



**Non-invasive prenatal  
testing: ethical issues**

SHORT GUIDE

NUFFIELD  
COUNCIL ON  
BIOETHICS

This is a short guide to the report *Non-invasive prenatal testing: ethical issues*, published by the Nuffield Council on Bioethics in March 2017. This guide sets out some of the issues, themes and conclusions that are discussed in more detail in the full report, available at [www.nuffieldbioethics.org/nipt](http://www.nuffieldbioethics.org/nipt)

The report is the outcome of a project carried out by an interdisciplinary Working Group that included people with expertise in genetic counselling and clinical medicine, psychology, ethics, public health, disability research and law. The Working Group sought the views of a wide range of people, such as doctors and midwives, women with personal experience of screening, and people with genetic conditions and variations, such as cystic fibrosis and Down's syndrome, and their families. These were obtained through an open call for evidence, an anonymous online survey, group meetings and one-to-one interviews. More information about this process and who was involved is available at [www.nuffieldbioethics.org/NIPT-evidence-gathering](http://www.nuffieldbioethics.org/NIPT-evidence-gathering)

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# Introduction to NIPT

## What is NIPT?

Non-invasive prenatal testing (NIPT) is a technique that can be used to test a fetus for a range of genetic conditions and variations using a blood sample taken from the pregnant woman. It is referred to as 'non-invasive' because it does not involve inserting a needle into the woman's abdomen or cervix, as is the case with invasive testing where cells are taken from the amniotic sac (in amniocentesis) or the placenta (in chorionic villus sampling, or CVS).

## How does it work?

Small amounts of DNA, often referred to as 'cell free DNA' (cfDNA), circulate in everybody's blood. From an early stage in pregnancy, this includes DNA from the placenta, which is very similar to the fetal DNA. By analysing these cfDNA fragments, it is possible to find out genetic information about the fetus. The amount of fetal cfDNA in the woman's blood increases as the pregnancy progresses. NIPT can be carried out from about 9 or 10 weeks of pregnancy, which is when there is usually enough fetal cfDNA in the woman's blood to get an accurate result.

## What does it test for?

Currently, NIPT can be used to test a fetus for a range of genetic conditions or variations, for example:

- It can be used to **estimate the chance** that a fetus has Down's, Edwards' or Patau's syndrome (an invasive test is still required to provide a definitive diagnosis).
- It can **provide a definitive diagnosis** for some genetic conditions, such as cystic fibrosis, achondroplasia and Apert syndrome, if they are inherited from the father or arise at conception.
- It can be used to **determine the sex** of a fetus.

The accuracy of NIPT varies depending on the condition or variation being tested for. Different circumstances can also affect the accuracy, such as whether it is a single or multiple pregnancy and if the fetus is already known to be at increased risk of the condition.

## What is new about NIPT?

NIPT has a number of features that distinguish it from other prenatal screening and testing techniques:

- It is **more accurate than other non-invasive prenatal screening tests** for Down's, Edwards' and Patau's syndromes, including the combined test currently offered to all pregnant women in the NHS.
- Because it is non-invasive, **NIPT carries no risk of miscarriage**.
- In some circumstances, **NIPT can provide earlier results** than current screening and diagnostic tests.
- **NIPT requires no specialist skills or equipment in the healthcare setting**, such as sonographers and ultrasound equipment.
- It is **less physically uncomfortable** for the woman than invasive diagnostic testing such as amniocentesis, which can be unpleasant and painful for some women.

## Screening and testing

**Prenatal screening** is when a test is offered to whole populations of pregnant women to find out the chance that their fetus has or might develop particular diseases or conditions.



**Prenatal diagnostic testing** generally refers to testing offered to particular individuals to confirm or rule out diseases, conditions or variations that it is suspected a fetus might have.



# NIPT in NHS screening for Down's, Edwards' and Patau's syndromes

Down's, Edwards' and Patau's syndromes are examples of 'aneuploidy' which is where there is an unusual number of chromosomes in some or all of a person's cells. This can express itself in a range of ways, including in appearance, physical and intellectual development and health.



Further information about Down's, Edwards' and Patau's Syndromes is available in [Chapter 1](#) of the report.

