# Consensus and Controversy: Clinicians' Views of Non-Invasive Prenatal Testing

For anyone who has children — or wants to have children in the future — genetic testing is an important part of the process. In the last several decades, aided by technologies including next-generation sequencing (NGS), fetal genetic testing has expanded significantly. NIPTs, also called noninvasive prenatal screening (NIPS) tests, are screening tests done on blood from the pregnant parent that use NGS to sequence short fragments of cell-free fetal DNA (cfDNA) to determine whether or not there are chromosomal abnormalities in the fetus.

Since their introduction, NIPTs have been moderately popular and controversial. In the United States, about 25 to 50 percent of women have NIPT performed during pregnancy. And while the American College of Obstetrics and Gynecology recommends that prenatal screening, including NIPT, be offered to all pregnant women regardless of their gestational history or risks, the organization notes the risk of false-positive and false-negative results. The FDA has cautioned pregnant people against making decisions about keeping or terminating pregnancies based on NIPT results, and instead advocates for confirmatory testing.

In January 2024, the Deerfield Institute, a division of Deerfield Management Company, an affiliate of Cure, conducted a study of 106 physicians and 125 genetic counselors who work with pregnant people to determine the use and benefits of NIPTs. The physicians surveyed included specialists in maternal-fetal medicine, obstetrics and gynecology, internal medicine and others. The results are revealed here publicly for the first time.

## NIPT marketplace

Since the 1970s when the first genetic screening test was introduced for neural tube defects, companies have created dozens of tests that claim to test for both common and rare genetic abnormalities. NIPTs can test for common chromosomal disorders such as Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13). These diagnostics can also screen for disorders that affect the X and Y chromosomes, such as Turner syndrome and Klinefelter syndrome. Some companies offer expanded NIPTs, which look for more rare disorders, including those caused by chromosomal microdeletions such as Angelman syndrome and Prader-Willi syndrome.

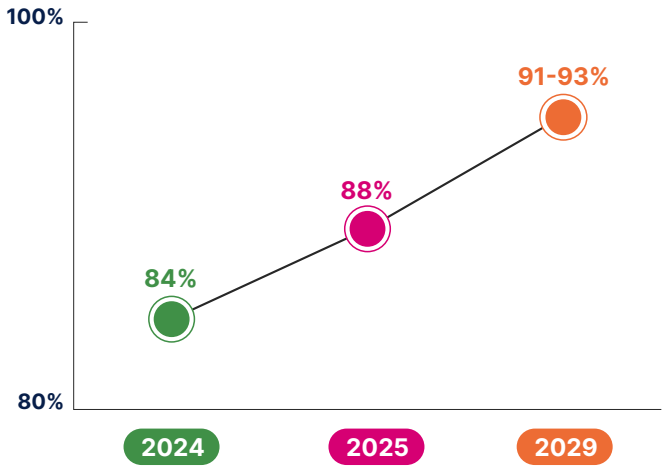
Still, the NIPT market has been pursued heavily by companies including Invitae, Natera, Quest, LabCorp, Billion to One, Myriad and more. These companies are competing for a slice of the genomic testing market that is predicted to reach $140 billion by 2026.

## NIPT vs. Carrier Tests — which is more popular?

In 2020, the American College of Obstetrics and Gynecology (ACOG) recommended that prenatal screening, including NIPTs, be offered to all pregnant women regardless of their gestational history or risks. Both ACOG and the FDA note the risk of false results and advocate for confirmatory testing.

The Deerfield survey found that 84 percent of genetic counselors and physicians use NIPTs in counseling their pregnant patients. The number is expected to rise to 88 percent in the next year, and between 91 and 93 percent in the next five years.

**NIPT Use Expected to Increase**



The survey respondents said that increased insurance coverage for NIPTs will be a driving force in uptake among patients. “I think insurance coverage will continue to expand,” one respondent said. “While some people decline testing for other reasons, the majority that I have decline are concerned about the cost.”

Currently, more patients get NIPT screenings compared to carrier screenings — screenings that test one or both parents for genetic diseases that could be inherited by offspring. Carrier screenings can test for genetic diseases including cystic fibrosis, fragile X syndrome, Tay-Sachs disease and sickle cell disease. Like NIPTs, carrier screenings can be done via a simple, noninvasive blood test. The genetic counselors surveyed said that 75 percent of their patients got carrier screenings, while the physicians surveyed said that 81 percent of their patients got carrier screenings.

