



# Non-Invasive Prenatal Testing

By Philippa Taylor

**Non-invasive prenatal testing (NIPT) is a technique used to screen fetuses for genetic conditions and variations. It is 'non-invasive' because it involves taking a blood sample from a pregnant woman and analysing the cell free fetal DNA that is circulating in the mother's blood. Direct 'invasion' of the fetus, and the consequent risk of miscarriage, are both avoided. NIPT is more accurate than other prenatal screening tests.**

**N** IPT is most commonly used to diagnose fetuses for trisomies such as Down's, Edwards' and Patau's syndromes,<sup>5</sup> as well as cystic fibrosis and achondroplasia in women where there is a family history or other indication. Some private healthcare providers offer NIPT for other genetic conditions, and also to find out fetal sex. For example, the IONA test developed by Premaitha Health has been developed to include 'a number of additional product and software features such as sex determination'.<sup>6</sup> Premaitha Health is planning to cover other prenatal conditions and cancer detection in the next few years. These private tests cost between £400 and £900, depending on what is included.

The developed world is moving towards wide scale adoption of NIPT, whether through private providers and/or government-funded healthcare provision. We need to consider the consequences of its increasing use, its impact on the way we view pregnancy, disability and difference and how Christians might respond.

## PRACTICAL ISSUES

### Increase in terminations

Some argue that NIPT is effectively an

### NHS Screening Policy<sup>1</sup>

NHS policy is to offer all women a prenatal screening test – the 'combined test' – for Down's, Edwards' and Patau's syndromes between ten and 14 weeks of pregnancy as part of the NHS fetal anomaly screening programme. This involves a blood test and an ultrasound scan. If a woman is too far into her pregnancy to have the combined test she will be offered other tests. For Down's Syndrome, a blood test called the 'quadruple test' is available from weeks 14–20 of pregnancy. For Patau's and Edwards' syndromes, a mid-pregnancy scan can check for physical abnormalities.

Diagnostic testing using chorion villus sampling (CVS) or amniocentesis is then offered as standard practice to women who through the screening have been found to have a high chance of having a fetus with one of these syndromes. These tests carry a small risk of miscarriage.<sup>2</sup>

However, where NIPT is available, pregnant women who are found to have a high chance of their fetus having Down's, Edwards' or Patau's syndromes after having the 'combined test' are offered NIPT, generally from around nine or ten weeks gestation. If the result is positive, then an amniocentesis or CVS is offered to confirm the condition.<sup>3</sup> The expectation is that with NIPT fewer women will proceed to the more invasive diagnostic testing.<sup>4</sup>

extension of the prenatal screening tests currently offered as standard practice, but with the additional benefits that it is more accurate and therefore leads to fewer women needing the more invasive tests that carry a risk of miscarriage. Thus, implementing NIPT would save lives because the number of (healthy) babies lost through miscarriage would fall dramatically.

A review of international research studies has found that NIPT identifies 97% of fetuses with Down's Syndrome (DS).<sup>7</sup> In the UK, statistical predictions have calculated that NIPT would result in around 3,368 fewer invasive tests and therefore an estimated 17 fewer miscarriages per year. However the same predictions also calculate that 195 more fetuses with DS would be detected each year.<sup>8</sup> Since the proportion of women having a termination after a diagnosis of DS ranges from 89–95% this would result in around 180 babies with DS being aborted each year than is currently the case.<sup>9</sup> These are statistical predictions but we know that in Iceland, where NIPT is widely implemented, *all* babies with DS are now aborted.<sup>10</sup>

A disability campaign group, 'Stop Discriminating Down' warns about the introduction of NIPT: 'With selective termination rates following prenatal diagnosis already higher than 90% in these

countries, the result will be catastrophic.'<sup>11</sup>

The belief that parents and families will be damaged by having a disabled child, and that this damage can be limited through abortion, is almost unquestioned today. However, psychological morbidity for mothers following abortion for disability is considerable and associated with long-lasting consequences for a substantial number of women.<sup>12</sup> Rather than leading to psychological wellbeing, abortion for fetal disability is an emotionally traumatic life event that can lead to post-traumatic stress response and intense grief reactions which are still detectable some years later.<sup>13</sup> Research has found that women who terminate pregnancies for fetal anomalies experience grief as intense as those who experience spontaneous perinatal loss.<sup>14</sup>

### Confusing results

While NIPT is marketed as being more accurate (NIPT will identify 97% of fetuses with DS), there remains a risk of false-positive results, which would be followed up with a recommendation to have the invasive tests. It is estimated that one in ten pregnant women who have a high chance of being affected will have a false-positive result from the NIPT test.<sup>15</sup> This will not only generate extra anxiety for parents but,

desperate to minimise risk, many will opt for invasive procedures and as a result increase the number of inadvertent miscarriages of healthy babies.