## Screening for aneuploidies in the NHS

Current NHS policy is to offer all women a prenatal screening test (the 'combined test') for Down's, Edwards' and Patau's syndromes between 10 and 14 weeks of pregnancy as part of the NHS fetal anomaly screening programme. Diagnostic testing using CVS or amniocentesis is offered to women who are found through screening to have between a 1 in 2 and a 1 in 150

chance (which is considered a high chance) of having a fetus with one of these syndromes. CVS can be carried out from around 11 weeks of pregnancy, and amniocentesis from around 15 weeks. Approximately 1 in 10 women who get a high chance combined test result will be carrying a fetus with Down's syndrome; 9 in 10 women will not. Amniocentesis and CVS carry a small risk of miscarriage.

Studies have found that NIPT can give a more accurate prediction of the chance that a fetus has Down's, Edwards' or Patau's syndromes than the combined test alone. NIPT is more accurate in pregnant women who are already known to have at least a 1 in 150 chance of having a fetus with these syndromes, compared to unscreened pregnant women. When NIPT is offered to these higher chance women, as a second stage screening test (ie, following the combined test), 9 in 10 women who receive a high chance NIPT result will have a fetus with Down's syndrome. This means there is still a small but significant chance a fetus is not affected when a high

chance NIPT result is received, and so CVS or amniocentesis should be carried out to get a definitive diagnosis. However, the increased accuracy of NIPT means significantly fewer women will have invasive diagnostic testing.

NIPT has been piloted as a second stage screening test for pregnant women in a number of NHS hospitals. The findings suggest that, if NIPT is offered to all pregnant women in the UK who are found to have at least a 1 in 150 chance of having a fetus with Down's, Edwards' or Patau's syndromes following the combined test, it will lead to:

- Nearly 200 more fetuses with Down's syndrome identified per year;
- Over 3000 fewer invasive diagnostic tests carried out; and
- An estimated 17 fewer procedure-related miscarriages.

Therefore, offering NIPT as a second stage test for Down's, Edwards' and Patau's syndromes in the NHS would give more women the opportunity to prepare for a disabled child or to choose to have a termination (in England, Wales and Scotland). It would also lower the number of invasive diagnostic tests that are carried out.

The UK National Screening Committee (UKNSC) is the body that advises the UK Government on national screening programmes. Given the benefits of accuracy and safety offered by NIPT, in January 2016, UKNSC recommended an 'evaluative' implementation of NIPT for Down's, Edwards' and Patau's syndromes as a second stage screening test in the NHS fetal anomaly screening programme. In November 2016, the Department of Health announced that NIPT will be made available on the NHS, as recommended, from 2018.

## Is NIPT an accurate test for Down's syndrome?

A review of over 40 international research studies has found that when NIPT is used in pregnant women already found, through initial screening, to have a high chance of having a fetus with Down's syndrome:

- NIPT will identify 97% of fetuses with Down's syndrome
- 9 in 10 women with a 'high chance' result will have a fetus with Down's syndrome; 1 in 10 will not (a false positive result).
- NIPT has very few false negative results: almost all women with a 'low chance' result will not have a fetus with Down's syndrome.

## NIPT in NHS screening for Down's, Edwards' and Patau's syndromes continued...

### Implications

In addition to the benefits offered by NIPT, the people and organisations we spoke to during our project suggested the introduction of NIPT in the NHS screening programme might have a number of consequences that require consideration:

#### Information and support

- Existing challenges in ensuring women and couples are making informed, autonomous choices about prenatal screening may be intensified by the introduction of NIPT. For example, there are concerns that some healthcare professionals, when imparting information about Down's syndrome, focus on medical problems, such as heart problems, and learning disability, without describing more fully what it is like to have a child with Down's syndrome. This may influence the decisions women and couples make. Women and couples' choices are also influenced by societal attitudes towards disability, the presentation of disability and prenatal testing in the media, and the perceived impact of a disabled child on the family.
- Given the accuracy and non-invasive nature of NIPT, there are concerns that women and couples will think it is equivalent to a diagnostic test, and that it is a 'routine' part of prenatal care. Also, women and couples might not appreciate the difficult choices they may be faced with after having NIPT.

#### Timing and accuracy of diagnosis

- NIPT can fail to produce a result, or the result can be inconclusive. This could prolong the screening pathway for some women and cause anxiety.
- Offering NIPT to women as a second stage test may lead to a delay in diagnosis for some women. Currently women who are found to have a high chance of their fetus having one of the syndromes following the combined test are offered a diagnostic test (CVS or amniocentesis). If these women then take up an offer of NIPT, and subsequently decide to have confirmatory diagnostic testing, there may be a delay of a week or more before a diagnosis is received. This delay may be significant to some women, particularly those considering termination of pregnancy.