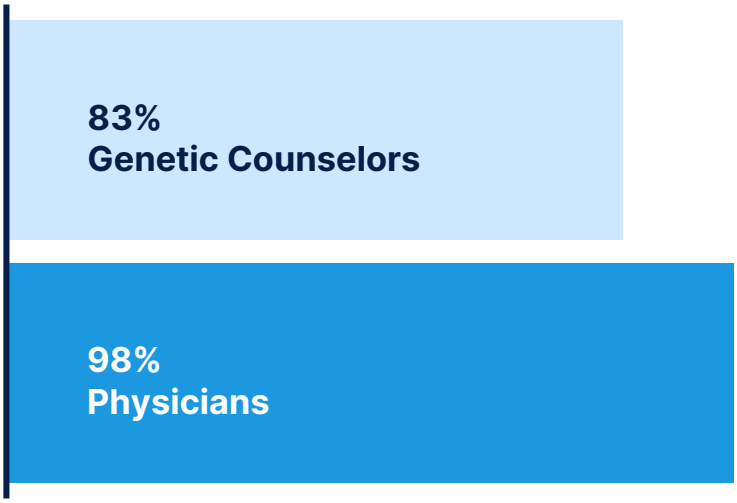
For those that refuse all screenings, providers say cost is often a factor. “Patients are concerned about large bills,” one respondent said, “whether for themselves, or their partner as follow up. Or further testing for the pregnancy.”

Other patients decline all genetic testing as a matter of personal or religious preference.

## Who is more likely to order a NIPT?

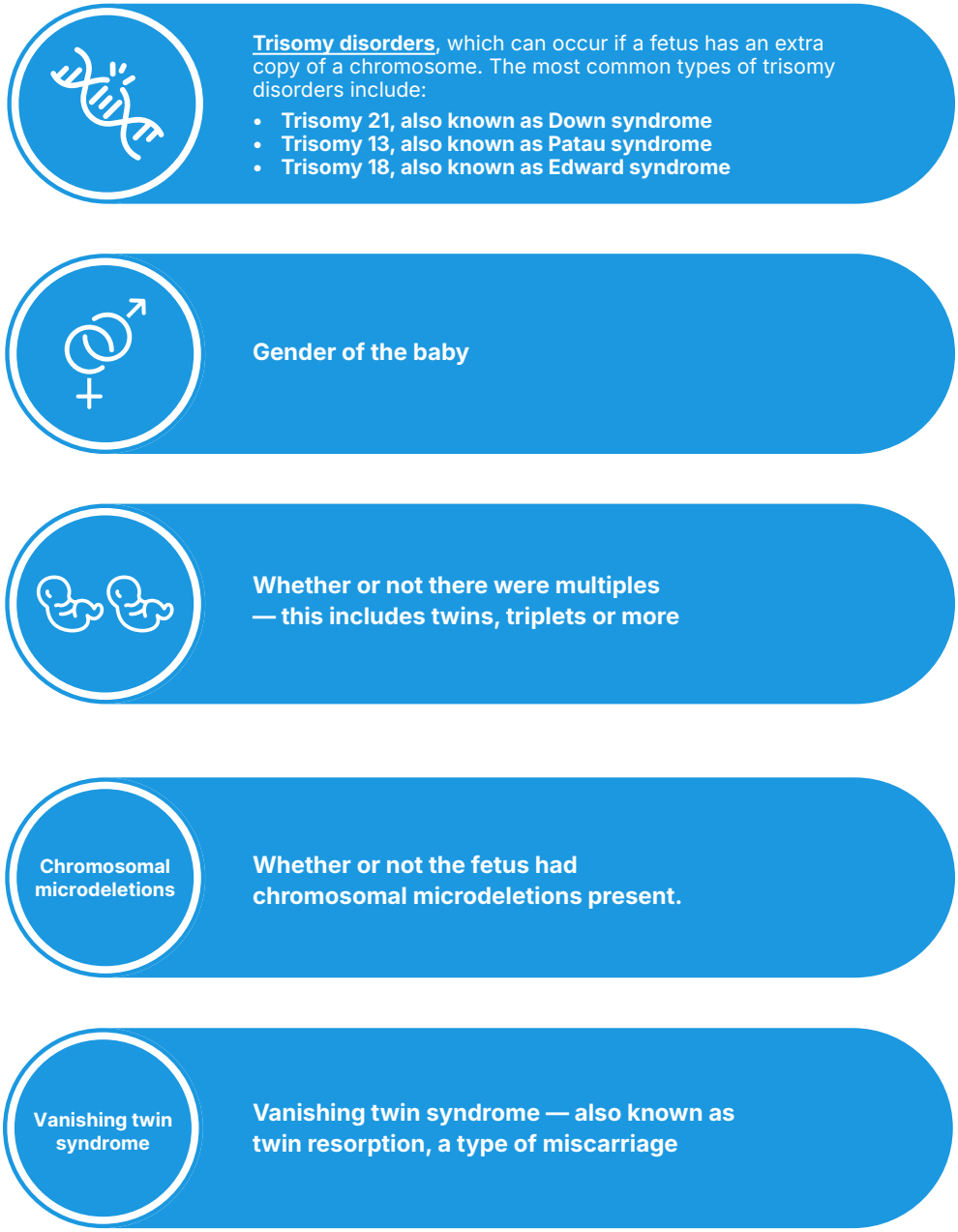
When it comes to ordering a NIPT, physicians are slightly more likely to order one compared with genetic counselors. Of genetic counselors, 83 percent reported that they ordered NIPTs, compared to 98 percent of physicians. Meanwhile, 100 percent of genetic counselors ordered carrier tests.

