

Comfort or conflict: Earlier Down syndrome test

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In this June 6, 2011 photo, Erin Witkowski, of Port Jervis, N.Y., and her 16-month-old son Grady pose for photos in New York's Central Park. Witkowski underwent a test to see if the baby she was carrying had Down syndrome. When the test came back positive, her doctor started talking immediately about abortion, a step Witkowski rejected. She changed doctors and gave birth to Grady in February 2010. In 2012, there may be an alternative _ a simple blood test that offers accurate results weeks before a pregnancy is obvious to others. (AP Photo/Richard Drew)

(AP) -- The results of the blood test revealed only a risk, but when she saw them, she still threw up. Now she had to find out for sure.

So she lay on her back at a doctor's office, praying, comforted by her Christian faith and her mother at her side, while a needle was slipped into her belly.

Erin Witkowski of Port Jervis, N.Y., was going to find out if the baby she was carrying had Down syndrome.

This is the first of a two-part series on [prenatal testing](#) and the [ethical issues](#) raised by it. The second part is [here](#).

For years, many [women](#) have gone through an experience like hers: a blood or ultrasound test that indicates a heightened risk of the syndrome, followed by a [medical procedure](#) to make a firm diagnosis by capturing DNA from the fetus.

Usually it's the needle procedure Witkowski had, called amniocentesis, done almost four months or more into the pregnancy. Sometimes it's an earlier test called CVS, or chorionic villus sampling, which collects a bit of tissue from the placenta. Both pose a tiny but real chance for miscarriage, and experts say highly skilled practitioners are not available everywhere.

But by this time next year there may be an alternative - one that offers accurate results as early as nine weeks into the pregnancy.

Companies are racing to market a more accurate [blood test](#) than those available now that could spare many women the need for an amnio or CVS. It would retrieve fetal DNA from the mother's bloodstream. And the answer could come before the pregnancy is obvious to others. For some women, that might mean abortion is a more tenable choice. For others it could be a mixed blessing.

Down syndrome slows mental and physical development, and people

with it usually show mild to moderate disability in intellect and skills for everyday living. Physically, they often have a flat face with a short neck and smaller hands and feet. They're at risk for complications like [heart defects](#) and hearing problems. Life expectancy is about 60 years.

Most cases are diagnosed after birth now, but if the blood test is widely adopted it could become chiefly a prenatal event.

A diagnosis before birth can pose a difficult challenge for couples as they decide whether to continue the pregnancy. It's not only about child-rearing, but also about what happens as the child grows into an older adult and may need care that the aging parents struggle to provide, says Dr. Mary Norton, a Stanford University professor of obstetrics and gynecology.

Dr. Brian Skotko, a Down syndrome specialist at Children's Hospital Boston who has written a research paper for doctors on how to deliver a diagnosis, said "the vast majority of people with Down syndrome and families affirm that their contributions to their communities are significant, and their lives are very valuable."

Current prenatal screening has already cut into the number of babies born with the syndrome, which now stands at about 6,000 each year in the United States, or about 1 in every 691 babies, says Skotko, who serves on the board of the National Down Syndrome Society. He cites one study that concludes the number of Down syndrome births in the nation dropped 11 percent between 1989 and 2006, a time when it would otherwise be expected to rise 42 percent.

Initially, doctors are expected to use the new blood test with women at risk for a Down syndrome pregnancy, such as those older than 35. A negative result would indicate a woman could skip the amnio or CVS; a positive result would suggest she get one done to be sure.

Eventually it might replace the routine screening tests offered to all pregnant women. Since the test sounds fewer false alarms than current tests, fewer women would be told they need the invasive follow-up procedures, experts say. And some suggest that with further fine-tuning, it could largely replace amnio and CVS. With no miscarriage risk, more women might be willing to take it, and so more women would find out they have a Down syndrome pregnancy.

