Genetic Support Foundation

Prenatal Cell-free DNA Screening

Prenatal cell-free DNA screening (also known as cfDNA screening) is an optional prenatal blood test that screens for certain genetic conditions.[1] It can be performed as early as 10 weeks of pregnancy. Our blood contains fragments of our DNA known as cell-free DNA (cfDNA). When a woman is pregnant, her blood also contains DNA fragments from the placenta that has a genetic makeup that is *usually* identical to that of the developing baby. By analyzing this DNA in mom's blood, it can be determined if there is an increased or decreased chance for certain genetic conditions.

What does cfDNA test for?

Though it began as a screening for Down Syndrome, over time the list of conditions that can be screened for has grown.[2] Not all cfDNA companies screen for the same conditions. Current guidelines from the American College of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine indicate that routine cfDNA screening should *only* be done for the common trisomies (Down syndrome, trisomy 13, and trisomy 18) and, if requested, sex chromosome variations. They do not recommend routine screening for microdeletion syndromes.[3] It is important to keep in mind that *no* prenatal genetic test, not even cfDNA screening, can look for all genetic conditions or birth defects.

[1] cfDNA screening goes by many different names including Noninvasive Prenatal Testing (NIPT) or Noninvasive Prenatal Screening (NIPS). You may have heard it referred to as the new "gender test," or be familiar with specific test brand names (i.e. Harmony TM , MateriniT21®, verifi®, Panorama TM, InformaSeq SM.)

[2] Some conditions include trisomy 18, trisomy 13, and differences with the X and Y chromosomes.

3 http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/Cell-free-DNA-Screening-for-Fetal-Aneuploidy

How certain are cfDNA screening results?

Prenatal cfDNA is a screening test that can determine if the chance of certain genetic conditions in a pregnancy is higher or lower. Since cfDNA screening is not a diagnostic test, it cannot give you “yes” or “no” answers. If a result indicates a higher or increased chance, then a diagnostic test such as amniocentesis can be performed, if desired, to determine whether the baby really has the condition or not. If the results indicate a low or decreased chance, the possibility the baby has the genetic condition is reduced, but not zero.

What are the risks associated with cfDNA screening?

cfDNA screening is performed on a sample of a pregnant woman’s blood. While there is no risk for pregnancy complications such as miscarriage, it is important to think about what these test results may mean for you. Most of the time the test will come back with a low chance for a genetic condition and many women feel reassured based on these results. However, if the test indicates an elevated chance of a genetic condition it may create worry and uncertainty about what to do next.

Making a decision that is right for you...

cfDNA screening has some benefits and drawbacks when compared to other screening tests such as traditional maternal serum screening (otherwise known as quad screen, first trimester screen, triple screen, integrated screen, and sequential screen). There are also benefits and drawbacks when compared to diagnostic testing, such as amniocentesis or CVS. Decisions about genetic testing in your pregnancy should be based on your own beliefs, values, needs, and personality.

You can find more information and videos about prenatal genetic testing options at: www.geneticsupportfoundation.org and www.doh.wa.gov .