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| SCWHCA_1 | **Southern Connecticut Women’s Health Care Associates****247 Broad Street, Milford CT 06460****P. (203) 783-0543 – F. (203) 874-5728** | Miriam Sivkin, M.D. FACOGMary Murray, M.D. FACOGErika Schaeffer, M.D. |

**What Is the Cell-Free Fetal DNA Test**

The cell-free fetal DNA (cffDNA) test is a relatively new test that may be used to assess the risk of a pregnant woman's developing baby (fetus) having a chromosome disorder, such as Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), or Patau syndrome (trisomy 13). It may be used to identify other rare conditions resulting from an extra chromosome or missing piece of chromosome (microdeletion).

Cell-free fetal DNA is genetic material that is released by the placenta and circulates in a woman's blood during pregnancy. It is present in small quantities starting in the first trimester and increases throughout pregnancy. CffDNA generally reflects the genetic makeup of the developing baby.

The American College of Obstetricians and Gynecologists (ACOG) currently recommends that the cffDNA test be offered to women at an increased risk for trisomy 13, 18, or 21, after pre-test genetic counseling. Factors that increase risk include:

* Advanced maternal age: 35 years or older
* A fetal ultrasound result that indicates an increased risk of a chromosome abnormality
* A previous pregnancy with a trisomy
* A positive first-trimester or second trimester maternal screening test
* A known balanced chromosome translocation (switched chromosome pieces) in the mother or father that is associated with trisomy 13, 18, or 21

Current routine prenatal testing includes the first trimester Down syndrome screen and the second trimester maternal serum screen. While studies have shown that the cffDNA test can be more specific and sensitive than these routine tests in high-risk women, ACOG currently recommends that the cffDNA test be offered separately, not incorporated into routine prenatal testing. The routine second trimester maternal serum screen detects neural tube defects as well as chromosome disorders.

The cffDNA test is used as a screening test, not a diagnostic test. If there are abnormal findings in routine prenatal testing or in cffDNA testing, then more invasive confirmatory testing using procedures such as chorionic villus sampling (CVS) or amniocentesis may be indicated to diagnose a chromosome abnormality.

### When is it ordered?

The cell-free fetal DNA test may be ordered:

* During or after the tenth week of pregnancy
* When a woman is at an increased risk of having a baby with a chromosome disorder
* After pre-test genetic counseling

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### What does the test result mean?

A negative cffDNA test result means that it is very unlikely that the baby has trisomy 13, 18, or 21. If the test is negative for other chromosomal abnormalities that the laboratory tested for, then it is unlikely that the baby is affected by those. However, these trisomies or genetic defects cannot be entirely ruled out, and other chromosome abnormalities could still be present. If a cffDNA screen is positive, then the fetus is at an increased risk of having the identified abnormality. However, the test is not diagnostic. The performance of more invasive procedures, including chorionic villus sampling (CVS) between the tenth and twelfth week of pregnancy or an amniocentesis procedure between 15 and 20 weeks of pregnancy, and subsequent chromosome analysis is required to rule out or diagnose a chromosomal abnormality.

Questions on interpreting the results of a cffDNA are usually best answered by someone experienced in genetic counseling.