Nuffield Council on Bioethics

Call for views and evidence on non-invasive prenatal testing

Briefing Paper by the Christian Medical Fellowship

Screening for genetic conditions such as Down Syndrome now can be carried out by means of a simple blood test from ten weeks of pregnancy. It is more accurate than other early screening tests, and carries less risk of miscarriage. Nuffield are consulting on the ethical and practical implications of increased use of the tests in NHS screening programmes.[1] There is a short survey primarily for people with genetic conditions and their families, and a longer consultation for others

This briefing paper sets out some concerns with providing these tests as part of a nationwide NHS screening programme. CMF recommends that the cfDNA test[2] should not be introduced into the NHS. Efforts should instead focus on supporting children with Down syndrome and other trisomies, and their families.

General Concerns with implementing cfDNA testing

- Findings from a pilot study suggest that an additional 102 babies with Down Syndrome would be detected every year. On the basis that 90% of women will choose to abort,[3] this would mean that an additional 92 babies with Down Syndrome could be aborted each year.
- Non-invasive testing, which is currently available through private healthcare, has already increased the number of children with Down Syndrome being aborted.[4]
- Detection of gender and a range of other genetic 'conditions' and predispositions would be enabled and lead to more abortions. [5]
- Widespread use of cfDNA testing to analyse more and more genetic features would produce a mass of complex data, unreliable estimates of risk, and an increase of false-positives for abnormalities whose relevance is not known. These would require confirmation by invasive tests. Huge anxieties would be generated for parents; many would opt for invasive procedures, with a resultant increase in inadvertent miscarriages.
- The pressure to incrementally extend the availability of the test, towards the point where it becomes part of routine screening, would be hard to resist and certainly not cost-neutral.
- The routinisation and ease of accessing the tests is likely to lead to societal attitudes that it is a 'duty' to test rather than an option.

---

[1] http://nuffieldbioethics.org/project/non-invasive-prenatal-testing/

[2] This abbreviation refers to the new screening test in which cell-free DNA from the fetus can be identified and analysed in a sample of maternal blood

[3] Morris JK, Springett A. (December 2014) The National Down Syndrome Cytogenetic Register for England and Wales: 2013 Annual Report. http://www.binocar.org/content/annrep2013_FINAL.pdf

[4] New blood test blamed as women choosing to abort babies with Down syndrome and other serious disabilities soar in three years, The Daily Mail, 13 June 2015 http://dailym.ai/1efQRPw

[5] Current proposals are to screen only for trisomies 13,18 and 21. However it is likely that whole genome screening for over 6,000 conditions will be available (privately) in a few years time.

Legal concerns

- New procedures in public health programmes, such as this, need to be assessed against the Equality Act 2010. • WHO screening guidelines state that: 'there should be a treatment for the condition'.[6] However prenatal screening for Down's provides no benefit to the fetus as most will be aborted.
- The UN International Bioethics Committee warns against a 'culture of "perfectionism" or "zero defect"'[7] and inappropriate screening as a 'discriminatory practice that sends the message that these people are unwelcome in society'.[8]
- The UN Convention on the Rights of Persons with Disabilities (CRPD) calls for: 'respect for difference and acceptance of persons with disabilities as part of human diversity and humanity'; and for action to prevent discrimination within abortion law on the grounds of disability.[9]

The UN Convention on the Rights of the Child (UNCRC) states that a child 'needs special safeguards and care, including appropriate legal protection, before as well as after birth.'[10]

