

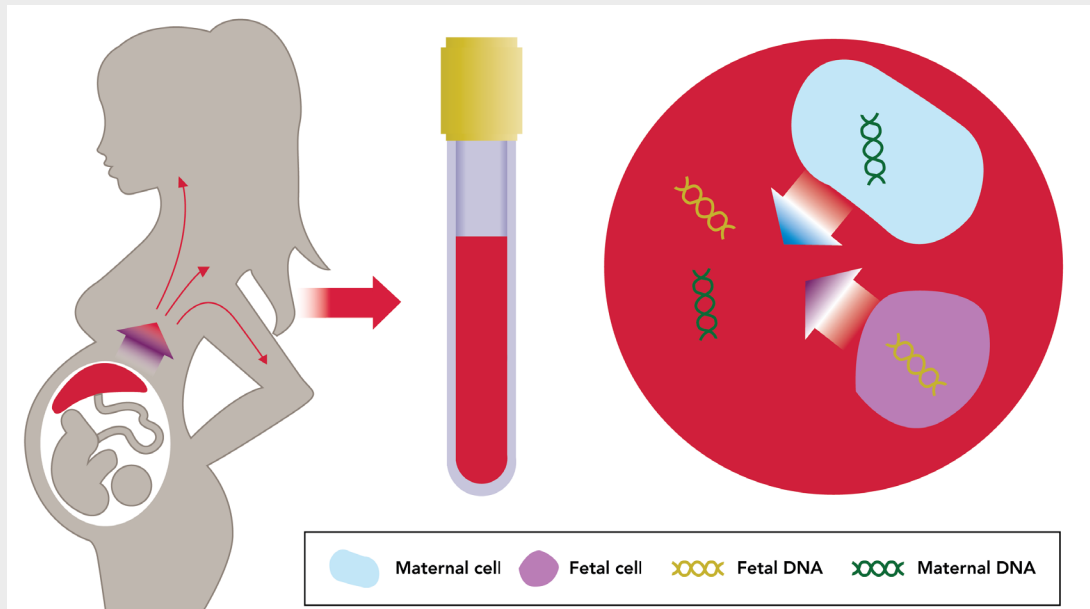


NONINVASIVE PRENATAL GENETIC SCREENING

In the last decade, advances in DNA sequencing have revolutionized prenatal screening for chromosomal disorders in fetuses. Now routinely carried out as part of prenatal care, noninvasive prenatal screening (NIPS) offers doctors a low-risk way to screen for specific genomic conditions in a fetus using just a small sample of a pregnant woman's blood. Early detection of a fetal genetic abnormality gives a woman time to understand the disease and discuss options for care with a healthcare provider.

Principle of Noninvasive Prenatal Testing

DNA from the woman and fetus is extracted from a maternal blood sample and screened to detect chromosomal abnormalities in the fetus.



Prenatal Genetic Testing

Birth defects affect about three percent of all babies born in the United States annually. Some genetic abnormalities can be detected before birth through procedures such as amniocentesis and chorionic villus sampling (CVS). CVS involves testing a sample of tissue from the placenta. However, these procedures are invasive and carry the risk of miscarriage.² The development of noninvasive testing grew from a desire to minimize risk to the health of the fetus from testing by limiting direct contact with the fetus or placenta.

NIPS: A Game Changer

Funding from the National Institutes of Health (NIH) have supported the development of sensitive, efficient methods for DNA sequencing and clinical trials aimed at isolating and detecting fetal cells from maternal blood. This funding has enabled a totally new approach to prenatal genetic testing.

NIPS works by analyzing fragments of fetal DNA that naturally circulate in the woman's blood during pregnancy. Introduced into clinical practice in 2011, NIPS involves a simple blood draw that can be performed early in the pregnancy. NIPS is primarily used to look for chromosome disorders

that are caused by the presence of an extra or missing copy of a chromosome. These include Down syndrome, the most common chromosomal abnormality, as well as trisomy 13, trisomy 18, and an irregular number of sex chromosomes.

A positive result from NIPS is typically followed by amniocentesis or chorionic villus sampling to confirm the diagnosis. A negative result precludes the need for the more invasive diagnostic testing; in fact, there has been a dramatic decline in such testing since NIPS was introduced.

Knowledge is Power

Many parents want to know beforehand if their baby could have a chromosomal abnormality. Prenatal genetic screening gives families time to learn about the disorder and discuss options with doctors. For instance about half of children with Down syndrome have heart defects, NIPS can help clinicians, who may need to plan for surgery soon after birth. Current research is also focused on developing new therapies to treat genetic disorders in fetuses before they are born.

Additionally, since NIPS analyzes both fetal and maternal DNA, the test may detect undiagnosed medical conditions in the pregnant woman. NIPS has identified maternal medical conditions, including vitamin deficiencies, uterine fibroids, and even cancer.

Accessible Technology

Due to its many advantages, the adoption of NIPS in routine clinical practice has been rapid and global. Several million pregnant women receive this test each year in the United States and Europe and tests are increasingly becoming available in the Middle East, South America, Asia, and Africa. Since NIPS requires only a blood draw, it may offer benefits in developing countries, such as reducing the need for trained medical personnel to perform invasive diagnostic procedures and making genetic testing more accessible in remote and resource-poor areas.

How Congress Can Support NIPS

NIH funding supports research and development efforts focused on overcoming current NIPS limitations and increasing the testing accuracy. As technology improves and the cost of genetic testing decreases, researchers expect that NIPS will become available for a larger range of genetic conditions, including those for which no prior screening protocols exist. With sustained NIH funding, NIPS may one day be a final diagnostic test, replacing invasive methods of screening for genetic abnormalities. Further research is needed to ensure NIPS works equally well in all populations of women. Congress can support advances in prenatal health, improved technology, and personalized genomic medicine by continuing to fund basic biomedical research..

Additional Resources

Noninvasive Prenatal Genetic Testing
genome.gov/dna-day/15-ways/noninvasive-prenatal-genetic-testing

What is NIPT?
genomicseducation.hee.nhs.uk/blog/what-is-nipt/

Prenatal Genetic Screening Tests:
acog.org/patient-resources/faqs/pregnancy/prenatal-genetic-screening-tests

References:
ashg.org/advocacy/fact-sheets/