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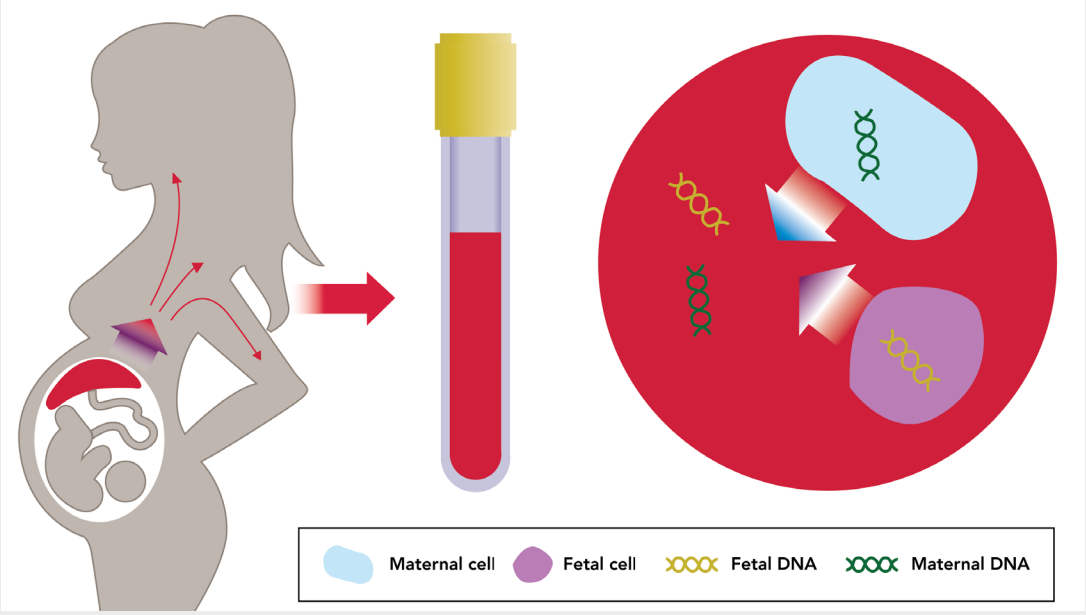
Success Stories
in Human Genetics and Genomics Research

PRENATAL CELL-FREE DNA 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, prenatal cell-free DNA (cfDNA) screening offers doctors a low-risk way to screen for specific genomic conditions in a fetus using just a small sample of a pregnant mother's blood. Early detection of fetal genetic abnormalities gives families and clinicians time to prepare for the care the child may require.

Principle of Prenatal Cell-Free DNA Screening



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 carry the risk of miscarriage.[2] The development of prenatal cell-free DNA screening grew from a desire to minimize risk to the health of the fetus from testing by limiting direct contact with the fetus or placenta.

cfDNA Screening: A Game Changer

Funding from the National Institutes of Health (NIH) has 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.

cfDNA screening works by analyzing fragments of fetal DNA that naturally circulate in the mother's blood during pregnancy. Introduced into clinical practice in 2011, cfDNA screening involves a simple blood draw from the pregnant woman that can be performed early in the pregnancy. It 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 cfDNA screening is typically followed by amniocentesis or chorionic villus sampling to confirm the diagnosis. A negative result precludes the need for the additonal diagnostic testing; in fact, there has been a dramatic decline in such testing since cfDNA screening was introduced.

Knowledge is Power

Many parents want to know beforehand if their baby will be born with a chromosomal abnormality. Prenatal genetic screening gives families time to learn about the disorder and prepare for the care the child may require. Since about half of children with Down syndrome have heart defects, cfDNA screening can also 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 cfDNA screening analyzes both fetal and maternal DNA, the test may detect undiagnosed medical conditions in the mother. Prenatal cell-free DNA screening has identified maternal medical conditions, including vitamin deficiencies, uterine fibroids, and even cancer.

Accessible Technology

Due to its many advantages, the adoption of cfDNA screening 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 cfDNA screening requires only a blood draw, it may offer benefits in developing countries, such as reducing the need for trained medical personnel to perform higher-risk diagnostic procedures and making genetic testing more accessible in remote and resource-poor areas.

How Congress Can Support cfDNA Screening

NIH funding supports research and development efforts focused on overcoming current cfDNA screening limitations and increasing the testing accuracy. As technology improves and the cost of genetic testing decreases, researchers expect that cfDNA screening will become available for a larger range of genetic conditions, including those for which no prior screening protocols exist. With sustained NIH funding, cfDNA screening may one day be a final diagnostic test, replacing higher risk methods of screening for genetic abnormalities. Further research is needed to ensure cfDNA screening 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 fundamental biomedical research.

Additional Resources

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

What is cfDNA?
https://www.genomicseducation.hee.nhs.uk/blog/cell-free-dna-prenatal-testing-and-beyond/

Prenatal Genetic Screening Tests:
https://www.acog.org/womens-health/faqs/prenatal-genetic-screening-tests

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

6120 Executive Blvd, Suite 500, Rockville, Maryland 20852 · policy@ashg.org · (301) 634-7300 · www.ashg.org

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