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 confined placental mosaicism). 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 Down syndrome. 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 here to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

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