#### Implications for people with genetic conditions

- Introducing NIPT on the NHS is likely to lead to an increase in the number of diagnoses of Down's, Edwards' and Patau's syndromes, and possibly a rise in terminations. If this leads to a significant reduction in the number of babies born with one of these syndromes, it is possible that the specialist health and social care they receive, and the importance attributed to research into these syndromes, will be affected in the future.
- Making NIPT available on the NHS could be perceived as sending negative and hurtful messages about the value of people with the syndromes being tested for.

- Disabled people and their families might be more vulnerable to discrimination, stigma or abuse if NIPT gives rise to perceptions that people are 'to blame' for having a baby with a disability.

#### Implications for NHS and healthcare professionals

- Introducing NIPT into the NHS prenatal screening programme may lead to changes in the demand for related NHS services such as genetic counselling, invasive diagnostic testing, termination and laboratory services.

#### Future uses of NIPT in NHS prenatal screening programmes

It is possible that NIPT for further genetic conditions or impairments could be proposed for inclusion in NHS prenatal screening programmes in the future. This raises particular issues for the appraisal of screening programmes:

- Although the central aim of prenatal screening is to promote informed choice, there may be unintended consequences for people with the conditions and impairments being screened for and others. This is not currently taken into account in the criteria used by the UKNSC to appraise screening programmes.
- There is a need for more transparency around how screening programmes are appraised, and for improvements in how ethical, social and legal issues are considered in the appraisals.

“Making NIPT available on the NHS could be perceived as sending negative and hurtful messages about the value of people with the syndromes being tested for.



For more information, see Chapter 2 of the full report

# NIPT for rare genetic conditions in the NHS

NIPT can be used to test for other, rare genetic conditions. Some of these conditions run in families, such as cystic fibrosis and Duchenne muscular dystrophy; some arise at the time of conception, such as thanatophoric dysplasia.

Pregnant women with a family history of a genetic condition, or whose fetus has been found to have an anomaly on a scan, are usually referred to a specialist genetic testing service by their obstetrician, midwife or GP.

Until recently, if it is suspected that a fetus has a rare genetic condition, the only option for women and couples seeking a diagnosis would be to have an invasive test such as amniocentesis or CVS, which carry a small risk of miscarriage. However, NIPT can now be used to test safely for some conditions if they are inherited from the father or arise at conception. NIPT for these conditions can usually be carried out earlier than invasive testing and is often diagnostic, removing the need for invasive testing altogether.

There are currently no UK-specific guidelines for healthcare professionals on offering NIPT through specialist genetics testing services. Decisions about what tests should be offered, and to which patients, are made on a case-by-case basis by doctors such as clinical geneticists.

## Implications

In addition to the benefits offered by NIPT, the people and organisations we spoke to during our project suggested that the increasing use of NIPT for rare genetic conditions in the NHS raises a number of issues that require consideration:

### Implications for the NHS

As NIPT for rare genetic conditions becomes more widely available, genetic counselling services will need to grow to meet the information and support needs of those undergoing testing.

### Implications for people with genetic conditions

Introducing NIPT on the NHS is likely to lead to an increase in the number of diagnoses of rare genetic conditions, and possibly an increase in terminations. This gives rise to concerns similar to those raised by NIPT for aneuploidies. For example, that it might send out negative messages about the value of people with genetic conditions, and make them and their families more vulnerable to discrimination, stigma or abuse.

## Possible future developments

This is a rapidly moving field and uses of NIPT for other single gene conditions, or 'panel tests' for several related conditions, are likely to be developed in future. The availability of NIPT for significant medical conditions or impairments can enable pregnant women and couples to make informed choices about their pregnancies regarding whether to continue and prepare for the birth of a disabled child or whether to have a termination.

It is also possible that NIPT could be developed in future to test fetuses for genetic conditions that are likely to affect them only in adulthood, or to test whether a fetus carries a copy of a gene that does not cause a condition on its own but might do so in future generations. Whole genome sequencing using NIPT might also become available to pregnant women and couples, where it is suspected that the fetus has a genetic condition but the origin is unknown.

