



**Down's Syndrome
Association**

A Registered Charity No. 1061474

Briefing for Members of Parliament

The implementation of non-invasive prenatal testing (NIPT)
and the provision of information to prospective parents

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Background

As a member of parliament you may be approached by constituents who are concerned about the implementation of NIPT (sometimes known as cell free fetal DNA, cffDNA) . The intention of this paper is to provide you with the information necessary for you to understand what the main concerns of the Down's Syndrome Association are in relation to NIPT and the provision of information to prospective parents involved in all prenatal screening.

The Down's Syndrome Association (DSA)

The aim of the organisation is to create the conditions that all people with Down's syndrome need to live full and rewarding lives and provide information, training and support to people (with Down's syndrome, their families and the professionals who work with them. The DSA maintains neutrality as a recognised source of impartial information and advice and acknowledges that the offer of prenatal testing for Down's syndrome is a routine part of antenatal care. However it is essential NIPT is delivered within an ethically sound and socially responsible framework which is appropriately regulated. Such a framework will ensure that each individual choosing to take the test is able to make a decision based upon accurate and balanced information provided by appropriately trained health professionals.

Non-invasive prenatal testing - NIPT

For many years the goal of researchers has been to develop a diagnostic prenatal test for Down's syndrome that does not involve an invasive procedure with an accompanying risk of miscarriage. The most recent developments have involved analysing cell-free maternal and fetal DNA in the mother's blood. This has come to be known as non-invasive prenatal testing (NIPT).

NIPT is a screening test, **not** a diagnostic test. An invasive test (amniocentesis or CVS) is still required to confirm a definitive diagnosis of Down's syndrome. NIPT is not yet available on the NHS as part of the UKNSC pilot. [Current recommendations](#) from the UK National Screening Committee (NSC) are to implement the test as part of an evaluation process. Once the evaluation has taken place the results will need to be further reviewed by the UKNSC before any further recommendations to the Government can be made. Even if the evaluation is favourable the UKNSC recommends that NIPT will **only** be offered to women who are deemed at higher risk following the current primary screen.

Concerns

There is general agreement amongst researchers and professional bodies in this field that the implementation of NIPT must be accompanied by the training of health professionals in the provision of accurate unbiased information. Professor Alan Cameron, Vice President of Clinical Quality for the Royal College of Obstetricians and Gynaecologists has stated recently that *'Should the test be rolled out, resources and training for health care professionals offering this testing will be necessary, in particular around communication and counselling expectant parents about the implications of the test results.'* The DSA goes one step further in recommending that health practitioners are able to provide information about the condition being screened for.

Dr Anne Mackie, director of screening at Public Health England, has said: *'While the evidence suggests that NIPT is much more accurate in predicting Down's syndrome than current tests, there are a number of questions about its use in a real-life screening programme that the evidence hasn't yet been able to clarify.'*

Private providers

Prior to the completion of the UKNSC evaluation, the DSA is concerned that NIPT has become widely available in the private sector. Private clinics, one in collaboration with major high street retailer Mothercare, are able to offer NIPT through a number of different laboratories, based here and overseas. The Association is also aware of at least one NHS hospital which is giving all prospective parents a leaflet on the Harmony NIPT test enabling them to access a discounted rate of £350 for a NIPT via the hospital.

The ease and availability of NIPT on-line, on the high street and in hospitals without due consideration given to the manner in which it is advertised and delivered is of major concern to the DSA. Companies have been using marketing strategies such as reaching out to pregnant women through social media or offering introductory pricing specials to capture a market share. The Nuffield Council on Bioethics raised this issue in a background paper written in November 2015¹

The aggressive marketing of NIPT to healthcare providers and to women raises concerns regarding premature implementation of NIPT into routine practice in the absence of sufficient evidence of clinical utility.

Parents experiences - the need for balanced accurate up-to-date information

A key objective for the Down's Syndrome Association has always been to ensure the provision of balanced, up to date [information](#) to prospective parents and the health professionals who support them.

Information should be delivered by health professionals trained in communicating and counselling individuals about the implications of screening test results prior to them embarking upon the process. The Association continues to hear from families who have received negative experiences of **the current** national screening programme for Down's syndrome. This feedback indicates that the regulations and guidance available **even** in the public sector are not meeting the needs of some patients who undertake the process. Whilst some health professionals handle the situation sensitively, we have evidence confirming that the information given by others when results are positive is not balanced, accurate or current.

¹ Nuffield Council of Bioethics Background Paper NIPT Key clinical ethical, social, legal and policy issues (<http://nuffieldbioethics.org/wp-content/uploads/NIPT-background-paper-8-Nov-2015-FINAL.pdf>)

I was 12 weeks pregnant and had just found out that I was carrying a little boy with Down's syndrome. I was distraught. The advice and information provided by my local hospital was less than reassuring. Not only were the risks of me having a healthy pregnancy and safe delivery (I was 45 and this was my fourth caesarean) considered to be too great to continue with the pregnancy, it wouldn't really be worth it anyway, especially given that there was a significant chance of the baby having health complications (perforated anus, hole in the heart, digestive problems...)

Statistics were shown to my husband and I from detailed medical tomes to 'evidence' such a bad state of affairs. Fortunately, my husband is a bit of a maths geek and told the consultant that the statistics didn't add up and more than that had been misinterpreted by her. As far as the evidence was presented, my prospects were so awful, if I made out of the hospital car park alive that evening without internally combusting, I would be doing well. Not only that, "...think about your other children."