Moreover, understanding risk of false-positives and negatives is complex and poorly understood, even by those offering NIPT. Some groups of women are more likely than others to have false-positive results, depending on their age and other factors.<sup>16</sup> It is also likely that many women will not fully appreciate that NIPT is not diagnostic – a positive result simply indicates a high *chance* that a fetus has a specific genetic condition and any positive result (including a false-positive result) would be followed up by an offer of CVS or amniocentesis for a more definitive diagnosis. The accuracy of NIPT also depends on the condition being tested for, whether it is a twin or singleton pregnancy, maternal weight and the length of gestation.<sup>17</sup>

### Incremental extension

This is a rapidly moving field. NIPT technology is currently being recommended to screen for a limited number of chromosomal conditions. The same technology, however, is able to detect a wide range of genetic conditions and predispositions, including adult onset conditions, as well as non-medical features such as fetal sex. NIPT can give a near definite diagnosis for a few specific genetic conditions, removing the need for further invasive testing.<sup>18</sup> The *New Scientist* has warned that more prenatal tests for single gene disorders are on the way.<sup>19</sup> Whole genome sequencing using NIPT has already been carried out in a research setting.<sup>20</sup>

Professor Tom Shakespeare, Chair of the Nuffield Council on Bioethics working group on NIPT, has warned that: 'Abortions on the basis of sex appear to be rare in Britain. However, this could change with a new DNA testing method that allows the baby's sex to be revealed to prospective parents much earlier than the standard 18–20 week scan'.<sup>21</sup>

While the NHS may offer free NIPT for a limited number of chromosomal conditions, private NIPT providers are not restricted to these applications. It is likely that over time there will be pressure on government to extend its use to include more conditions.

### Support and information

The impact on the parents, learning of a risk of disability in their unborn child, is life

changing; how it is handled by healthcare professionals can significantly affect the decisions made. Many parents have said that the information they were given by health professionals was negatively biased, uninformed and even incorrect; only occasionally was it described as 'fantastic' or helpful.<sup>22</sup> The Nuffield Council on Bioethics has cited '...concerns that some healthcare professionals, when imparting information about DS, focus on medical problems, such as heart problems, and learning disability, without describing more fully what it is like to have a child with DS. This may influence the decisions women and couples make'.<sup>23</sup> They also cite the influence of society towards disability, the presentation of disability and prenatal testing in the media and the perceived impact of a disabled child on the family.

In reality, disability does not preclude a satisfying life. Most people with DS now live into their 50s and 60s, finding fulfilment and contributing greatly to family and community life. Research published in the *American Journal of Medical Genetics* on children and families affected by disabilities found that nearly 99% of people with DS are happy with their lives, more than three-quarters of parents of a child with DS had a more positive outlook on life and almost 90% of siblings said they considered themselves better people because of their family member with DS.<sup>24</sup>

Many problems attributed to the existence of a disability actually stem from inadequate social arrangements that public health professionals should work to change.<sup>25</sup> A reduction in the number of babies born with certain syndromes may well diminish the specialist health and social care that they receive, as well as the research carried out on these syndromes, with fewer people and groups to fight for support and care for people with the syndromes, and fewer able to offer mutual support to those affected.

### Ethical dilemmas

The fact that a technology such as NIPT can be abused or misused does not make it inherently wrong. The diagnostic test is a morally neutral procedure. Much depends on the *purpose* of the test and the *context* in which it is offered. NIPT could be a benefit if the medical information is used to help prepare parents for the birth of a baby with special needs, for example by undertaking more screening, determining where the

baby should be born and setting up a network of help and support for them.<sup>26</sup> A prenatal diagnosis can give women time to learn about the genetic condition. It may help for the parents to be able to grieve for the child they thought they were going to have, which can allow a greater sense of celebration when the child is born.

Many further argue that the decision to use NIPT should be left to the choice of the parents, along with the personal responsibility about what to do with the results. Society today lays great weight on the importance of freedom of choice, and indeed autonomy is an important concept with Christian origins.<sup>27</sup>

*'Those who do not believe in a particular approach to reproductive choice are free not to use it. We should remember the imbalance in these arguments: when we restrict reproductive-choice options, no one gets to use them; when we make reproductive-choice options available, it's an individual decision whether or not to take up that option.'*<sup>28</sup>

NIPT is already available privately for pregnant women for a range of genetic conditions, but only for a fee. Therefore, it could be argued that it is discriminatory for NIPT only to be available for those who can afford it and NIPT should be freely available for all women who want it. Likewise the range of tests on offer should not be arbitrarily restricted to just some women or some tests.

