

Simple test, complex questions

Mothers seek peace of mind, but critics raise spectre of eugenics

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CAROLYN ABRAHAM
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Learning about the genetic health of an unborn child could soon be as simple as giving blood.

A new prenatal test is slated to hit the market this summer that requires nothing more than a sample of a pregnant woman's blood for doctors to analyze the DNA of her developing fetus.

Current methods to collect fetal DNA, such as amniocentesis, involve an intrusion into the uterus that can trigger a miscarriage – a risk that makes many couples refuse the procedure. But after 30 years of effort, science appears to be on the cusp of delivering a safe, non-invasive test that can detect Down syndrome and other genetic conditions by capturing the minute bits of fetal DNA in a pregnant woman's bloodstream.

Yet as with most advances in reproductive medicine, the new technology is raising tricky social questions.



Supriya James, who is 19 weeks pregnant, hugs her 5-year-old daughter, Veda. (*Deborah Baic/The Globe and Mail*)

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While some see it as a better way to prepare parents and hospitals to care for newborns with special needs, others fear it smacks of eugenics as science makes it ever easier to reject a less-than-perfect baby.

Sequenom Inc., a biotech firm based in San Diego, says it will begin selling the first non-invasive prenatal genetic test, pioneered at the University of Oxford, online in June. Meanwhile, researchers at Stanford University have developed a similar version they expect will be available within two years.

Neither test has been tried in large patient studies to evaluate accuracy. But if the promising preliminary results hold up, experts say the tests, which can be performed early in pregnancy, will revolutionize the field.

“This is a powerful technology,” said Doug Wilson, head of the genetics committee of the Society of Obstetrics and Gynaecology of Canada.

“If it can be proven to be as accurate [as current diagnostic methods] it will become the new diagnostic gold standard.

“If it can be done at 10 weeks, instead of 16 weeks, it will relieve the stresses of pregnancy early.”

At the Canadian Down Syndrome Society, however, news of the technology coming to market has only added to stress levels.

“People with Down syndrome are very concerned about the emergence of tests that aim to eliminate them from the world,” said the society's executive director, Krista Flint.

“Down syndrome is a bellwether – if this is an easy test, it is just a matter of time before it's used to determine other things, and sometimes just because we can do something doesn't mean we ought to.”

Scientists have long searched to find a non-invasive way to gather fetal cells from a pregnant woman's blood. But the rare few that exist have been shed off the placenta, and are on their way to dying, Dr. Wilson explained. Adrift in the woman's blood, they burst.

Then in 1997, scientists Dennis Lo and James Wainscoat, at the University of Oxford, pioneered a technique to pick up the fetal genetic material that lingers in a woman's bloodstream even after the

fetal cells have died off.

Sequenom licensed the Oxford technology in 2005 and says it will likely sell the test for roughly \$700 (U.S.).

Tests to determine the sex of a fetus, screen for Down syndrome and chromosomal abnormalities known as trisomies 13 and 18 will be the first on offer.

But CEO Harry Stylli said the company plans to develop screening tests for a range of other disorders, such as cystic fibrosis, sickle cell anemia and Tay-Sachs disease.

Yet even before the completion of large trials for Down syndrome, the company has been fielding calls from women across North America anxious to try the new screening test, he said.

“This [type of non-invasive test] has been viewed as the Holy Grail of testing,” Mr. Stylli said. “This is just a safer, more precise test. It is going to save women a great deal of anxiety.”

Supriya James, a 41-year-old Toronto woman 19 weeks pregnant with her second child, knows all about the anxiety. Given her age, Ms. James realizes her chances of carrying a child with a genetic abnormality are high – “one in 65 for Down syndrome, one in 43 for any chromosomal abnormality,” she said, quoting statistics from a prenatal screening brochure.

But the current screening process to identify women carrying a child with Down syndrome or another genetic condition can be faulty and stretch into the fourth month of pregnancy.

“It's daunting enough to get pregnant beyond 40, naturally,” Ms. James said. “But it's hard to celebrate it fully because of this incredibly long and arduous road of genetic testing that lies ahead of you.”

The standard noninvasive prenatal tests include a series of ultrasounds and blood tests that together aim to identify women at high risk of having a baby with genetic disorders or birth defects.

Women who receive a positive screening result are then referred on to an amniocentesis some time between 16 to 19 weeks into their pregnancies.

