

DNAdots

Simple explanations
of modern genetic techniques

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Cell-free Fetal DNA Screening (cffDNA)

What it is:

Non-invasive prenatal genetic testing

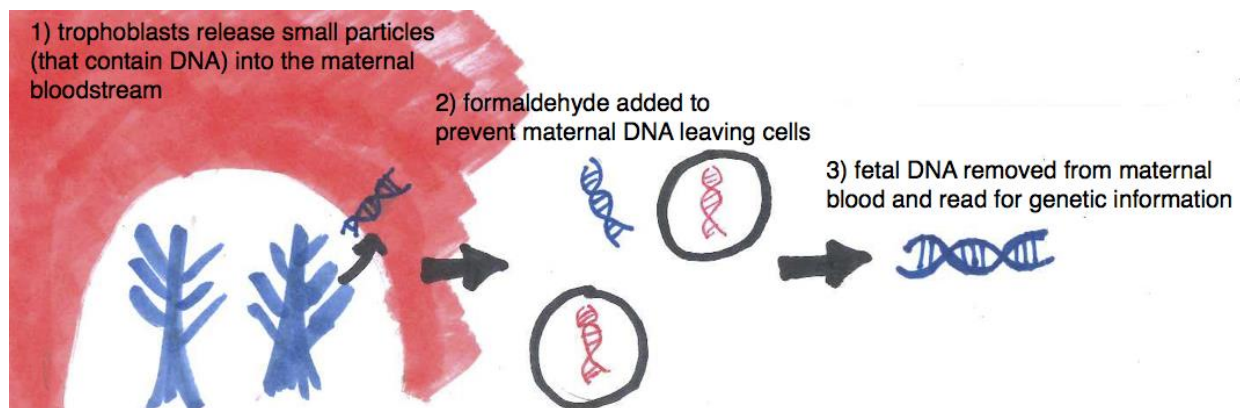
New research has discovered that it is possible to obtain genetic information prenatally directly from the mother's blood, without disrupting the developing fetus or placenta. These tests can detect fetal DNA (cffDNA) directly in the mother's blood as early as 7 weeks into pregnancy. They can allow for sequencing of the fetus genome and for genetic screening without increasing the risk of miscarriage or pregnancy complications.

How it is used:

Screening for genetic risks early in pregnancy

cffDNA genetic testing is mainly used to detect genetic abnormalities that can cause neural tube defects, Down syndrome, and other disorders early during gestation. Currently, cffDNA screening is only being done based on high risk factors, such as maternal age, pre-existing medical conditions, or high blood pressure during pregnancy.

While the technology has been on the market since October 2011, it has yet to be approved by the FDA, so cffDNA testing is currently used as a screening method, meaning it is used to determine risk but remains non-diagnostic. Positive results should be followed up with more invasive diagnostic tests, such as amniocentesis. More invasive tests are not suggested for all pregnancies because they are associated with small risks of miscarriage, therefore the power of cffDNA lies in informing when a pregnant individual should consider a more invasive genetic test.



How it works:

Reading fetal DNA from maternal blood

cffDNA screening starts by detecting small fragments of fetal DNA found in the maternal bloodstream. Fetal DNA can make up to 11-13% of all cell-free DNA in the mother's blood. It enters the maternal bloodstream as trophoblasts that make up the placenta shed small DNA-containing particles. In order to detect fetal DNA, a sample of maternal blood is extracted by a simple venipuncture, then treated with formaldehyde to prevent maternal DNA from being released from cells. Then qPCR (another DNAdots topic!) or shotgun sequencing is used to study fetal DNA. Fetal DNA is often distinguished from maternal DNA by looking for paternally inherited regions. For example, if the fetus is male, Y-chromosome markers can be assumed to represent fetal DNA and used to determine fetal sex, as well as screen for paternally-inherited mutations. Also, in families carrying X-linked mutations that can pose a high risk to male progeny, early gender determination through cffDNA testing can help assess the risk of the fetus developing a genetic disorder.

The future:

How to respond to genetic testing results?

Prenatal testing is done for a variety of reasons including to provide timely medical treatment of a condition before or immediately following birth, time to prepare psychologically, socially, financially, and medically for a child with a health problem or disability. As more accessible prenatal testing technologies emerge, more couples will be faced with difficult choices in the face of genetic results.

It is possible that in the future cell-free fetal DNA screening will become a common test for all pregnant women. cffDNA testing can already be used for whole-genome fetal sequencing, revealing the complete genetic makeup of a fetus. As more genetic information can be assessed prenatally for more people, there must be a societal consensus on what responses to this information are appropriate. If genetic testing is administered to all women, there must be support provided to help individuals handle the implications of results, and sufficient genetic counselors to meet the growing demand. There is an ongoing legislative debate around genetic testing and abortion in many states. Because it is available early in a pregnancy, cffDNA testing raises the question of when a pregnancy could be terminated based on a genetic finding. As cffDNA testing advances, it will become important for society to discuss the appropriate responses to genetic information obtained before birth.

Learn more:

- "Cell-free fetal DNA." *Wikipedia*. http://en.wikipedia.org/wiki/Cell-free_fetal_DNA
- White, Michael. "Next-Generation Prenatal Genetic Tests are Turning Fate into Choice." *Pacific Standard*. <http://psmag.com/next-generation-prenatal-genetic-tests-are-turning-fate-into-choice-1d901c49a445#.6k636xhes>