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Noninvasive prenatal testing: Effective, safe, preferred by parents

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Non-invasive prenatal testing (NIPT) for Down's syndrome is feasible, acceptable to parents, and could be introduced into the National Health Service (NHS), UK researchers say. The results of a National Institute for Health Research (NIHR) study carried out by the first NHS laboratory to provide NIPT testing will be reported to the annual conference of the European Society of Human Genetics today (Saturday).

Presenting her team's findings, Professor Lyn Chitty, from the UCL Institute of Child Health and Great Ormond Street Hospital, London, UK, will announce the results of the study evaluating the possibility of introducing NIPT into the NHS screening programme for Trisomy 21 (Down's syndrome). The researchers will present their study to the UK National Screening Committee later this month and hope that it will inform their decisions on if and how to implement NIPT in the NHS.

As part of the study, carried out by the NHS laboratory at Great Ormond Street Hospital, women at high and medium risk of having a child with Down's syndrome were offered NIPT, and over 2,500 undertook the test.

Prof Chitty says: "There was a very high uptake of testing and we saw invasive test numbers fall sharply. NIPT performed well in identifying problems, and women were very positive about it.

"The cost of providing an NIPT service will depend on the cost of the test itself and how it is implemented. There will be significant savings

resulting from a decrease in invasive testing whilst increasing the detection of affected babies. The reduction in invasive testing also means there will be a reduction in miscarriages and loss of unaffected babies which is much better for parents."

Commenting on NIPT, a woman classified as 'high risk' who was involved in the study said: "You get told 1 in 30 and although that sounds relatively high...we probably wouldn't have done [invasive testing] because there's a risk of miscarriage. ..I think that we were very lucky. It's enabled us to make an informed choice about what happens for the rest of our lives."

Another woman involved in the study said: "I think it's a real advancement. At the moment, if you are put in a high risk category you're automatically offered the invasive test, whereas this will reduce the amount of invasive tests that need to take place."

Reporting the results of a second study from the same group, Dr Suzanne Drury, a translational research and development scientist from Great Ormond Street Hospital, will describe the team's experience in the use of NIPD (non-invasive prenatal diagnosis) to diagnose the disorder congenital adrenal hyperplasia (CAH). CAH exposes a female fetus to male hormones, which can result in the development of masculinised external genitalia. It is an autosomal recessive (AR) disorder, in which the defective gene must be passed on from both parents in order to cause disease.

Dr Drury will say: "We chose CAH because the gene that causes it is particularly challenging to study. It is the most common adrenal disorder in childhood and affects one in every 18,000 live births. In the UK, NIPD for fetal sex determination is carried out for an average of 13 pregnancies per year at risk of CAH because it is the female fetuses that are at risk.

"Fetal sex determination allows targeting of invasive testing to see if the female fetus is carrying two mutant copies of the CAH gene and is therefore affected. As we were already carrying out NIPD for sex determination, and there is a potential in utero treatment for CAH available, we felt that this was a good condition to select to allow treatment to be very specifically targeted to only those female fetuses that are affected."

In 2014, the researchers say, 32% of prenatal diagnostic tests for monogenic disorders in their laboratory were non-invasive. NIPD for single gene disorders in a fetus is diagnostic, as it targets specific genetic changes present in a high risk family. For this reason it will remove the need for invasive testing completely, reducing the risk of miscarriage and making prenatal diagnosis for these conditions safer and more accessible to families who would not otherwise be prepared to take the risk.

Dr Drury adds: "Our results have shown NIPD to be sufficiently precise to be diagnostic and therefore we do not recommend confirmatory invasive testing. Currently we are developing non-invasive tests for other conditions caused by mutations in a single gene, including cystic fibrosis, sickle cell anaemia, and beta-thalassaemia. At present invasive testing is required for definitive prenatal diagnosis of these disorders, but our experience with CAH leads us to believe that NIPD will have the same diagnostic efficacy in other AR disorders."

Provided by European Society of Human Genetics