PATIENT HANDOUT

Prenatal Screening Using Cell-Free DNA

Society for Maternal-Fetal Medicine

By the Society for Maternal-Fetal Medicine (SMFM) with the assistance of Nancy Rose, MD, and the SMFM Education Committee

What is cfDNA (cell-free DNA) and how is it used in pregnancy?

cfDNA refers to small fragments of DNA found in the maternal blood during pregnancy. Most cfDNA fragments come from the placenta and can be used to test for common chromosome disorders in a developing fetus during pregnancy. When a whole extra chromosome is identified by this test, the finding is known as a trisomy, such as trisomy 21 or Down syndrome. Trisomy 21 is the most common form of inherited intellectual disability and also is associated with other health problems. CfDNA screens for trisomy 21, 18, or 13, and for missing or extra genetic material in sex chromosomes X and Y. Sometimes other rare chromosome problems are also included.

How accurate is cfDNA aneuploidy screening?

cfDNA screening is most accurate for the diagnosis of Down syndrome and is somewhat less accurate for the other trisomies and sex chromosome disorders. The accuracy of cfDNA screening depends on several factors, including a woman's baseline risk of carrying a fetus with a trisomy. The chance of having a trisomy depends mostly on a woman's age at the time of screening. For example, the chance of having a baby with Down syndrome at age 40 is about 1% or 1 in 100 at delivery. The risk of having a child with Down syndrome at age 30 is much lower, about 0.1% or 1 in 1000. Therefore, this screening test is most accurate in women over age 35 who have a higher baseline risk of chromosomal abnormalities. In younger women who have a lower risk, there is a much higher chance of a false-positive test, that is, a test result that indicates a high risk of chromosomal problems when the baby is actually normal.

What does it mean to have a positive test result from a cfDNA screen?

CfDNA screening is not a diagnostic test and a positive cfDNA test result DOES NOT MEAN that your baby definitely has a chromosome problem. The test can only tell that there is a higher chance of a chromosome problem. If you have a positive test result, irreversible decisions, such as pregnancy termination, should not be made immediately. You should meet with a maternal-fetal medicine specialist, geneticist, or a genetic counselor to review your results and to discuss other testing options. Patients whose test is positive are usually offered an ultrasound evaluation and a diagnostic test, such as chorionic villous sampling (CVS) or amniocentesis, to determine the actual fetal chromosomes.

What does it mean if the cfDNA screen of my sample doesn't give a test result?

Sometimes cfDNA screening will not be able to provide a test result. This can happen because there isn't enough free fetal DNA available or because there is a problem interpreting the test result in the laboratory. The most common reasons for this are that the test was sent before 10 weeks of pregnancy, the mother's weight is over 250 lb, or because the baby has a chromosome abnormality. Depending on your age and other risk factors, your provider may suggest a repeat cfDNA test, or a diagnostic test (amniocentesis or CVS). You may or may not get a result from a repeat test, and it will take another 7 to 10 days to know.

What are other limitations of cfDNA aneuploidy screening?

At this time, cfDNA screening only evaluates the pregnancy for three common trisomies as well as fetal sex chromosome abnormalities. It does not identify other birth defects.

CfDNA screening is less accurate in women with twins, particularly if one twin stopped developing during the early part of the pregnancy. No information is available on the test accuracy in women who are carrying three or more fetuses.

Your doctor may also recommend MSAFP (maternal serum alpha-fetoprotein) screening, a blood test to evaluate the baby for defects such as an open spine defect (spina bifida).

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