Two California companies, Sequenom Inc. and Verinata Health Inc., hope to offer the test to doctors in the United States by next April. They say it could be done in the first trimester, with Sequenom aiming as early as 10 weeks, and Verinata as early as eight weeks. Results would be available 7 to 10 days later. In addition, LifeCodexx AG of Germany says it wants to start offering its test in Europe by the end of this year, to be performed at 12 to 14 weeks initially. None of the companies would discuss its cost.

"I would have definitely taken a noninvasive test over the struggle for deciding whether to do an amnio or not," says Nancy McCrea Iannone of Sewell, N.J., who gave birth six years ago to a daughter with Down syndrome. She'd been alerted by screening results, but hesitated to get amniocentesis because of the risk of a miscarriage and the prospect of "a needle in my belly," she recalls. Ultimately, she did have one.

Iannone now counsels women who plan to deliver babies with Down syndrome. Her charitable group, Down Syndrome Pregnancy Inc., lists several reasons for getting a diagnosis before birth, such as more time to adjust, grieve and learn about the condition, preparing friends and family, and checking on available medical care and insurance.

But detecting the condition earlier in a pregnancy through the new blood test would be a mixed blessing, Iannone said.

The time between diagnosis and birth is "an unnatural state," she said, and "the longer that time period is, the harder it is."

"All you know is that they have Down syndrome. You're coping with that diagnosis in a vacuum, without a baby... It's fear of the unknown, you haven't met your baby yet. You spend a lot of time worrying."

That might weigh heavily on women who haven't decided whether to continue the pregnancy or not, she said.

Since the new blood test could deliver an answer so early - before a pregnancy is showing or the baby is kicking - it might make getting an abortion easier, several observers said. Women haven't bonded so much, and "they wouldn't have to explain to as many people," said Christie Brooks, who moderates an online support group for women who've gotten abortions for medical reasons.

"No one needs to know you're pregnant," said Skotko. "Maybe you haven't even told your husband."

Skotko said he respects that a woman's right to continue or terminate a pregnancy is a personal one for couples. But he's concerned that in the case of Down syndrome, many women may be getting bad information about what having the baby would mean. And if the new test became routine it would only exacerbate that problem, he said.

Studies show medical students are poorly trained about people with disabilities and that some doctors who make a prenatal diagnosis emphasize negative information about the condition, he said.

"We have a fleet of physicians who are saying they're untrained, unprepared and sometimes knowingly inserting their own personal bias," he said. "How are women today able to make a truly informed

decision?"

Others say the blood test could thrust some women into a choice they didn't ask to make.

Hank Greely, a Stanford University law professor, said women sign forms for plenty of blood tests during prenatal care and often don't focus on them. Many California women are surprised to learn they'd authorized the screening test for Down syndrome, he said.

If these tests are someday replaced by the new blood test, many women may be told out of the blue not simply that they're at risk, but that in fact their baby almost surely has Down syndrome, Greely says.

"They're going to jump directly to the final answer, which is not necessarily something they wanted to get," Greely said. So the new test poses a challenge to the medical establishment about how to assure that women get adequate counseling to make an informed choice, he said.

Witkowski, who prayed as that needle was slipped into her swollen belly in 2009, got her answer: It was [Down syndrome](#). As her doctor gave her the news, her baby kicked her and "I could see my belly move," she recalled.

Her doctor started talking immediately about abortion, a step Witkowski rejected. She changed doctors and gave birth to Grady in February 2010.

"When they first gave him to me," Witkowski said, "I saw tiny little hands, and he had the most beautiful eyes... He didn't have 'Down syndrome' stamped on his forehead. He cried and he peed and he pooped. He was a baby."

More information: Current prenatal tests: <http://bit.ly/iev0ZG>

National Down Syndrome Society: <http://www.ndss.org/>

Erin Witkowski's blog: <http://skiingthroughlife.wordpress.com/>

Down Syndrome Pregnancy Inc.: <http://downsyndromepregnancy.org>

Brooks' group about pregnancy termination: <http://bit.ly/jJzQuy>

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