Making decisions about whether NIPT should be offered for this kind of use and who it should be offered to will involve consideration of:

- how best to respect the autonomy and protect the interests of the future child or adult, particularly where a test makes available detailed genetic information about them, or reveals that they are likely to develop a serious genetic condition later in life;
- whether the information being sought is medically useful;
- whether NIPT might inadvertently reveal previously unknown genetic information about the pregnant woman or her partner; and
- what genetic counselling and support will be available to women and couples undergoing testing.

## What is a 'significant medical condition or impairment'?

We use the term 'significant medical condition or impairment' in this report to describe what would be grounds for termination under section 1(1)(d) of the Abortion Act 1967 (the 'fetal anomaly ground'). We recognise that what constitutes a significant medical condition or impairment is a judgement that depends on several factors, including the likely level of impairment, the available treatment options, and the views of and potential impact on the family and the individual themselves. In this report, we refer to 'less significant medical conditions or impairments' as those that would not have a significant impact on the life of the child or family, or where remedial treatment is available, and would not usually be considered grounds for termination.



## NIPT in the private sector

In the UK, women and couples have been able to access NIPT for Down's, Edwards' and Patau's syndromes in the private healthcare sector since 2012. Some NIPT manufacturers offer to test for other conditions, such as those caused by an unusual number of sex chromosomes (sex aneuploidy) or where tiny pieces of chromosome are missing (microdeletions), that are not currently offered in the NHS. Most offer NIPT to determine the sex of the fetus.

The majority of NIPT providers only offer their tests to women through hospitals and healthcare clinics, although some clinics are located in retail chains on high-streets. NIPT is also available on a direct-to-consumer basis from a small number of websites.

### Regulation and professional standards

There is no law or regulation that covers all aspects of NIPT in the private sector, but various legal instruments and guidelines cover different elements of how NIPT is made, sold and provided.

The **manufacture** of NIPT tests in the UK are currently regulated under the UK Medical Devices Regulations 2002, which implements an EU Directive. This only applies where the test itself is carried out within the EU. Many tests currently being offered to women and couples in the UK involve sending their blood samples away to be analysed in the US or China and are not therefore covered by the Directive.

**Private hospitals and clinics** in which NIPT is offered are regulated by the Care Quality Commission (CQC) in England and by equivalent bodies in Scotland, Wales and

Northern Ireland, and must carry out their services to certain standards.

**Healthcare professionals** that offer and provide NIPT services in the private sector must adhere to standards set by professional regulators such as the General Medical Council (GMC) or the Nursing and Midwifery Council (NMC). There is no specific professional guidance on using NIPT to test for genetic conditions or variations.

**Advertising** of products and services including NIPT is monitored by the Committee of Advertising Practice (CAP). CAP produces codes of conduct for advertising in broadcast and non-broadcast media. The codes are enforced by the Advertising Standards Authority.

### Implications

Many issues raised by the offer of NIPT in the private sector are similar to those in the NHS, such as the possibility of failed or inconclusive tests and of unanticipated or secondary findings about the pregnant woman. However, we heard a number of concerns from people we spoke to during our project specifically relating to the way NIPT is offered:

#### Marketing and information

Although there are examples of good practice, there is commonly a lack of good quality information from manufacturers, private hospitals and clinics about the limitations of NIPT and the conditions being tested for. The information currently provided to women and couples is frequently incomplete, unsubstantiated,

inaccurate or misleading, and some use emotive language. We also heard concerns that the follow-up support offered to women with a high chance result is inadequate, with the NHS being left to 'pick up the pieces'. These concerns are particularly relevant to the provision of NIPT on a direct-to-consumer basis, in which the test might be offered and results delivered without the provision of adequate information or support.

#### NIPT for other conditions

The accuracy of NIPT for conditions such as microdeletions and sex aneuploidy has not been widely researched and the chance of a false positive result is often much higher than for Down's, Edwards' and Patau's syndromes, which could lead to more women seeking invasive tests to confirm a diagnosis.

#### NIPT for sex determination

The offer of NIPT to reveal the sex of the fetus at an early stage in pregnancy may increase the risk of sex selective abortions taking place. This practice is opposed by many, believing it is sexist and wrong. There is some evidence that sex selective abortions have happened in the UK and they are known to occur in other countries. It is also known that people who live in countries where prenatal sex determination is illegal, such as China and India, travel to countries where it is legal to have tests.