Concerns for mothers and their partners

- The test will create increased anxiety for those whose risk levels are considered just below the threshold level (currently 1:150 on initial screening) for the new test and for women confronted by ambiguous results.[11]
- There is a significant risk of mental health complications, following termination. Women who terminate pregnancies at advanced gestation for fetal anomalies experience grief as intense as those who experience spontaneous perinatal loss, with approximately a fifth developing major depression and/or requiring psychiatric intervention. [12]
- Participating in a programme of 'informal eugenics' would raise issues of conscience for many healthcare professionals.
- Ground E of the Abortion Act does not specify what amounts to 'serious handicap'. In practice it is left to individual clinicians to 'interpret' the results of screening tests to parents and to inform them of likely implications. It is likely that the introduction of widespread cfDNA testing will exacerbate the differences (and even conflict) of opinion between doctors concerning whether or not trisomies, and in particular Down Syndrome,fall within the scope of the law. The Parliamentary Inquiry into Abortion on the Grounds of Disability[13] found that many medical professionals assumed that parents ought to opt for abortion if their child was diagnosed with having a fetal disability.

Concerns for people with disabilities

---

[6] Wilson JMG, Jungner G. (1968) Principles and practice of screening for disease (large pdf). WHO Chronicle Geneva:World Health Organization. 22(11):473. Public Health Papers, #34.
[7] http://unesdoc.unesco.org/images/0023/002332/233258e.pdf, section IV, para 125
[8] Report on the IBC on Updating its reflection on the Human Genome and Human Rights, International Bioethics Committee, United Nations Educational, Scientific and Cultural Organization (UNESCO), Paris, 2 October 2015, page 22, para 88
http://unesdoc.unesco.org/images/0023/002332/233258E.pdf
[9] UN Convention on the Rights of Persons with Disabilities 2006, Article 3 http://www.un.org/disabilities/convention/conventionfull.shtml
[10] Preamble to the 1989 UN Convention on the Rights of the Child
[11] Research in the US found that conversations between patients and healthcare providers about screening for fetal aneuploidy lasted just 1.5 minutes on average *Obstetrics & Gynecology*, June 2016. http://bit.ly/29bAp2I
[12] Zeanah CH et al. Do women grieve after terminating pregnancies because of fetal anomalies? A controlled investigation. ObstetGynecol 1993;82(2):270-5 (August)
[13]http://cdn.thejournal.ie/media/2016/06/abortion-and-disability-report-17-7-13.pdf

- The tests will have a profound and discriminatory effect on the community of people with Down Syndrome as a result of increased selective screening and elimination of babies diagnosed with a risk of having Down Syndrome.
- These tests foster the notion that the lives of people with disability are 'not worth living' and that it is socially desirable to prevent people with disabilities from being born.[14]The correct response is to provide better support for families and children. Article 23 (3) of the CRPD calls for early and comprehensive support for children with Down's, and their families.

We recommend:

- The cfDNA test[15] should not be introduced into the NHS. Efforts should instead focus on supporting children with Down syndrome and other trisomies, and their families

For existing screening tests, we recommend:

- Provision for informed 'reflection before decision' for every woman receiving the news that she may be carrying a baby with Down Syndrome (and other trisomies). Opportunities for every parent to meet a family who has a child with that diagnosis or a similar condition.[16]
- Ensuring that printed information written by those who have the same disabilities, and their families, is given to every woman following any screening test.
- Information provision to be neutral, balanced, compassionate and well-informed.
- Improved training for medical professionals so there is no presumption, if disability is detected, that parents will automatically want an abortion.
- Ensure that signposting to telephone and online helplines manned by trained professional counsellors, and to local and national support groups for those with specific conditions, is given to every woman.
- Challenging societal attitudes to disability. Disabled people say they have a quality of life as good as people who are not disabled and often possess an inner strength and resilience that able-bodied people do not have.

Following her decision, and regardless of what choice the woman and her family may make, ongoing support must be part of that provision.

Public Policy Department
July 2016

[14] Wyatt, J. 2001. 'Medical paternalism and the fetus'. Journal of Medical Ethics. http://jme.bmj.com/content/27/suppl_2/ii15.full
[15] This abbreviation refers to the new screening test in which cell-free DNA from the fetus can be identified and analysed in a sample of maternal blood
[16] Parens E and Asch A. The Disability Rights Critique of Prenatal Genetic Testing: Reflections and Recommendations. Hastings Center Report 1999; 29(5): S1-S22