FDA NEWS RELEASE

FDA Warns of Risks Associated with Non-Invasive Prenatal Screening Tests

Inappropriate interpretation of results may lead to potentially improper medical decisions

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Español (/news-events/press-announcements/la-fda-advierte-sobre-los-riesgos-asociados-las-pruebas-de-deteccion-prenatales-no-invasivas)

Today, the U.S. Food and Drug Administration is warning the public of the risk of false results, inappropriate use and inappropriate interpretation of results with non-invasive prenatal screening (NIPS) tests, also called cell-free DNA tests or non-invasive prenatal tests (NIPT). These tests look for signs of genetic abnormalities in a fetus by testing a sample of blood from the pregnant person. Given the increased use of these tests and recent media reports, the FDA is providing this information to educate patients and health care providers and to help reduce the inappropriate use of NIPS tests.

"While genetic non-invasive prenatal screening tests are widely used today, these tests have not been reviewed by the FDA and may be making claims about their performance and use that are not based on sound science," said Jeff Shuren, M.D., J.D., director of the FDA's Center for Devices and Radiological Health. "Without proper understanding of how these tests should be used, people may make inappropriate health care decisions regarding their pregnancy. We strongly urge patients to discuss the benefits and risks of these tests with a genetic counselor or other health care provider prior to making decisions based on the results of these tests."

NIPS tests can provide information about the possibility a child will be born with a serious health condition. However, NIPS tests are screening tests – not diagnostic tests. They only provide information about the risk that a fetus may have a genetic abnormality, and additional testing may be needed to confirm whether or not a fetus is affected.

Genetic abnormalities may be caused by a missing chromosome or an extra copy of a chromosome, known as an aneuploidy, a small piece missing from a chromosome called a microdeletion, or an extra piece of chromosome called a duplication. These genetic abnormalities can cause serious health conditions. Conditions caused by a missing chromosome or an extra copy of a chromosome are more common and may be easier to detect, such as Down

syndrome, which can cause physical and intellectual challenges. A missing or extra piece of a chromosome may result in rarer conditions, such as DiGeorge syndrome, which can cause heart defects, feeding difficulties, immune system problems and learning difficulties.

All NIPS tests on the market today are offered as laboratory developed tests (/medical-devices/in-vitro-diagnostics/laboratory-developed-tests) (LDTs). Most LDTs, including NIPS tests, are offered without review by the FDA. While LDTs are medical devices under the Federal Food, Drug, and Cosmetic Act, the FDA has had a general policy of enforcement discretion for most LDTs since the Medical Device Amendments were enacted in 1976. That means that FDA does not generally enforce applicable regulatory requirements for most LDTs. The FDA is continuing to work with Congress on legislation to establish a modern regulatory framework for all tests, including LDTs.

Many laboratories offering these tests advertise their tests as “reliable” and “highly accurate,” offering “peace of mind” for patients. The FDA is concerned that these claims may not be supported with sound scientific evidence. While these laboratories claim their tests are highly accurate, there are limitations due to the rarity of some of the conditions included in the screening. For example, when screening for a very rare condition, a positive screening result may be more likely to be a false positive than a true positive, and the fetus may not actually be affected. In other cases, a positive screening result may accurately detect a chromosomal abnormality, but that abnormality is present in the placenta and not in the fetus, which may be healthy.

Patients and health care providers should be aware of the risks and limitations of using these genetic prenatal screening tests and that they should not be used alone to diagnose chromosomal (genetic) abnormalities. However, the FDA is aware of reports that patients and health care providers have made critical health care decisions based on results from these screening tests without additional confirmatory testing. Pregnant people have ended pregnancies based on the results of genetic prenatal screening alone, without understanding the limitations of the screening tests and that the fetus may not have the genetic abnormality identified by the screening test.

The FDA recommends that patients and health care providers discuss the benefits and risks of all prenatal genetic testing, including NIPS tests, with a genetic counselor or other health care provider before considering such testing or making any decisions about their pregnancy. Please see the safety communication linked below for a full list of recommendations for patients and health care providers.

The FDA will continue to closely monitor safety issues around the use of NIPS tests and is committed to protecting public health.

Related Information

- Genetic Non-Invasive Prenatal Screening Tests May Have False Results: FDA Safety Communication (/medical-devices/safety-communications/genetic-non-invasive-prenatal-screening-tests-may-have-false-results-fda-safety-communication)

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