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Experimental Prenatal Test Helps Spot Birth Defects

It's noninvasive, but don't expect to see it on the market soon

By **Amanda Gardner**
HealthDay Reporter

FRIDAY, Feb. 2 (HealthDay News) -- A new, noninvasive method of prenatal testing may one day help to detect birth defects in unborn babies.

But it'll be a while before the technique comes to market.

"We've shown the first abnormality, and this is proof of principle to show that this technology can be a noninvasive test from maternal blood," said study author Dr. Ravinder Dhallan, founder and chief executive officer of Ravgen Inc., in Columbia, Md. "Next, we will do a larger study and start building the infrastructure to go to market."

Dhallan wouldn't speculate on when such a test might become commercially available, saying only, "We don't intend to rush it. The health of a baby is the most important thing in anyone's life, but we are moving with deliberate speed."

Dr. Michael Katz, senior vice president for research and global programs at the March of Dimes, called the experimental test a "good system if it can be sustained by more extensive tests. It also dovetails with the current suggestion by the American College of Obstetricians and Gynecologists that prenatal diagnosis should be applied to all women."

The study findings are published online Feb. 2 in *The Lancet*.

Currently, tests to detect chromosomal abnormalities before a baby is born pose a number of problems.

Ultrasound is noninvasive but does require subsequent invasive testing before a final diagnosis can be made. And invasive diagnostic tests, such as amniocentesis and chorionic villus sampling, can pose risks to the pregnancy, including miscarriage.

"Even though the current invasive tests are quite safe, relatively speaking, they are invasive, and there are complications," Katz said. "Moreover, they have to be done somewhat later [in the pregnancy], and the sooner one makes this diagnosis, the better."

Scientists have known for several decades that fetal cells are present in the mother's blood as early as five weeks into the pregnancy. The problem is that the cells are extremely rare, representing only about one in a million of total cells.

The reason for their rarity? The mother's cells are very much like water balloons: From the moment blood is drawn, her cells burst and let loose maternal DNA into the blood stream. This serves to dilute the fetal DNA, Dhallan explained.



"Every time a mother's cell bursts, it dilutes the fetal DNA, particularly when you transport samples and when you centrifuge and process samples to get the DNA out of the plasma," he said.

In March 2004, Dhallan solved that problem in a study that showed that adding formaldehyde to the blood sample caused the mother's cells to harden ("like Ping-Pong balls"), reducing dilution and increasing fetal DNA to 25 percent.

The next challenge was how to distinguish maternal DNA from fetal DNA and how to identify abnormalities.

For the new study, the researchers examined blood samples from 60 pregnant women and the "stated biological fathers," and analyzed single nucleotide polymorphisms (SNPs or "snips"), the tiny variations in the DNA sequence that exist between individuals.

There are about 3 million variable sites where human DNA differs from person to person, out of a total of 3 billion base pairs. "The key is finding where they differ," Dhallan said.

Combining mathematics and genetics technology, the researchers determined the ratio (between mother and child) of SNPs on different chromosomes. That ratio should be the same for all chromosomes. "If it's not, you have a problem," Dhallan said.

The method correctly identified the number of chromosomes in 58 of 60 samples, including two cases of trisomy 21 (which causes Down syndrome), Dhallan said. One case of trisomy 21 went undetected, while one normal sample was incorrectly identified as trisomy 21.

Katz said: "They have one mistake one way and one the other way. That's a little too risky if you compare this with invasive techniques where you have virtually 100 percent accuracy. Also, one can never be absolutely certain that this is the biological father. In addition, there may be some times when the father is not available."

A second study, this one appearing in the Feb. 3 *British Medical Journal*, questions the rise of "boutique ultrasonography," where commercial companies offer "keepsake" scans to expectant parents without medical supervision.

Three- and four-dimensional, as well as moving images, can sell for up to \$500, the study author stated.

While the companies claim the ultrasound poses no danger to mother or baby, several official organizations, including the American Institute of Ultrasound in Medicine and the U.S. Food and Drug Administration, have expressed concerns about the practice.

More information

To learn more about the types of prenatal tests that are currently available, visit the [March of Dimes](#).

SOURCES: Ravinder Dhallan, M.D., Ph.D., founder and chief executive officer, Ravgen Inc., Columbia, Md.; Michael Katz, M.D., senior vice president for research and global programs, March of Dimes, White Plains, N.Y.; Feb. 2, 2007, *Lancet*, online; Feb. 3, 2007, *British Medical Journal*

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