First national study of noninvasive prenatal testing shows it works

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Results from a national study of non-invasive prenatal testing (NIPT) in women at high risk of having a baby with Down's syndrome will be presented at the annual conference of the European Society of Human Genetics today (Saturday). The Netherlands is the first country in the world to include NIPT in a government supported, healthcare-funded trisomy syndrome screening programme. In many other countries, such screening is offered by commercial companies and without governmental guidance, so studying the accuracy of the programme and its acceptability to prospective parents was important, the conference will hear.

Dr Erik Sistermans, head of the Genome Diagnostics department, VU Medical Centre Amsterdam, The Netherlands, will present results from the TRIDENT (Trial by Dutch Laboratories for Evaluation of Non-Invasive Prenatal Testing) study, of which he is the project leader. Together with colleagues from other academic centres in The Netherlands, he formed a consortium including all stakeholders - gynaecologists, clinical geneticists, midwives, and laboratory specialists - to apply for the ministerial licence which is needed to screen for untreatable disorders such as Down's.

"The study was in two parts; first concerned with its implementation in eight university laboratories, and the second to examine the perspective of pregnant women. After receiving positive advice from the Health Council, a temporary licence for two years was granted allowing us to enrol women with an elevated risk of carrying a foetus with three syndromes caused by the presence of an abnormal number of chromosomes - Down's (trisomy 21), Edward's (trisomy 18), and Patau (trisomy 13)," Dr Sistermans will say. Entry to the study was based on the results of a combined test, taking account of maternal age and involving a blood test and a nuchal fold measurement - a measurement of a skin fold in the neck of the foetus.

After nine months, 3306 high-risk pregnant women had undergone NIPT and 3278 reports on the status of the foetus issued. The researchers found 89 cases of trisomy 21, 11 of trisomy 18, and ten of trisomy 13. Follow up of 103 cases revealed only nine false positives.

Although NIPT carried a much lower risk of miscarriage than do invasive tests, it is slightly less accurate, because it only analyses DNA from the outer layers of the placenta. In some cases a trisomy will be present in these outer layers, but not in the foetus. "For this reason a positive NPT result should always be followed by an invasive test," says Dr Sistermans.

"Currently the costs for NIPT and invasive tests are about the same, but we believe that with a greater uptake of NIPT the cost will fall. Given that the vast majority of the women we surveyed preferred to undergo NIPT in order to be more certain of avoiding risks to the child, it seems likely that the demand for this test will increase over the years to come."

The researchers now expect the Dutch Heath Ministry to continue the offer of NIPT as a second test to women at high risk of carrying a child with a trisomic disorder. "Whether NIPT will be offered as a first screening test to all pregnant women is currently under debate, and will depend partly on the reaction of the health ministry to our study. Based on our findings, the TRIDENT consortium believes this test can benefit all pregnant women, not just those who are identified as being at risk, and has applied for a licence to offer it to them," Dr Sistermans will conclude.

Provided by European Society of Human Genetics

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