

Our DNA is inside nearly every cell of our body, and is the instruction manual for how everything in our body grows and functions. Our cells are continuously dividing to create new cells. As cells break down, the DNA inside the cell is released into the blood as fragments or pieces of DNA. These DNA fragments in the blood are known as cell-free DNA.

All pregnant women will have DNA fragments in their blood (some that are her DNA, and some that is from the placenta). The placenta develops during pregnancy and provides oxygen and nutrients to the developing baby. The placenta has the same genetic information as that of the developing baby approximately 98-99% of the time.

However, approximately 1-2% of the time, the genetic makeup of the placenta and that of the developing baby are not the same (called <u>confined placental mosaicism</u>). This can lead to false positive (the results are high-risk but the fetus does not actually have the condition) and false negative results (the results are low-risk but the fetus actually has the condition). This is one reason why tests such as cfDNA are considered screening tests; they cannot tell you 'yes' or 'no' for sure if your pregnancy has any of these conditions. It can only tell you if there is an increased or decreased chance.

When it comes to how the test is run, there are slight differences in the technologies that various labs may use. In general, regardless of the technology used, the lab is looking for differences in the overall amount of chromosome material in mom's blood.

For example, if the testing finds an increased amount of material that is from chromosome 21, then the test result would say that there is an increased risk that the pregnancy has <u>Down syndrome</u>. Again, the result from a cfDNA test would never be able to tell you 'yes' or 'no', but can help give you more information about the risk for the conditions tested in your pregnancy.

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for pregnancyrelated questions.

Related Articles

<u>Conditions Screened for with Cell-Free DNA Tests</u>

cfDNA started as a screening test for Down syndrome, but over time the list of conditions that can be screened for is expanding. Most labs now provide screening for Down syndrome, trisomy 18, and trisomy 13, as well as the sex chromosomes (X and Y). Down syndrome, trisomy 18, and trisomy...



• cfDNA Results

Interpreting any prenatal genetic screening result can be challenging. Because of this, it may be helpful to meet with a specialist, such as a genetic counselor, who can review the specifics of your situation to help you understand what your cfDNA results mean for your pregnancy. It is important to...

• cfDNA Testing: No-Call Results

With cfDNA testing, there is a possibility to get a no-call result. A no-call result means that the lab was not able to run the test, or that the test did not produce a result. There are a few possible reasons that prenatal cfDNA screening may not provide a result....

• cfDNA Testing vs. Traditional Screening

There are pros and cons to both cfDNA screening and traditional screening, such as first trimester screening, second trimester screening, sequential screening, and ultrasound. For some conditions, particularly Down syndrome, cfDNA has been shown to be a more accurate screening test than traditional screening tests. cfDNA may also potentially screen...

• <u>cfDNA Testing: How to Decide</u>

The decision of whether or not to pursue prenatal genetic testing is up to you. Your doctor and genetic counselor are available to you with all of the information you need to make an informed decision that fits with your beliefs, values, needs, and personality. Your healthcare providers should also...