**Physicians** are slightly more likely to order NIPTs compared to genetic counselors



## What is the most important information from an NIPT?

A lot of information can be gleaned from NIPTs. But the physicians and genetic counselors surveyed agreed some data were "more important" to them, and their patients, than others. These include five data points



## Microdeletion testing: A controversy

Whether or not to use NIPTs to screen for chromosomal microdeletions is a controversy within the obstetrics community. Microdeletions occur when a small piece of chromosome is missing. While some microdeletion syndromes — like Angelman syndrome and Prader-Willi syndrome — have been associated with abnormal developmental outcomes, other microdeletions are harmless.

ACOG does not recommend the use of NIPTs to detect microdeletions. The reasoning is that while NIPTs generally perform well at ruling out chromosomal abnormalities, there is a high rate of false positive results, especially when it comes to microdeletions. To truly detect microdeletions, more invasive testing, such as an amniocentesis, must be performed.

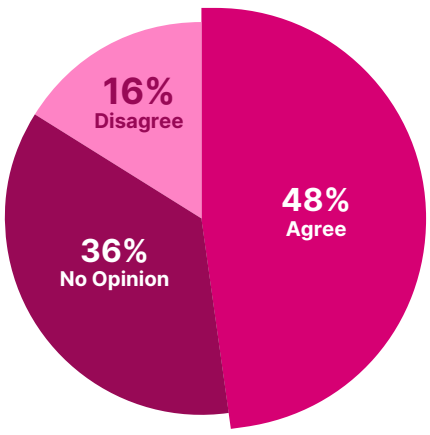
When physicians and genetic counselors were asked about the clinical utility of microdeletion testing with NIPTs, 48 percent of physicians and 56 percent of genetic counselors agreed that there was enough clinical utility that insurance coverage was justified.

"A positive result can direct further diagnostic testing," said one survey respondent who was in favor of testing for microdeletions. "If a baby is confirmed to be affected, this can guide obstetrical management, options for pregnancy management, and neonatal care which may reduce morbidity and mortality."

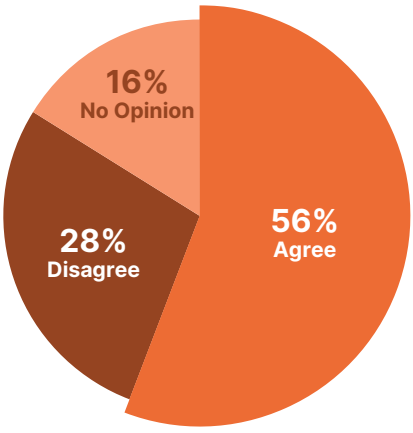
Another said that microdeletion testing with NIPTs can provide "early information to prevent delayed diagnosis and ensure children and families get the support and resources they need."

However, 16 percent of physicians and 28 percent of genetic counselors disagreed that NIPTs for microdeletion testing are useful enough to justify insurance coverage, while 16 and 28 percent, respectfully, had no opinion. Those that were against microdeletion testing said that patients would ultimately have to undergo invasive testing anyway. "I feel that microdeletion NIPT testing gives patients the wrong impression that it is an alternative to amniocentesis," one respondent said.

**48 percent of physicians and 56 percent of genetic counselors agreed that NIPTs for microdeletions have enough clinical utility to justify insurance coverage.**




Physicians




Genetic Counselors

## The future of NIPTs

Until 2020, NIPTs were only recommended for pregnant people with certain risk factors: those at advanced maternal age, or who had a previous child with chromosomal abnormalities, fetal abnormalities seen on an ultrasound, or a family history of genetic disease.

That year, however, the American College of Obstetricians and Gynecologists recommended that NIPTs be made available to all pregnant people, regardless of risk. In the years since, improved accuracy of the NIPT, expanded screening offerings and a commercialization push by companies mean that more people than ever are getting NIPTs. While the tests continue to spark conversation surrounding ethical issues and debates about microdeletion detection, it is clear that NIPTs are a game changer in the genetic testing space that will only continue to grow.

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