The procedure entails a doctor inserting a long needle directly into the uterus to extract amniotic fluid and the fetal cells and DNA it contains. Amnio results are considered more than 99-per-cent definitive, but the procedure carries a miscarriage risk that ranges from one in 200 to one in 1,000.

The other diagnostic test, chorionic villus sampling, involves the invasive collection of fetal cells from the placenta. While CVS can be performed earlier in the pregnancy, it also carries a slightly higher miscarriage risk than amnio.

It was these risks that prompted Ms. James and her husband Brett James to refuse the amnio when they had their daughter Veda in 2003, and why – after recently receiving very encouraging screening results – they decided to skip the procedure again.

“I've had three previous miscarriages and the idea of an amnio absolutely terrified me,” Ms. James said.

“It's only after getting that [screening] result back that I'm finally in a relaxed state and can celebrate this pregnancy ... but it was 18 weeks in.”

The prospect of a simple blood test that could be performed early in a pregnancy “will be warmly welcomed by all pregnant women,” Ms. James predicted.

“It would spare so much mental anguish.”

Some experts feel the new tests will only be a significant advance if they prove to be as accurate as amniocentesis.

But others believe they will be a major step forward if they can improve the considerable rate of false positives and negatives in the pre-amnio screening process.

Dr. Wilson of the obstetrics society noted that even the best current screening methods result in a number of women being told they are carrying a baby with Down syndrome when they are not.

“You could have 10 to 15 women who screen positive, but only one of them will be a true positive,” said Dr. Wilson, also head of obstetrics and gynecology at the University of Calgary and Foothills Hospital.

Sequenom has so far tried its test in 399 pregnant women and found it has an accuracy rate of 99.1 per cent, a figure “comparable to amniocentesis,” Mr. Stylli said.

Yet in the course of conducting their research, they found that 136 of those 399 women had received positive, high-risk screening results and were recommended to have an amnio or CVS.

But only six of those 136 women actually had a child with Down syndrome, Mr. Stylli said. Meanwhile, two per cent of women who screen positive choose to terminate their pregnancies without an amnio.

“It's scary actually,” he said of the current technology. “Is losing even 2 to 2.5 per cent of babies you look at [through amnio] acceptable?”

The Stanford group, which published its results with a non-invasive prenatal test last fall, found no false positives or negatives in a study of 18 patients.

Mr. Stylli admits Sequenom needs results from much larger trials before it can be considered a diagnostic test and says the company will restrict its claims as it begins selling the procedure.

But he added that trials are under way with more than 800 patients, with plans to publish the results in a peer-reviewed journal later this year.

Jon Barrett, chief of maternal fetal medicine at Toronto's Sunnybrook Hospital, cautioned that the company is still in its research phase, raising the question, “are people being sold a technology that's not quite ready yet?”

The company could use its early customers as part of its patient trials, but then, he said, they should not be charging for the test.

Mr. Stylli said only doctors will be able to order the test, sending blood samples from their pregnant patients to a Michigan lab where the procedure will be performed and the results returned by mail.

It is the online, mail-order process that has the Canadian Down Syndrome Society particularly worried about the new tests.

While the CDSS believes families should be entitled to the best prenatal information and make their own decisions, Ms. Flint said the prime concern is that results from these new tests will come without any insights offered as to “the rich and rewarding lives of citizens with Down syndrome.”

Just last year, the obstetrics society recommended that every pregnant woman in Canada be offered prenatal genetic screening regardless of age and, since then, the Down Syndrome Society has been working to ensure doctors and health-care professionals dispense the screening results with balanced information.

“Prospective parents tend to be told negative things ... like ‘this child will ruin your life ... it will be hard on your life, hard on your kids, ruin your marriage....’

“You don't hear about those who enrich the lives of others, who go to school, get married, get jobs,” said Ms. Flint, who, along with other advocacy groups, has been in talks with Sequenom.

U.S. statistics suggest that 80 to 95 per cent of women who receive an early prenatal diagnosis of Down syndrome choose to end their pregnancies, she noted. In Canada, the number cannot be tracked due to privacy regulations.

Renate Lindeman of Nova Scotia, an activist mother of two daughters with Down syndrome, says Down syndrome birth rates are already falling.

“This is just the result of doing ultrasound screening,” Ms. Lindeman said. “I am not opposed to prenatal screening and I am pro-choice ... but as time goes by it will get easier and cheaper to screen for Down syndrome and other things ... and there is a view that Down syndrome in particular will be a thing of the past.”

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