### Possible future developments

It is possible that in future NIPT will be available in the private sector for a wider range of genetic conditions and features, and that these may include less significant medical conditions or impairments, or non-medical traits. Whole genome sequencing may also be offered, raising the possibility that large amounts of information about the genetic make-up of the fetus could be made available to prospective parents and stored for the future.

Key issues to consider regarding these possibilities include:

- whether the information being sought is medically useful;
- how best to respect the autonomy and protect the interests of the future child or adult, particularly the risk that such tests might undermine the future person's ability to make their own choices about accessing and allowing others access to information that relates to their health, abilities, personality or physical attributes.
- whether allowing such tests would encourage discrimination against people with certain genetic features, including sex, or create damaging perceptions of what constitutes a 'normal' or 'healthy' baby.



## Ethical values

The development and increasing availability of NIPT raises a range of ethical issues, some of which are similar to those raised by prenatal screening more widely. We propose that these can be broadly understood through consideration of the following ethical values.

### Choice, autonomy and consent

Our ability to make free, informed choices about the medical tests and treatments we undergo is considered to be an important principle in modern healthcare. *Reproductive autonomy* refers to the capability men and women have to make choices about when they become parents, how many children they have and whether or not to make use of technologies such as prenatal testing.

NIPT can enhance or facilitate reproductive autonomy in different ways, including by enabling women and couples to prepare for a baby with a condition or trait, or decide to have a termination, potentially at an earlier stage of pregnancy. However, NIPT could also undermine autonomy and choice if accurate and balanced information about the test and the conditions being tested for is not available, or if women and couples feel they are expected to make a particular decision.

### Avoiding harm

The Government has a duty to protect its citizens from harm. As part of this, it has a role to play in eliminating or reducing any harms that might be caused by healthcare interventions such as NIPT that are available through the NHS, or to consumers in the private healthcare sector.

Providing tests and treatments in publicly funded healthcare services that are safer, more effective and involve less discomfort than other available tests could be seen

as a way of meeting these responsibilities. NIPT has the potential to reduce harms, for example where it can replace or reduce the need for invasive testing.

NIPT itself could also give rise to harms. For example, it could cause anxiety if the information and support provided to women and couples is inadequate or misleading, or where inaccurate or unreliable results are returned. If NIPT leads to a significant decrease in the number of people born with genetic conditions or impairments, it could lead to fewer resources being invested in research, healthcare and education relating to and available to people with genetic conditions, and cause offence, social exclusion and discrimination.

### Equality, fairness and inclusion

It is generally accepted that the state has a duty to promote equality and work to ensure that all people are treated fairly. This involves taking into account how policies such as a new health intervention might reduce or worsen existing inequalities. It also entails the duty to ensure that public money is spent fairly.

NIPT has the potential to contribute to women's ability to exert control over the circumstances of their pregnancies, with implications for their role in the workplace and wider society. Introducing NIPT into the NHS screening programme will mean that more women will have access to safer, more accurate prenatal testing.

However, NIPT has the potential to undermine equality, fairness and inclusion for disabled people in a number of ways. For example, it may give rise to perceptions that people are to blame for having a baby with a disability, and make disabled people and their families more vulnerable to stigma and abuse.

## The Working Group's ethical approach

The tensions that exist between the potential benefits of current and possible future uses of NIPT and the risks with which these uses are associated, create challenges for public policy. We suggest three general principles that, taken together, can provide a foundation for promoting reproductive autonomy and providing choice, while minimising potential harms and supporting an equal, fair and inclusive society.

**PRINCIPLE 1: The wider societal environment in which NIPT is provided and developed should be considered when developing policy relating to NIPT.**

We believe the state has a duty to promote an equal and inclusive society, and that wider social inequalities and injustices should be taken into account when policy, regulation and law relating to NIPT is developed.

Concerns about the inequality and challenges that disabled people face, for example in accessing adequate health and social care and support, educational and employment opportunities, are likely to influence the ways that women and couples think about their choices in pregnancy and prenatal testing. It is our view that women and couples will be better able to make genuine choices about their pregnancies if all disabled children are actively welcomed into the world and valued as equals to those without disabilities.

