

Health & Science

A safe prenatal genetic test is gaining popularity with young moms-to-be and their doctors

By Sarah Elizabeth Richards

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During Carrie Wells’s first OB/GYN visit after learning she was pregnant this summer, the 30-year-old was surprised when her doctor suggested a blood test that could detect her baby’s risk of Down syndrome and other chromosomal abnormalities as early as 10 weeks. At her age, she hadn’t been worried about conceiving a child with a genetic disorder.

Wells’s obstetrician/gynecologist explained that the test, called a “cell-free DNA” screen, analyzes the fetal genetic information that travels from the mother’s placenta and poses no risk to the baby. The doctor had been routinely prescribing it for her patients who were older and higher risk, now she was prescribing it to younger women, as well.

The doctor’s reasoning: women under 35 had more babies overall and account for 80 percent of country’s 6,000 annual Down births, according to the Centers for Disease Control and Prevention. She explained that the test was more accurate than the current standard of care for younger women, which can include other blood tests and an ultrasound to measure the fluid at the base of the fetus’s neck that can signal the presence of Down. Plus, the parents could learn the gender during the first trimester if they wanted, rather than wait to see the baby’s anatomy on the 20-week ultrasound.

“It was an easy blood draw, and we didn’t see a downside,” says Wells, a public relations specialist from Baltimore. “My husband and I hadn’t discussed what we would do if we got a bad result, but I thought if we did, wouldn’t we want to know as far in advance as possible?”

Wells is part of a new wave of younger women gaining access to what is marketed as the “noninvasive prenatal test” (NIPT). Becoming available in 2011, the NIPT was rapidly embraced by OB/GYNs and covered by insurers for women over 35 because it was less risky than two other tests, [amniocentesis](#) and [chorionic villus sampling](#) (CVS), and more sensitive in picking up genetic abnormalities. It also could be performed earlier in pregnancy than amniocentesis.

In a [2015 study in the New England Journal of Medicine](#) of nearly 16,000 pregnant women undergoing first trimester screening, cell-free DNA testing detected the presence of trisomy 21, the marker for Down syndrome, in 38 of 38 women, compared with just 30 of 38 women who were given current standard-of-care tests.

Diana Bianchi, director of the Eunice Kennedy Shriver National Institute of Child Health and Human Development, argued that cell-free DNA testing has led to a “global transformation of prenatal care” in [an](#)

[article](#) last summer in the New England Journal of Medicine. Not only did it spare women unnecessary angst from fewer falsely abnormal results than current methods, but the increased use of the NIPT led to fewer invasive amniocentesis or CVS procedures that carry a small risk of miscarriage, she wrote.

Today, as many insurers expand coverage of the test — and more women request it — more OB/GYNs are making the NIPT a standard offering for all patients. In 2017, nearly 60 percent of OB/GYNs prescribed it to younger women — up from 35 percent the year before, according to the asset management firm Piper Jaffray.

Yet the NIPT's entry into the mainstream has raised some questions as busy health-care providers scramble to figure out how to offer it en masse to a public with varying understanding of genetic testing and little access to genetic counselors. "Sometimes, OB/GYNs don't have time to explain the nuances of the tests before they order them," says OB/GYN Neeta Vora, who works as the director of reproductive genetics at the University of North Carolina School of Medicine.

That communication gap is further exacerbated by what Houston prenatal genetic counselor Blair Stevens calls the "elephant in the OB/GYN's office": some patients agree to take the NIPT simply to learn the sex of their baby. They may not understand that the NIPT is not 100 percent accurate and should be confirmed by a diagnostic amniocentesis or CVS. And they also might not be prepared for — or even expecting — the possible genetic problems the test can reveal.

When Casey Delperdang was pregnant with her son, Simon, two years ago, she says the main appeal was the chance to learn she was carrying a boy. "I was 25 at the time and wasn't even thinking about genetic abnormalities because of my age. I sort of took the test for granted," says Delperdang, who works in health-care marketing in Houston.

When she received her results, she and her husband threw a "gender reveal" party that involved an elaborate scavenger hunt leading guests to a tray of custom-made cupcakes. The blue frosting hidden inside indicated they were having a boy. "I have a lot of similarly aged friends who've been pregnant recently, and it's not even a question that they get the test for the same reason," she said. Thankfully, her test results were genetically normal.

Although disclosing fetal sex information is forbidden in some countries — such as China and India — out of concern that girls will be selectively aborted, [research shows](#) that is not an issue in the United States. Rather, Blair says, she is worried about parents-to-be getting emotionally blindsided by upsetting genetic results.

