

SUMMARY OF REPORT

Non-invasive prenatal testing: ethical issues

Published 1 March 2017

Non-invasive prenatal testing (NIPT) is a technique that can be used to test a fetus for genetic conditions and variations. It involves taking a blood sample from the pregnant woman at around 9 or 10 weeks of pregnancy. NIPT is more accurate than other screening tests, it carries no risk of miscarriage and, in some circumstances, NIPT can provide earlier results than current screening and diagnostic tests.

From 2018, NIPT for Down's, Edwards' and Patau's syndromes will be available to pregnant women as a second stage screening test in the NHS fetal anomaly screening programme. NIPT is already used in the NHS to diagnose fetuses for other genetic conditions, such as cystic fibrosis and achondroplasia, in women where there is a family history or another indication. NIPT for a range of genetic conditions, and for finding out fetal sex, is widely available through private healthcare providers. NIPT for more genetic conditions and variations is likely to be available in the future. Whole genome sequencing using NIPT has already been carried out in a research setting.

The Nuffield Council on Bioethics report considers, at this early stage of its use, how NIPT could change the way we view pregnancy, disability and difference, and what the wider consequences of its increasing use might be.

Key recommendations

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 the potential wider harms of NIPT are minimised.

- To offset the possibility that the increased use of NIPT might adversely affect disabled people, the Government and those subject to the Public Sector Equality Duty have 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.
- Before the introduction of NIPT in NHS screening, Public Health England should produce accurate, balanced and non-directive information for women and couples about NIPT and the conditions for which it tests. High quality education and training must be compulsory for all NHS healthcare professionals involved in prenatal screening.
- The Committee of Advertising Practice should more closely monitor the marketing activities of private NIPT providers to ensure that they are not being misleading or harmful.
- Certification from information quality schemes should be sought by private NIPT providers to help women and couples to know that the information they provide has been quality checked.
- Private hospitals and clinics should be required by their regulatory bodies to only offer NIPT as part of an inclusive package of care that should include, at a minimum, pre- and post-test counselling and follow-up invasive diagnostic testing if required.

Key recommendations – continued...

NIPT should not generally be used to find out whether a fetus has a less significant medical condition or impairment, has an adult onset condition, or carries a copy of a gene that does not cause a condition on its own. Nor should it be used to reveal non-medical features of a fetus, such as sex.

- The Government should ensure that private NIPT providers stop offering fetal sex determination.
- The Government should consider establishing a moratorium with NIPT manufacturers to ensure whole genome sequencing of fetuses is not offered outside research environments (apart from in exceptional circumstances).

Screening for Down's syndrome 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 Chorionic Villus Sampling (CVS) or amniocentesis is offered to women who are found through screening to have a high chance of having a fetus with one of these syndromes. 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. From 2018, 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 will be offered NIPT. It is likely that fewer women will proceed to invasive diagnostic testing, thereby reducing the number of miscarriages that occur.

Key facts and figures

- Around **three quarters (74%)** of pregnant women currently decide to have a screening test for Down's syndrome – a figure that varies across the UK.
- Around **1 in 200 (0.5%)** of invasive tests (amniocentesis and CVS) result in miscarriage – although this figure is contested.
- Improvements in screening methods have led to an increase in the number of prenatal diagnoses of Down's syndrome over the past 25 years. For example, in 1989, **320 fetuses** were diagnosed with Down's, and in 2012, **1250 fetuses** were diagnosed.
- The proportion of women having a termination after a diagnosis of Down's syndrome ranged from **89 to 95 per cent** between 1989 and 2012, meaning that the actual number of terminations has increased.
- The number of live births of babies with Down's syndrome has remained fairly constant during this period with around **700 babies** born each year. This is likely due to an increased incidence of Down's syndrome in fetuses caused by an increase in the average age of mothers.
- Studies have suggested that when NIPT is offered to pregnant women with a high chance of having a fetus with Down's it would lead to:
 - nearly **200** more fetuses with Down's syndrome being identified per year
 - around **3000** fewer invasive tests
 - an estimated **17** fewer procedure-related miscarriages.