**PRINCIPLE 2: Pregnant women and couples should have access to NIPT within an environment that enables them to make autonomous, informed choices.**

When NIPT is offered to women and couples in the NHS, it will be important to emphasise that it is an optional test and that there are no expectations as to what is the 'right' decision.

In both the private and public sectors, it is important that accurate, balanced and non-directive information and support are made available to women and couples deciding whether to have NIPT, whether to have further testing, and whether to continue or terminate a pregnancy. High quality support must be available both to women who choose to proceed with pregnancies and to those who do not.

**PRINCIPLE 3: Efforts should be made to reduce any risks of significant harms posed by the growing use and development of NIPT.**

Action should be taken to minimise risks of harm resulting from the availability of NIPT.

For example, the use of NIPT to test for conditions where the accuracy of the test is unknown or poor can cause unnecessary anxiety for women and couples and may increase the number of women seeking invasive diagnostic testing.

Possible harms may also arise from extending the use of NIPT beyond testing for information that has a bearing on the immediate or early health of fetuses and future children. If, for example, NIPT could be used in future to test for less significant or adult onset conditions, or non-medical features, this would raise concerns about the capability of future children and adults to make their own choices about accessing information relating to their genetic makeup and health, and to be able to access the same opportunities and services as those who do not have this information about themselves.





## Conclusions and recommendations

### NIPT for significant medical conditions and impairments

Women and couples should be able to access NIPT to enable them to find out at an early stage of pregnancy, if they wish, whether their fetus has a significant medical condition or impairment that manifests at birth or in childhood. This information can be clinically useful and enable women and couples to have meaningful reproductive choice. However, we believe that it should only be available within an environment that enables women and couples to make informed choices, and where steps are taken to minimise potential harms.

The Government has a duty to provide disabled people with high quality specialist health and social care and to tackle discrimination, exclusion and negative societal attitudes experienced by disabled people. This is important for offsetting the potential harms posed by the use of NIPT for significant medical conditions or impairments to disabled people and their families. Women and couples will be better able to make genuine choices about their pregnancies if all disabled children are actively welcomed into the world and valued as the equals of those without disabilities.

NIPT should only be offered if it provides an accurate prediction of whether a fetus has the condition or impairment that is being tested for. Private providers should stop offering NIPT for conditions where test

performance is poor or unknown, such as NIPT for some microdeletions.

We support the introduction of NIPT for Down's, Edwards' and Patau's syndromes in the NHS for women who have been found to have at least a 1 in 150 chance of having a fetus with one of these conditions. Given the higher rate of false positives when NIPT is used in the general population of pregnant women and the significant failure rate of NIPT, we believe offering it to women only in the higher chance category is a proportionate approach.

All providers of NIPT for significant medical conditions or impairments have a responsibility to provide high quality information and support. NIPT should only be offered in a healthcare setting by health care professionals with the knowledge and skills needed to support women and couples to make informed choices. High quality education and training must be compulsory for all health and social care professionals involved in NHS prenatal screening. Accurate, balanced and non-directive information should be readily available to women and couples in accessible written and multimedia formats. This information and training should be developed with the support of those with genetic conditions or their families.

To ensure NHS patients receive the information and support they need to make decisions relating to NIPT for rare genetic

conditions, it will be important for the NHS to ensure it has sufficient genetic counselling resources.

As the only prenatal testing support organisation to which the NHS directs pregnant women, it is important that Antenatal Results and Choices (ARC) provides balanced, non-directive and impartial advice to parents, and balanced information via training to health professionals.

A number of private providers of NIPT in the UK are currently not meeting their obligations to offer good quality information and support to pregnant women and couples. We suggest that providers in the private sector should be encouraged to seek certification from recognised information quality schemes, such as NHS England's Health Information Standard, to help women and couples know that their information has been quality checked.

The Committee on Advertising Practice should pay closer attention to the advertising practices of NIPT manufacturers and providers to ensure that they are not misleading, harmful or offensive.

Private hospitals and clinics should only offer NIPT as part of a package of care that should include, at a minimum, counselling before and after testing and follow-up diagnostic testing where this is required. NIPT should not be available on a direct-to-consumer basis unless it is offered as part of this package.

## Conclusions and recommendations continued...

### NIPT for other conditions and variations

In the future, NIPT may open up possibilities for testing fetuses for a much wider range of genetic conditions and variations than is currently possible at an early stage of pregnancy. It is important for policy makers to be prepared and consider the potential consequences of these developments.

