Prenatal cfDNA Screening

What is Prenatal cfDNA Screening and How Does it Work?
Prenatal cell-free DNA screening (also known as cfDNA screening) is an optional prenatal blood test that screens for certain genetic conditions. It can be performed as early as 10 weeks of pregnancy.

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 (Harmony™, MaterniT21®, verifi®, Panorama™, InformaSeq SM...)

Our blood contains fragments of our DNA—known as cell-free DNA. 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 in the baby.

What does cfDNA Screening Test For?
Though it began as a screening for Down syndrome, over time the list of conditions that can be screened for has grown (Down syndrome Trisomy 18, Trisomy 13, differences with the X and Y chromosomes). Not all cfDNA companies screen for the same conditions. These conditions are all very different from one another. It is not always possible to know before a child is born how much their health will be affected. It is important to know that cfDNA screening cannot detect all genetic conditions or birth defects. No test can guarantee the birth of a healthy baby.

How Certain are cfDNA Screening Results?
Prenatal cfDNA screening 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 high 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 lower, but not zero.

cfDNA screening has been shown to be more predictive for Down syndrome compared to other screening tests such as maternal serum screening. However, similar to maternal serum screening, it is possible to have a cfDNA result indicating a higher chance of a genetic condition even though the condition is not present in the baby.

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 relief based on these results. However, if the test indicates a higher chance of a genetic condition it may create worry and uncertainty about what to do next.

**How to Decide About cfDNA Screening?**
cfDNA screening has some benefits and drawbacks when compared to other screening tests such as maternal serum screening. Decisions about genetic testing in your pregnancy should be based on your own beliefs, values, needs and personality. See our video, *How to Decide About Prenatal Genetic Testing*, as you consider your prenatal testing options.

**Credits**
This video was created by the Washington State Department of Health and Genetic Support Foundation. You can find more information and additional videos about prenatal genetic testing options at: [www.doh.wa.gov](http://www.doh.wa.gov) and [www.geneticsupportfoundation.org](http://www.geneticsupportfoundation.org).

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