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New research has found that **routine screening** using a **non-invasive test** that **analyzes fetal DNA** in a **pregnant woman's blood** can accurately **detect Down's syndrome and other genetic fetal abnormalities** in the **first trimester**

. Published early online in

Ultrasound in Obstetrics & Gynecology

, the results suggest that the test is superior to currently available screening strategies and could reshape standards in prenatal testing.

Current screening for Down's syndrome, or trisomy 21, and other trisomy conditions includes a combined test done between the

11th and 13th weeks of pregnancy, which involves an ultrasound screen and a hormonal analysis of the pregnant woman's blood. Only chorionic villus sampling and amniocentesis can definitely detect or rule out fetal genetic abnormalities, but these are invasive to the pregnancy and carry a risk of miscarriage.

Several studies have shown that non-invasive prenatal diagnosis for trisomy

syndromes using fetal cell free (cf) DNA from a pregnant woman's blood is highly sensitive and specific, making it a potentially reliable alternative that can be done earlier in pregnancy.

An Ultrasound in Obstetrics & Gynecology study by

Kypros Nicolaides, MD,
of the Harris Birthright
Research Centre for
Fetal Medicine at
King's College London
in England, and his
colleagues is the first to
prospectively
demonstrate the
feasibility of routine
screening for trisomies

21, 18, and 13 by
cfDNA testing. Testing
done in 1005
pregnancies at 10
weeks had a lower
false positive rate and
higher sensitivity for
fetal trisomy than the
combined test done at
12 weeks. Both cfDNA
and combined testing

detected all trisomies, but the estimated false-positive rates were 0.1% and 3.4%, respectively.

“This study has shown that the main advantage of cfDNA

testing, compared with the combined test, is the substantial reduction in false positive rate.

Another major advantage of cfDNA testing is the

reporting of results as very high or very low risk, which makes it easier for parents to decide in favor of or against invasive testing," the authors wrote.

A second *Ultrasound*
in Obstetrics
& Gynecology
study by the
group, which
included
pregnancies
undergoing

screening at
three UK
hospitals
between March
2006 and May
2012, found that
effective
first-trimester

screening for
Down's
syndrome could
be achieved by
cfDNA testing
contingent on
the results of the
combined test

done at 11 to 13 weeks. The strategy detected 98% of cases, and invasive testing was needed for confirmation in

less than 0.5%
of cases.

“Screening for
trisomy 21 by
cfDNA testing

contingent on
the results of
an expanded
combined test
would retain
the advantages
of the current

method of
screening, but
with a
simultaneous
major increase
in detection
rate and

decrease in the rate of invasive testing,” the authors concluded.

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