### The fallacy of autonomy

While the test itself may be morally neutral, the development and context of prenatal screening, including NIPT, is not neutral or value free and does not allow for the exercise of individual procreative autonomy.

NIPT is purely a screening test, it is not a treatment. However if a test is offered as standard procedure, and free of charge, the implication will be that it is a 'good' test to have, and women 'should' ideally have it. However if abortion is the only option on offer this strongly implies that it is socially desirable to prevent the birth of certain babies. This puts pressure on women to proceed with termination and it places a negative value on people with the genetic condition. It thus creates a coercive environment where true choice is constrained.

This is compounded by negative perceptions of the experience of life with disability and rhetoric about the 'prevention

of suffering' and the 'burden of care'. Neonatal Paediatrician John Wyatt argues that there is strong evidence that many professionals overtly or covertly influence the decisions that parents make:

*'Autonomy is a slippery concept. It may appear to have a clear theoretical meaning to the philosopher, but in my experience it is extraordinarily elusive in the realities of the fetal medicine clinic or intensive care unit... the choices of patients are heavily influenced by the exact way in which information is presented and by whom it is given. Patients are extremely vulnerable when presented with devastating news... People vary hugely in their capacity to comprehend the implications of technical information... The truth is that the goal of genuine neutrality in areas as emotive as procreation and abortion is impossible and even inhumane.'*<sup>29</sup>

A further limitation of the parental autonomy argument is that it ignores the rights of the other life directly affected by any decision. It is a choice for the mother but not the fetus.

Although legally a fetus has few rights, *'the absolutist position that the fetus is owed no duties until the moment of birth seems counterintuitive, illogical and hard to defend'*.<sup>30</sup> Since Hippocrates, the practice of medicine in the West has always enshrined moral commitments that include a primary orientation to protect vulnerable life.

As NIPT develops it is likely to be used to predict genetic conditions that have their onset later in life. This would reveal knowledge about the child – without his or her consent – that would deprive the child of the right to choose to know what his/her chances of developing a genetic condition later in life would be. One person's freedom to choose would remove the freedom to choose from another. Indeed, it would have still wider impact because NIPT would in effect reveal familial genetic information about the pregnant woman, her wider family, her partner and his family, that they most likely had not chosen to know.

We cannot ignore the reality that the 'choice' most women take when given a diagnosis of a disability for their unborn child is abortion, so the result of allowing so-called 'freedom of choice' in a coercive context is a significant increase in the number of children with genetic conditions who are eliminated by abortion.

## Discrimination

While the apparent freedom of choice for

some women to have NIPT and a subsequent abortion might be restricted according to income, a far greater discrimination is against the lives of unborn children with genetic conditions. NIPT is promoted as being beneficial for women and society because fewer lives will be lost from miscarriage. However these lives saved are the 'healthy' ones and the many extra lives that would be detected and aborted are those with disabilities. A view that the life of a disabled person, born or unborn, is of less worth than that of fellow human beings is discrimination. NIPT is discriminatory because it deliberately targets genetic disorders and fetal sex.

In the UK, equality laws provide protection for people with disability to ensure they are treated the same as those with no disability – from the moment of birth. Before birth however there is no such protection from disability discrimination.<sup>31</sup> The expansion of NIPT and free abortion stands in stark contrast to the social progress made towards an inclusive and equal society.

Wyatt comments that: *'Social approval of abortion of fetuses with Down's Syndrome could even be seen as "chromosomalism", enshrining social discrimination against certain forms of DNA'*.<sup>32</sup>

## Eugenics and a pursuit of 'perfection'

The Nuffield Council on Bioethics has warned that NIPT will lead to babies being aborted simply because they are the wrong gender or have other 'undesirable' characteristics.<sup>33</sup> Selecting who is born based on their biological or genetic difference, such as is the case with DS for example, enables eugenic attitudes to flourish.<sup>34</sup> It fosters in society the notion that only the (genetically) 'perfect' are acceptable and that it is socially desirable to prevent people with disabilities from being born. Through trying to eradicate congenital differences, we promote a cult of perfectionism.<sup>35</sup>

The UN International Bioethics Committee warns that: *'The widespread use of genetic screening and in particular of [the new test] NIPT may foster a culture of "perfectionism" or "zero defect" and even renew some "eugenic trends", with the consequence that it could become more and more difficult to accept imperfection and disability as a part of normal human life and a component of the diversity we are all called on to acknowledge and respect. The right of an individual to make autonomous choices is to be made consistent with the right not to be subjected*

