness, 'Law, Reproduction, and Disability: Fatally 'Handicapped'?', *Medical Law Review*, XXI (2013) pp 213-242 at p 227
- ⁶¹ McGuiness, Op.cit, p 213
- ⁶² A Grubb, 'The New Law of Abortion: Clarification or Ambiguity?' *Criminal Law Review*, [1991], p 661
- ⁶³ Grubb, Op.cit, pp 659-670

⁶⁴ Royal College of Obstetricians & Gynaecologists (RCOG), *Termination of Pregnancy for Fetal Abnormality* (London, RCOG, 2010), p 10

⁶⁵ For example, whether we ought to assess from a certain perspective or taking into account the social or medical options to ameliorate any disability or condition. For a discussion of these issues, see McGuiness, 'Law, Reproduction, and Disability: Fatally 'Handicapped'?', pp 213-242

⁶⁶ Hall, Bostanci & John, 'Ethical, Legal & Social Issues', p37

⁶⁷ De Jong et al., 'Prenatal Screening', p 8

⁶⁸ A De Jong, W J Dondorp, C EM de Die-Smulders, S GM Frints & G MWR de Wert, 'Noninvasive Prenatal Testing: Ethical Issues Explained', *Eur J of Human Genetics*, XVIII (2010), p 274

⁶⁹ De Jong et al. 2010, Op.cit, p 275

⁷⁰ The Act applies across the UK

⁷¹ Schedule 2 para 1ZA

⁷² Schedule 2 para 1ZA, Op Cit

⁷³ So this would preclude publicly funded testing for adult onset disabling conditions, genetic abnormalities that may never manifest in disability or only manifest as minor disability in childhood.

⁷⁴ Ie Information that is available as a bi-product of collecting other genetic data through cell free fetal DNA analysis

⁷⁵ These might include the compatibility and convergence arguments already discussed and issues of distributive justice etc.

⁷⁶ Ie NIPT in conjunction with conventional screening and invasive diagnostic tests