

BRIEFING PAPER

Down's Syndrome - Non-Invasive Prenatal Testing (NIPT)

Further to the BBC2 documentary 'A World Without Down's Syndrome?' shown on 5th October 2016 and recent press coverage on the issue of screening, Down's Syndrome Scotland (DSS) compiled a short briefing to address some of the issues raised.

1. Down's Syndrome Scotland (DSS)

1.1 We are the only Scottish charity focused solely on the needs of people with Down's syndrome (Ds) and their family carers. We provide information, support and services for people with Ds, their families, carers and those with a professional interest. We also seek to improve public knowledge and understanding of Down's syndrome and to champion the rights of people with Ds. Our Family Support Service provides "all through life" support to families throughout Scotland, supported by local branches that hold activity groups for people with Ds and their families. We also conduct school consultation visits and training courses and offer a wide range of resources dealing with issues such as education and health.

1.2 DSS knows that people with Ds can and do lead full and rewarding lives. We acknowledge that the offer of pre-natal screening and testing for Down's syndrome is a routine part of antenatal care offered in Scotland and it is our policy to maintain neutrality as a recognised source of impartial information. We respect parents' right to make choices about antenatal tests and their outcomes.

1.3 Nevertheless we maintain that decisions regarding the implementation of NIPT should not be about the cost of the test or the costs of care. We continue to campaign for up-to-date, accurate information to be provided in a balanced way by well trained professionals to parents both before and during the screening process.

2. Background to NIPT

2.1 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 the accompanying chance of miscarriage. The most recent developments have involved analysing cell-free maternal and fetal DNA (cffDNA) in the mother's blood. This has come to be known as non-invasive prenatal testing (NIPT).

2.2 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. Current recommendations from the UK National Screening Committee (UKNSC) are to implement the test as part of an evaluation process to assess what impact NIPT

would have on the existing NHS Fetal Anomaly Screening Programme. Once the evaluation has taken place the results will need to be further reviewed by the UKNSC before any further recommendations to the UK 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 chance following the current primary screen. Women who are low chance will still be able to access the test privately¹. This is of concern too as there is not yet a clear picture of the quality of information provided prior to testing in the private sector or how it is regulated and by which body.

2.3 DSS's understanding is that the Scottish Government have not yet approved these new recommendations for implementation. A Scottish Government spokesperson said: "While we are fully committed to ensuring that pregnant women in Scotland have access to high quality, safe and effective screening services, there are a number of questions about the use of a Non-Invasive Prenatal Test (NIPT) that require further examination. No change will be made to the pregnancy screening programme without full consideration of the clinical evidence. This issue will be considered by the Scottish Screening Committee later this year."

2.4 Public Health England has established a group to look into the development of quality information for prospective parents about the offer of screening, the screening pathway and the development of training resources for NHS staff delivering care along the screening pathway. The first meeting will take place in November 2016 and the Down's Syndrome Association (DSA)² will be a contributor.

> DSS is of the view that a similar group to that set up by Public Health England should be established in Scotland and we would welcome involvement in this process.

2.5 Moreover DSS is currently involved with the Nuffield Council on Bioethics who are exploring the ethical, legal and regulatory implications of recent and potential future scientific developments in non-invasive prenatal testing, including its use in both NHS and commercial services. The Council will report the findings of the project early 2017.³

3. Accurate Information

3.1 We know from our membership that some parents received great support from medical staff throughout their pregnancy. However all too often we also gather evidence from families across the country about their negative experiences regarding screening, diagnosis or pre/post-natal support. It is worth noting that such experiences can be quite distressing for parents and can have longer term implications in their relationships with health professionals.

3.2 Calls to DSS indicate that there is already some confusion about the NIPT test. Some of the issues raised are:

- **A belief that NIPT is a diagnostic test and it means that no further tests such as Amniocentesis or CVS sampling will be required.**

¹ At present NIPT is available to all women privately – costs vary between £400 and £900.

² DSA is the only organisation to focus solely on all aspects of living successfully with Down's syndrome in England, Wales and Northern Ireland. For more information, see: <http://www.downs-syndrome.org.uk/>.

³ For more information, please see: <http://nuffieldbioethics.org/project/non-invasive-prenatal-testing/>.

- **Confusion about how to interpret the NIPT results.** Whilst DSS can provide callers with general information about the test, the screening pathway and Down's syndrome, it is for a professional to interpret results. DSS has received calls from women confused about what the results mean. We are therefore concerned about women who receive a positive result and go on to terminate/continue with the pregnancy, without access to accurate, current and balanced information.

3.3 If NIPT were to be incorporated into the Scottish NHS screening programme, healthcare professionals offering and providing prenatal screening and testing should have access to current information about Down's syndrome and should receive training which supports them in the delivery of that information to pregnant women and their partners.

3.3 Without accurate, current and balanced information from health professionals, pregnant women and their partners may not be able to make an informed choice. It is therefore vital that work is completed regarding the development of education resources and professional training prior to the implementation of NIPT within the NHS screening programme. The target group for the test and its purpose and accuracy **must** be explicit.

Mandatory training must:

- be delivered to all health practitioners involved in the screening process
- be included as part of their initial professional qualifications in universities and hospitals
- be part of the suite of Continual Professional Development courses
- include communicating with and counselling individuals about the implications of the screening prior to them embarking upon the process
- provide information about the condition which is current and balanced
- involve people with Down's syndrome/families so the trainee is able to understand that Down's syndrome is a spectrum condition and each person with Down's syndrome an individual

Conclusion

With the publication of the *Keys to Life strategy* in 2013, the Scottish Government demonstrated its commitment to improve the quality of life of people with learning disability and their families. DSS therefore believes that guarantees should also be put in place by the Scottish Government to ensure that health professionals can support prospective parents appropriately.

Down's Syndrome Scotland is calling for:

- 1) The establishment of a group to look into the development of quality information for prospective parents about the offer of screening and the screening pathway.**
- 2) The development of training resources for health professionals delivering care along the screening pathway, including the provision of accurate, balanced and current information about Down's syndrome.**
- 3) Assurances that private providers will adhere to agreed standards and that effective monitoring/regulations will be in place.**

We welcome the potential opportunity to work with the Scottish Government on this and will continue to offer to support expectant parents who contact the organisation during this time, through our family support service.

If you have any questions on this issue or if you would like to receive further updates in due course, please do not hesitate to contact our Policy Officer, Rachel Le Noan, on 0131 442 8849 or at Rachel@dsscotland.org.uk.