*to discrimination or stigmatisation based on genetic characteristics and the duty to respect every human being in her or his uniqueness'*.<sup>36</sup>

The argument that it can help prepare couples for the birth of a child with a disability should not be ignored but it is a side argument because the primary purpose of NIPT is to detect genetic differences, and we need to question why society wants to do so. Society now has a wrongly narrow view about who is truly 'normal' or 'healthy'. Disabled Peer Lord Shinkwin has written: *'The irony is that this isn't really about abortion. Ultimately, it's about power, the power of non-disabled people to determine the fate of other – disabled – human beings, whether we should live or whether we should die. Prejudice must not prevail'*.<sup>37</sup>

A Christian perspective is strikingly different and regards the most vulnerable human beings, the baby in the womb with a genetic 'condition', as being of equal value and worth as any other person, regardless of ability or disability.

## CHRISTIAN PERSPECTIVES

Raising a child with special needs involves substantial costs in many areas, and few of us – if honest – would actually choose to bring a child with disabilities into the world. Caring for a child with any disability is challenging and calls for sacrifice – yet so does all parenting.

## Care for the weak

Throughout the Bible we see God's concern for the weak and, as stewards of his creation, we are called to emulate this; 'bearing one another's burdens' lies at the very heart of Christian morality.<sup>38</sup> We must 'defend the weak'<sup>39</sup> and 'help the weak'.<sup>40</sup> This mandate involves compassionate caring like that demonstrated by the Samaritan man in Jesus' parable, rather than seeking our own human means to obliterate weakness (and the weak) from the world.<sup>41</sup>

## Autonomy

While autonomy has Christian origins we must be cautious about approaches to self-determination that deny responsibility or moral accountability. Our God-given ability and right to exercise free will is only appropriately used when we look not just to our own interests but also those of others, as Paul tells the Philippians.<sup>42</sup> Similarly the Galatians are told to 'serve one another humbly in love'.<sup>43</sup> In such a context, we can



see that Christians are to seek autonomy that is not only limited by God's sovereignty, but also by concerns for the community and its common good.

God has not made us independent individuals, but placed us in families where 'mutual burdensomeness' is part of the created order. None of us can act in isolation and affect no one else. All of our decisions should take account of the needs of these shared relationships, not just our own wants and desires.<sup>44</sup>

## The role of the Church

Should the weak be sacrificed for the strong or should the strong make sacrifices for the weak? The Christian answer is clear that bearing one another's burdens is at the very heart of the gospel. We walk in the steps of the all-powerful Creator who laid aside everything and entered this world at great personal cost to rescue, care and serve.

There is no doubt that providing life-time support for people with genetic disabilities can be costly in emotional and economic terms. This is where the Church is needed, to provide these most vulnerable of human beings with the honour, respect, love and protection that they deserve and to be places of support to their parents and families. In such an environment, there would seem to be little need to make use of NIPT tests. The unborn child should be welcomed unconditionally into the world, and the family supported in their parenting, regardless of any genetic difference the baby may have. If parents do use NIPT, and face an unexpected diagnosis, they need a life-affirming environment where their child will be welcomed and supported with loving care and acceptance.

## Four challenges that NIPT presents:

- To the Church: to be seen to be a place where we bear one another's burdens, so that women do not feel any need to use NIPT. The Church must also challenge the pursuit of perfectionism in our culture and stand against disability discrimination that is inherent in most use of NIPT.
- To Christian doctors, because bearing one another's burdens involves not only seeking to provide unbiased information and the best medical care for *all* members of our society, especially the most vulnerable, but also proactively supporting their families in the longer term.

- To our culture, with its inextricable link to abortion. By using it to search out (and destroy) genetic 'difference' it enables discrimination against vulnerable human lives and ultimately promotes a culture of perfectionism.
- To couples expecting a baby. Freedom to choose is real and biblical, but a tension exists between freedom, accountability and responsibility. The challenge is to make choices within God-given boundaries. It is not simply just 'our' life to do with as we please. NIPT might be more accurate than other

tests but it does not remove the need for invasive testing. While the test itself is morally neutral, it opens up the option to screen for many more conditions and traits in the future, with the consequent removal of unborn babies with such conditions as its premise. The basis of any testing (if it is needed at all) should be the health and furthering of those with disabilities, but the likelihood is that society will not use the test well. NIPT puts us under a moral temptation that we would be sensible to flee from.

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