

DNA test for Down's syndrome on the NHS could save lives

Offering pregnant women a DNA test for Down's syndrome on the NHS would reduce the dependency on invasive tests and potentially save the lives of unborn babies, a study shows.

The research – led by Professor Kypros Nicolaides and his team at King's College Hospital and King's College London – is published in the *Ultrasound in Obstetrics and Gynecology* journal today (Monday 26 October).

In the NHS study, of those pregnant women judged to be at high risk for carrying a baby with Down's syndrome, a far greater number opted for the 'cell-free' DNA test compared to those requesting invasive testing. 60% of women in the study chose the cell-free DNA test, compared to 38% opting for invasive testing.

At present, a woman's risk of carrying a baby with Down's syndrome is based on an assessment of her age, the level of hormones in her blood and the results of a 'nuchal scan', which measures fluid collection at the unborn baby's neck.

Women deemed to be high risk following this 'combined assessment' are given the option of further tests, which involve using a needle to take a sample of the placenta (chorionic villous sampling, or CVS) or the fluid that surrounds the baby (amniocentesis). CVS or amniocentesis are still the only tests that can provide a definitive diagnosis of Down's syndrome, but both carry a risk of miscarriage.

The study involved 11,692 women with single pregnancies treated at two different NHS hospitals; King's College Hospital, London, and the Medway Maritime Hospital, Kent. A small proportion of these women (395) were found to be at high-risk for Down's syndrome.

The cell-free DNA test – which involves a far less-invasive blood test of the pregnant woman – is a highly reliable test for Down's syndrome, but is currently only available privately and costs hundreds of pounds.

The results of the study suggest that the best approach to screening for Down's syndrome is to continue to offer the combined test to all pregnant women, as is standard currently, but to begin offering the cell-free DNA test to all women judged to be at an increased risk.

As well as showing the cell-free DNA test is the preferred option for pregnant women, the study also provides evidence that the test can be easily incorporated into routine clinical care within the NHS. Introducing the cell-free DNA test on the NHS would lead to a higher proportion of affected pregnancies being identified, resulting in a small increase in the overall cost to the NHS, but a major decrease in invasive tests.

Crucially, the study also showed that although Down's syndrome could be detected in more babies, this did not translate into a higher number of pregnancy terminations. Many women want to know if their baby is affected so that they can make an informed decision for themselves and their family.

Professor Kypros Nicolaides, Director of the Harris Birthright Research Centre for Fetal Medicine at King's College Hospital, and Professor of Fetal Medicine at King's College London, said:

"Our research puts the case for offering the cell-free DNA test on the NHS. This would improve the performance of screening, and reduce the number of unnecessary invasive tests and miscarriages."

ENDS

For more information contact ISUOG Secretariat at:

E: info@isuog.org

T: +44 (0)20 74719955

Or visit our website: isuog.org

Notes to Editors

Article:

Clinical implementation of routine screening for fetal trisomies in the UK NHS: cell-free DNA test contingent on results from first-trimester combined test.

Ultrasound in Obstetrics & Gynecology; Published early online, 26/10/15

Follow link for full article:

<http://onlinelibrary.wiley.com/doi/10.1002/uog.15783/abstract>

About ISUOG:

1,400 women die from pregnancy related causes every day, worldwide. The International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) is dedicated to ensuring that all women have access to competent ultrasound and that obstetric and gynecological conditions are effectively diagnosed. ISUOG delivers high quality learning through its education program and World Congress and disseminates research information and clinical guidance through its journal *Ultrasound in Obstetrics & Gynecology*. With more than 13,500 members across 128 countries, ISUOG is committed to improving health outcomes for women and their families across the world.

About Wiley:

Wiley is a global provider of content-enabled solutions that improve outcomes in research, education, and professional practice. Our core businesses produce scientific, technical, medical, and scholarly journals, reference works, books, database services, and advertising; professional books, subscription products, certification and training services and online applications; and education content and services including integrated online teaching and learning resources for undergraduate and graduate students and lifelong learners.

Founded in 1807, John Wiley & Sons, Inc. (NYSE: JWa, JWb), has been a valued source of information and understanding for more than 200 years, helping people around the world meet their needs and fulfill their aspirations. Wiley and its acquired companies have published the works of more than 450 Nobel laureates in all categories: Literature, Economics, Physiology or Medicine, Physics, Chemistry, and Peace. Wiley's global headquarters are located in Hoboken, New Jersey, with operations in the U.S., Europe, Asia, Canada, and Australia. The Company's website can be accessed at <http://www.wiley.com>.