

## Blood test detects Down syndrome during pregnancy

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Second company develops noninvasive screen for condition.

(HealthDay) -- A second company reports that it has developed a prenatal blood test to detect Down syndrome, potentially providing yet another option for pregnant women who want to know whether their unborn child has the condition.

Last fall, Sequenom Inc. announced that it was making a prenatal Down syndrome <u>blood test</u>, available in 20 cities in the United States. It marked the first time that pregnant women could undergo a Down syndrome test without having to go through <u>amniocentesis</u> or chorionic villus sampling, which are invasive and pose a small risk of miscarriage.

Now, two studies published online Feb. 21 and in the April print issue of the <u>American Journal of Obstetrics and Gynecology</u> suggest that another



blood test, this one developed by Aria Diagnostics, can detect Down syndrome and a genetic disorder known as Edwards syndrome, which can cause severe birth defects and is often fatal. In both studies, testing was conducted before 20 weeks gestation.

One of the studies, written by Aria Diagnostics researchers, correctly detected 44 cases of the two disorders out of 167 prenatal blood samples. The other study, by researchers from University of London and University College London, correctly detected all Down syndrome cases and 98 percent of Edwards syndrome cases.

The test "would be useful as a secondary test contingent upon the results of a more universally applicable primary method of screening," senior study author Dr. Kypros H. Nicolaides, of the University of London, said in a journal news release. "The extent to which it could be applied as a universal screening tool depends on whether the cost becomes comparable to that of current methods of sonographic and biochemical testing."

Dr. Brian Skotko, a physician with the Down Syndrome Program at Children's Hospital Boston, said the test's accuracy is "pretty good," although the studies didn't test as many samples as Sequenom did for its test.

Several other companies are developing prenatal tests for Down syndrome, said Skotko, who predicted that competition in the prenatal blood test market would lead to lower prices. Sequenom has said that its test won't cost mothers more than \$235 in out-of-pocket costs.

The growth of these kinds of tests raises major questions, Skotko said: Will the tests become routine? If so, "will babies with Down syndrome slowly start to disappear?"



Some pregnant mothers choose to abort their unborn children after they are diagnosed with Down syndrome. Statistics suggest that their numbers have risen in recent decades, Skotko said.

**More information:** For more on Down Syndrome, go to <u>U.S. National</u> <u>Library of Medicine</u>.

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