

New prenatal blood tests can check fetal DNA, raising ethical questions

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Soon a simple blood test will be able to tell newly pregnant women if they are carrying a child with Down syndrome - raising the prospect, and perhaps peril, of a world with fewer imperfections.

Based on fast-moving DNA science, one test is likely to be available by late fall; a second is due out early next spring. While screening tools already exist, the new blood tests will provide an easier and earlier way to help us decide the delicate question of what kind of babies we want.

"The implications are enormous," said Stanford University law professor Hank Greely, who studies the legal and ethical implications of emerging technologies.

"It could lead to a greater reduction in the number of children born with genetic disease," if proven accurate, affordable and then improved to detect hundreds of other congenital disorders, Greely predicts.

The day may come, say experts, when parents are spared the trauma of a baby who perishes at birth - or faces a life filled with pain or profound disability.

But are we ready, they ask, for a future fetal DNA test that forecasts a life filled with cancer? Depression? Alzheimer's disease?

The leap forward is driven by profound and rapid advances in technology that followed the completion of the human genome

sequencing project - making it possible to identify and count millions of short sequences, or fragments, of DNA from blood samples.

The biotech companies Verinata Health of San Carlos, Calif., and Fluidigm Corp. of South San Francisco have licensed a technique designed by Stanford biophysicist Stephen Quake. It precisely counts the millions of [DNA molecules](#) from both the mother and baby and can detect whether some [genetic material](#) is excessive.

A similar approach, but using a different way of counting, is enlisted by the San Diego company Sequenom. It just wrapped up a global study of 2,200 [pregnant women](#); once published, later this fall, the company plans to offer its DNA test.

The market is huge: 4.5 million U.S. births a year, of which an estimated 750,000 are "high risk," due to age or family history. The first application will be Down syndrome screening, because it is relatively simple. A positive result will be confirmed through additional testing.

"There is an eagerness on the part of women not to undergo amniocentesis and chorionic villus sampling," which require needles and can induce miscarriages, said Sharon Terry, director of the Washington D.C.-based nonprofit Genetic Alliance.

"And there are many women who want to make their choices early in a pregnancy," she added, "if there is a potential problem. Or they simply want to be better prepared."

To be sure, there is a lot that is not yet understood about how a person's DNA sequence is linked to disease or disability.

But the impact on fetuses with clear-cut diagnoses, like Down syndrome, would be immediate - and profound.

"It would be a very lonely world," said Martha Hogan of Danville, Calif., whose 33-year-old son Blair has Down syndrome. "Once you have lived with and loved these adorable people, you see how much they add to life.

"Some women, carrying a piece of tissue that they haven't bonded with yet, might let the baby go. It looks like just a blip on a screen. In that case, an earlier test is good, because you're not killing a live baby with a heartbeat and limbs," said Hogan, who counsels women at the Down Syndrome Connection, based in Danville.

"But it would have been tragic to have missed out on my son," she said.

Few women seek screening with current tests - nonetheless, since their introduction, the number of babies with Down syndrome has declined, falling 11 percent between 1989 and 2006, according to Dr. Brian Skotko, a physician with the Down Syndrome Program at Boston's Children's Hospital. The latest figures show 1 out of every 691 babies is born with Down syndrome, an extra copy of the 21st chromosome, which causes cognitive delays and some disability.

"When a new blood test is available, nearly all pregnant women will use it," he predicted. This is likely to result in more elective abortions, he predicted.

He fears that women won't seek counseling, so will never meet people like his sister Kristin, 32, who has Down syndrome and "has taught me extraordinary life lessons. How to keep smiling. How to keep going. How to find the hidden treasures that exist in every day. She has a very rich life - and I am a better person because of her.

"Are we seeing our last generation of Down syndrome?" he asked.

And the broadened application of such a trouble-free test could deliver

new worries, experts caution. Perhaps insurance companies will pressure parents not to bring handicapped children into the world. Public support for social and medical services could weaken. Maybe there'd be less pressure to find cures.

"Now is the time to ask," said Terry, "Are there certain conditions that should be selected out of the mainstream of humanity?"

"What kind of diversity, and disability, are we comfortable with?"

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