My consultant booked me in for a termination the following day at 8.30am. I could not help being concerned. The consultant had no good news, no impartial advice. In fact, she had only delivered two "live" babies with Downs in the last two years as most terminated their pregnancies. It felt like propaganda rather than a balanced consultancy.

The Down's Syndrome Association conducted two surveys (2009 and repeated in 2014) to identify the antenatal and neonatal experiences of parents of a child with Down's syndrome. Both surveys revealed that women did not recall being provided with enough information about Down's syndrome during their pregnancy.

I would hate anyone to go through what we went through who didn't have access to the information and support available from yourselves. I think that it is vital that health professionals are aware of Down's syndrome and are able to present a true and balanced picture so that parents can make an informed decision on all of the available information. Not just the scary stuff!

We received the diagnosis of Down's syndrome when I was 33 weeks pregnant. I was given that diagnosis over the phone. I was told that a specialist nurse would contact me and she did but she was unable to see me for a couple of weeks so I felt left on my own at that point and sought out my own information.

The paediatrician that told us they suspected Down's syndrome. He made us feel as if it was our fault, we were asked why we did not have testing before and when I said that they were unable to do the test because of the baby's position his reply was 'maybe you should of had it done again'. We were given lots of leaflets with facts and figures but no one to talk to who really understood.

Data from the 2014 survey identified that 67% of respondents reported not being provided with any information about Down's syndrome by their healthcare services prior to screening. Of those respondents who went on to receive a positive test result only 25% received any form of counselling. Without access to accurate up to date balanced information provided by trained healthcare professionals, prospective parents accessing NIPT are put in a highly vulnerable position when having to make a life-changing decision.

DSA training

The DSA continues to campaign for funding to deliver its comprehensive [training course](#) for all those involved in the ante-natal care of women. The training, Tell it Right, Start it Right (TIR), is predominately delivered to midwives, but is also extremely relevant to obstetricians, paediatricians and ultrasonographers. It is accredited by The Royal College of Midwives, with the aim of ensuring that those people involved in providing information, supporting women through the antenatal period, carrying-out tests and giving test results, do so in an accurate and balanced way.

Crucially, health professionals need up to date and correct information about Down's syndrome in order to answer the questions expectant parents may ask. Through its TIR programme the DSA has built a body of evidence which proves that a significant number of prospective parents taking part in current national antenatal screening programmes for Down's syndrome continue to receive inadequate information. The Down's Syndrome Association therefore fears that the introduction of NIPT to the private sector can only place further pressure on the ability of all health professionals in both sectors to deliver appropriate information and support.

The DSA believes the availability of the NIPT test in the private sector as a test for Down's syndrome is premature. There has been insufficient consideration given to the ethical implications of NIPT, its regulation and the implications NIPT has upon the additional training needs of healthcare professionals. The Royal College of Obstetricians and Gynaecologists has recognised this in their Impact Paper 15².

Resources for the education and training of health professionals offering this testing and pre-test information and discussion with the patient will be required.

The Down's Syndrome Association are campaigning for:

- The implementation of mandatory training for anyone involved in the delivery of NIPT, in particular around communication and counselling expectant parents about the implications of the test results. We continue to receive evidence that current training is not meeting the needs of many prospective parents.
- A simplification of regulations surrounding the delivery of NIPT in both the public and private sectors
- An ethically sound and socially responsible framework in which NIPT can be delivered.

² RCOG Scientific Impact Paper No 15 <https://www.rcog.org.uk/en/guidelines-research-services/guidelines/sip15/>

Further reading

- Down's Syndrome Association website: www.downs-syndrome.org.uk
- House of Commons Science and Technology Committee [Report](#) on National Screening(2014-2015 sitting)
- [Government response](#) to the House of Commons Science and Technology Committee on National Screening (2014-2015 sitting)
- Best Ethical Practices for Clinicians and Laboratories in the Provision of NIPT [MA Allyse](#), PhD, [LC Sayres](#), BA, [M Havard](#), BA, [JS King](#), JD, PhD, [HT Greely](#), JD, [L Hudgins](#), MD, [J Taylor](#), MS, CGC, [ME Norton](#), MD, [MK Cho](#), PhD, [D Magnus](#), PhD, and [KE Ormond](#), MS, CGC
- [Report](#) on the Human Genome and Human Rights (UNESCO)
- A New Era in Non-Invasive Prenatal Testing Stephanie Morain, M.P.H., Michael F. Greene, M.D., and Michelle M. Mello, J.D., Ph.D. *N Engl J Med* 2013; 369:499-501 [August 8, 2013](#) DOI: 10.1056/NEJMp1304843 <http://www.nejm.org/doi/full/10.1056/NEJMp1304843>
- Nuffield Council of Bioethics Background Paper NIPT Key clinical ethical, social, legal and policy issues <http://nuffieldbioethics.org/wp-content/uploads/NIPT-background-paper-8-Nov-2015-FINAL.pdf>
- RCOG Scientific Impact Paper No 15 <https://www.rcog.org.uk/en/guidelines-research-services/guidelines/sip15/>
- NIPT a review of international implementation and challenges [Allyse M¹](#), [Minear MA²](#), [Berson E³](#), [Sridhar S³](#), [Rote M³](#), [Hung A³](#), [Chandrasekharan S⁴](#). <http://www.ncbi.nlm.nih.gov/pubmed/25653560>
- Prenatal Screening Reproductive Choice and Public Health Stephen Wilkinson <http://www.ncbi.nlm.nih.gov/pubmed/25521971>
- FAQs from UKNSC on NIPT (June 2015) [FAQs NIPT from UKNSC](#)
- Review of the UK National Screening Committee - [Recommendations](#)