Genetic Alliance UK
Supporting. Campaigning. Uniting.

Consultation response

Cell-free DNA testing in the first trimester in the Fetal Anomaly Screening Programme

Response from Genetic Alliance UK, 2nd October 2015

Introduction

1. Genetic Alliance UK is the national charity supporting all those affected by genetic conditions. We aim to improve the lives of people affected by genetic conditions by ensuring that high quality services and information is available to all who need them. Our membership represents more than 180 voluntary organisations working for a wide range of conditions, many of which pose complex health and social care needs. We actively support research and innovation across the field of genomic medicine.

2. Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK, working towards the delivery and implementation of a national strategy for rare diseases in the UK. At least 80% of rare diseases have an identified genetic origin. The UK Strategy for Rare Diseases[1] was published by the Department of Health in November 2013. Pertinent to this consultation, in this strategy all four Governments of the UK committed to:

   "Continue to work with the UK National Screening Committee to ensure that the potential role of screening in achieving earlier diagnosis is appropriately considered in the assessment of all potential new national screening programmes and proposed extensions to existing programmes"
   Commitment 9, UK Strategy for Rare Diseases, November 2013

3. We welcome the recommendation by the UKNSC to introduce screening for trisomy 21, 18 and 13 in women found to have a combined test risk score equal or greater than 1 in 150. The recommendation to introduce non-invasive prenatal testing as part of the National Health Service represents a step towards further equality in reproductive autonomy.

The value of cell-free DNA testing to women and couples

4. Couples' views and experience of non-invasive prenatal testing have been shown to be overwhelmingly positive. Women who have received news that their pregnancy was affected were found to be equally positive towards the technology as those who received good news[2].

5. Invasive testing is associated with a risk of miscarriage (around 0.5-1in 100). Under the current screening programme only 5-10% of the population that undergoes invasive testing is found to have an affected pregnancy. The reduced risk of miscarriage is one of the most important, positive, aspects of the cfDNA testing for women. Women whose cfDNA test was positive for a

[1] UK Strategy for Rare Diseases. Department of Health, published November 2013, available at: www.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf
[2] Lewis. C et al (2012) *Fetal sex determination using cell-free DNA: service users' experiences of and preferences for service delivery* in Prenatal Diagnosis.

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trisomy will still have to undergo invasive testing, but the use of the non-invasive test used in the interim between the combined test and invasive test allows women to make the decision to undergo invasive testing with more accurate information. This will lower the number of women with an unaffected pregnancy undergoing invasive testing.

6. Women value the opportunity to have tests earlier, as it gives couples more time to make decisions about their pregnancy, bringing substantial psychological benefits. Women report feeling in control of the pregnancy, and having time to prepare themselves for what is to come. The test also gives women, whether found to be at risk or not, peace of mind much earlier on in their pregnancy.

Implementation

7. While we support the use of cfDNA testing for women at risk of T21, T18 and T13, it is important to make sure that testing is done in an appropriate way.

8. Non-invasive testing should be offered through specialised services. Patients have shown a preference for receiving pre and post test counselling from a specialist genetic counsellor, with specialist knowledge about the particular condition.

9. Some have argued that as tests such as this become routine in clinical practice they become normalised and present potential concerns for informed consent. This can easily be overcome by the presence of appropriate, detailed and non-directive counselling, which impresses on couples thinking about undertaking cfDNA testing, the impact that the results of this test may have on their lives. Couples should be given them all the information available to them as well as the space to make an informed decision.

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