DNA blood test can cut invasive testing for Down's syndrome by 98 percent

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Women in high risk pregnancies for Down's syndrome could have a DNA blood test to detect the disorder and avoid invasive procedures such as amniocentesis or chorionic villus sampling, finds a large scale study published in the British Medical Journal today.

The blood test could mean that 98% of invasive procedures could be avoided, say the authors. The test uses the latest DNA technology to analyse genetic components in the mother's blood that indicate whether the foetus has Down's.

Down's syndrome or trisomy 21 occurs in around 1 in 800 births and older women are at higher risk.

Women in high risk groups tend to undergo a combination of scans and hormone level tests in order to determine if they need to have an invasive test such as amniocentesis or chorionic villus sampling. The latter tests take samples of genetic material from the foetus but they carry a 1% risk of miscarriage and are therefore reserved for high risk pregnancies. Invasive testing still takes place in 3 to 5% of pregnant women in the UK.

The research team, led by Professor Dennis Lo from The Chinese University of Hong Kong, used the most up-to-date DNA technology to test the blood samples from 753 pregnant women (all were at high risk of having a baby with Down's) based in Hong Kong, the UK and the Netherlands. Eighty-six of the women were found to be carrying a foetus

with Down's syndrome.

The results show that the test is highly accurate in detecting Down's syndrome in unborn babies and does not give false negative results.

The authors conclude that the blood test could be used to accurately rule out Down's syndrome among high risk pregnancies before amniocentesis or chorionic villus sampling is considered. In this way the number of women requiring invasive procedures can be reduced.

Provided by British Medical Journal