"As a culture, we've gotten so swept in gender reveal parties that we've forgotten the real purpose of the test," she says. "I see women in my office with positive results who don't know what they've have their blood drawn for."

In addition to testing for Down syndrome, a cognitive impairment disorder with a risk of heart defects, vision and hearing problems, the NIPT also detects the rarer Edwards and Patau syndromes that occur in 1 in 5,000 and 1 in 16,000 births, respectively, and usually result in death shortly after birth.

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Despite the test's increase in popularity, public health experts have not uniformly embraced the technology as the preferred first-line test for all women — and some major insurers are not yet covering it for “average-risk” women, generally meaning under age 35.

For instance, in a [2016 bulletin](#) advocating prenatal screening for women of all ages, the American College of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine stopped short of explicitly endorsing the NIPT.

“We say that all women should be offered testing, but we don't recommend one kind,” says Nancy Rose, a maternal fetal medicine physician at the University of Utah Health Sciences Center and a former genetics chair of ACOG. “All these tests are good, and they all offer advantages and disadvantages.” For example, some of the older blood tests also assess neural tube and ventral wall defects.

Two years ago, six leading genetic NIPT manufacturers and providers formed an alliance to expand coverage for the test and persuade two of the largest private insurers, UnitedHealthcare and Aetna, which cover more than 50 million people in the U.S. combined, to offer NIPT for younger women.

Both Aetna and UnitedHealthcare, which only cover NIPT for high-risk pregnancies, said they cover traditional blood and ultrasound tests for younger women in line with the professional groups' recommendations.

ACOG is reviewing its position, says Chris Zahn, ACOG's vice president for practice activities. (A [recent study](#) suggested that NIPT may also detect some ovarian and cervical cancers in the mother.)

“There's new evidence every day about how this technology is expanding to detect abnormalities earlier,” he says. “But from a clinical perspective, we still need to figure out how to counsel women to think about the results.”

At UNC, Vora teaches OB/GYN residents how to talk to patients about the NIPT and genetic testing in general.

“I encourage residents to ask the basics, such as, ‘Have you ever heard of Down syndrome?’ ” she says. “Based on what patients say, you're obligated to spend more time with them and explain the general concepts of chromosomes.”

Vora's department is developing an educational program that patients can scroll through on an electronic tablet during an office visit. Similarly, Illumina, the San Diego maker of DNA sequencers used in 90 percent of NIPT samples processed in the United States, plans to launch a multilingual app soon that patients could consult before or after an appointment.

Part of adequately counseling women, Stevens says, is realizing that not all women want to know everything modern science can tell them about their pregnancies. “It causes them anxiety,” she says. “They don't want to know because they don't know what they would do with a positive result.”

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Although a [2012 retrospective analysis of 24 studies](#) found that 67 percent of U.S. women who learned their fetuses probably had Down syndrome had terminated their pregnancies between 1995 to 2011, Stevens says she also tells patients that they can use such information to prepare for a special-needs child by connecting with other parents or finding the right doctors and birth centers.

Yet, unlike previous prenatal testing that might occur in the second trimester, some observers say the NIPT is poised to change the American pregnancy experience as we know it — by making early testing a standard part of the OB/GYN checklist, whether a woman really wants it or not.

“Just the fact that it’s routinely offered forces you into a decision, or you’re an irresponsible mother,” says Vardit Ravitsky, a bioethicist at the University of Montreal who studies the social impact of prenatal testing. “With an amniocentesis, you could say you didn’t want to risk miscarriage, but with NIPT, what’s your excuse?”

The focus on testing also could dampen the joy of those early months of pregnancy. “It’s no longer ‘Congratulations! You’re pregnant!’ It’s ‘Here’s a test to see what could be wrong,’” says Barbara Katz Rothman, a sociologist at City University of New York and the author of “Tentative Pregnancy: Prenatal Diagnosis and the Future of Motherhood.” “It induces a kind of anxiety that’s not healthy.”

Yet for Wells, receiving a negative NIPT result before the end of her pregnancy’s first trimester gave her peace of mind — and made her pregnancy more real.

“It was a huge relief,” she says. “I thought, ‘Now we can tell people and celebrate.’”

She and her husband declined to hold a gender reveal party, but they did engage in another modern ritual of pregnancy: a funny Facebook post.

The couple took a photo of their two male cats posed next to an ultrasound image and a copy of book “The Expectant Father.”

“I just told my sons that we have a third son on the way,” read the caption. “He’ll be here in early February.”

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