Having weighed up the potential benefits and harms, and the wider societal consequences, we believe that NIPT should not generally be used:

- to test whether a fetus has a less significant medical condition or impairment;
- to test a fetus for a condition that will not affect the future person until adulthood; or
- to find out whether the fetus is a carrier of a gene for a medical condition or impairment; or
- to reveal non-medical features such as sex.

Although some women and couples might like to have this information, we believe that in most cases it would not be medically useful and would undermine the capability of the future person to make their own choices about accessing their genetic information. An exception might be posed by women and couples with a family history of an extremely serious adult-onset condition,

such as Huntington's disease, who want to find out if their fetus will develop the condition, if there is no treatment available and if termination of pregnancy is an option.

The ability of NIPT to reveal the sex of the fetus at a much earlier stage increases the risk that terminations on the basis of sex could take place. The Working Group believes this will in most cases be based on discriminatory attitudes. If NIPT for sex determination continues to be available in the UK, there is a real possibility that sex selective terminations may be encouraged within the UK, both among UK residents or through 'sex selection tourism'. **We recommend that NIPT providers should not offer sex determination of fetuses.**

### Whole genome sequencing

Our reasons for recommending that NIPT normally should not be used for less significant medical conditions or impairments, adult onset conditions, carrier status or non-medical features also apply to whole genome sequencing, which would reveal this information and more – much of which would be difficult to interpret. The prospect of whole genome sequences of fetuses being generated and stored raises additional concerns relating to the rights of the future person, given that information we are not currently able to interpret may be analysed and understood in future. We recommend that **the use of NIPT for whole genome sequencing of fetuses should not be offered outside research environments**, and that the Government should consider establishing a moratorium with NIPT manufacturers on the use of NIPT for whole genome sequencing.

There may be rare exceptions when it is appropriate to use NIPT for whole genome sequencing, for example to assist in obtaining a diagnosis when it is suspected that a fetus has a serious medical condition or impairment but the cause is unknown.

### Guidance for practitioners

There is currently no professional guidance on NIPT in the UK healthcare context. **We recommend that the relevant Royal Colleges and other professional bodies work together to produce guidance for health and social care professionals on the availability and provision of NIPT.**

We further recommend that the guidelines of the Royal College of Obstetricians and Gynaecologists on termination of pregnancy for fetal anomaly should be renamed immediately to make it clear that they also cover continuation of pregnancy after an anomaly has been diagnosed in the fetus and that this section is significantly expanded, or additional guidelines created. For example, it should be emphasised that where there is a high chance that the fetus has a particular condition, women and couples should have access to expert advice, including from those with a first-hand knowledge of what life is like for children and adults with the condition and their families. We also recommend that NICE should produce clinical pathway guidance on continuation of pregnancy after diagnosis of fetal anomaly.



# Overview

The Nuffield Council on Bioethics has published a report that explores ethical issues arising from current and possible future uses of non-invasive prenatal testing (NIPT). The report considers views and evidence gathered from a wide range of people and organisations, and from the academic literature. It sets out the potential implications of this technique for prenatal screening and testing for a range of genetic conditions and variations both in the NHS and in the private sector.

We suggest three general principles that should be adopted by policy makers:

1. The wider societal environment in which NIPT is provided and developed should be considered when developing policy relating to NIPT.
2. Pregnant women and couples should have access to NIPT within an environment that enables them to make autonomous, informed choices.
3. Efforts should be made to reduce any risks of significant harms posed by the growing use and development of NIPT.

## Key recommendations include:

- Women and couples should be able to access NIPT to enable them to find out, if they wish, whether their fetus has a significant medical condition or impairment, but only within an environment that enables them to make autonomous, informed choices, and when steps are taken to minimise the potential wider harms of NIPT.
- NIPT should not normally be used to test whether a fetus has a less significant medical condition or impairment or an adult-onset condition; or to find out whether the fetus is the carrier of a gene for any kind of medical condition or impairment; or to reveal non-medical features of the fetus, including sex. NIPT for whole genome or exome sequencing of fetuses normally should not be offered.
- Professional guidance for health and social care professionals on the availability and provision of NIPT in the UK should be developed by relevant Royal Colleges and other professional bodies.

**This guide and the full report are available on the Council's website:**  
[www.nuffieldbioethics.org](http://www.nuffieldbioethics.org)

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