Non-invasive first trimester blood test reliably detects Down's syndrome

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Cell-free fetal DNA testing, which measures the relative amount of free fetal DNA in a pregnant woman's blood, is a new screening test that indicates the risk of Down syndrome (trisomy 21), Edward syndrome (trisomy 18), and Patau syndrome (trisomy 13).

A recent analysis of 37 published studies shows that the test can detect more than 99% of Down syndrome cases in singleton pregnancies, with a very low false positive rate of less than 0.1%. This makes it superior to all other testing methods.

The test is much less accurate for Edward syndrome and Patau syndrome, however, with respective detection rates of about 96% and 92% and a false positive rate of 0.26%. The analysis is published in *Ultrasound in Obstetrics & Gynecology*.

More information: Gil, M. M., Quezada, M. S., Revello, R., Akolekar, R. and Nicolaides, K. H. (2015), Analysis of cell-free DNA in maternal blood in screening for fetal aneuploidies: updated meta-analysis. *Ultrasound Obstet Gynecol*. DOI: 10.1002/uog.14791

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