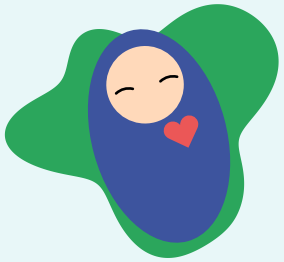
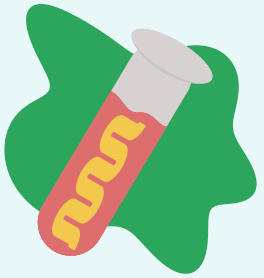
Genetic Support Foundation



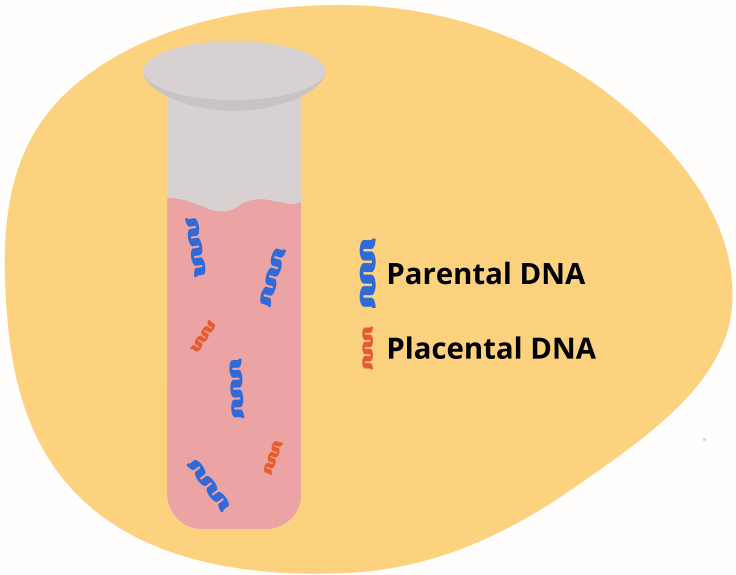
A Patient's Guide to

Cell-Free DNA Screening



What is cell-free DNA (cfDNA) screening?

- cfDNA screening, also known as NIPS or NIPT, is an **optional** blood test that can be performed any time **after 10 weeks** of pregnancy. This screen can tell whether a baby has a higher or lower chance of having a common **chromosome condition**.
- Chromosomes contain DNA, the instruction manuals for how our bodies grow and function, and extra or missing copies of chromosomes can cause **health complications**.



- cfDNA starts with a **blood draw** from the pregnant person. This blood contains the pregnant person's own DNA as well as DNA from the placenta, which gives a clue as to what the baby's DNA looks like.
- The lab will look for differences in the overall amount of DNA that indicate an increased chance that the baby has a chromosome condition.

What conditions does cfDNA screen for?

cfDNA screening looks for multiple chromosome conditions that **vary in symptoms and severity**. While some babies with chromosome conditions pass away shortly after birth, others may simply need extra support to live long and fulfilling lives. Most labs screen for the following conditions:

- **Down syndrome** is caused by an extra copy of chromosome 21. People with Down syndrome can have variable intellectual differences and health issues including heart defects, digestive complications, and hearing and vision problems.
- **Trisomy 18** and **trisomy 13** are caused by an extra copy of chromosome 18 and 13, respectively. These conditions result in many complex medical issues, and most babies pass away before or shortly after birth. With medical support, a small percentage live longer lives with significant disabilities.
- **Sex chromosome variations** are caused by extra or missing X or Y chromosomes. The symptoms of these conditions vary widely, with some individuals having only mild symptoms and others experiencing infertility, learning differences, and heart and kidney problems. There is also a high risk of miscarriage in pregnancies with a missing X chromosome. Because this screen detects the sex chromosomes, it can also incidentally suggest the sex of the baby.
- **Microdeletion syndromes** are caused by small (*micro*) missing (*deletion*) pieces of chromosomes that contain important information. Different labs will screen for different microdeletion syndromes, and the health issues associated with these conditions vary from mild to significant. As this is a newer screen, there are currently no medical guidelines in place to support cfDNA screening for microdeletion syndromes.

What are the possible results?

Negative/Low Risk

- A negative result means there is a **decreased chance** that the baby has one of the screened chromosome conditions.
- cfDNA screening detects the vast majority of these conditions, so it is very likely that the baby does not have one of these conditions.

Positive/High Risk

- A positive result means there is an **increased chance** that the baby has one of the screened chromosome conditions. However, this does *not* tell you whether or not they definitively have this condition.
- A diagnostic test is needed to provide a definitive "yes" or "no" answer.

Other Possible Results

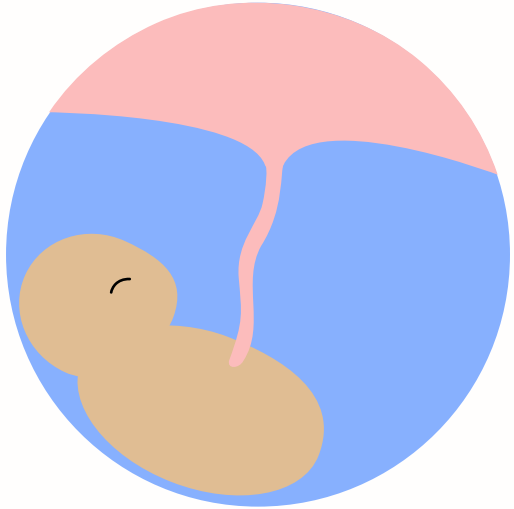
It is also possible to get a result that is neither positive nor negative.

- **No result**: The lab may not be able to perform the test due to poor DNA quality, low amounts of DNA (influenced by factors such as gestational age and the pregnant parent's weight), or shipping errors.
- **Unexpected result**: In some cases, cfDNA screening can find evidence of other health conditions, such as cancer in the pregnant parent or genetic differences in either the parent or the baby.

What are my options if there is a positive result?

Diagnostic Testing

- Diagnostic testing can **definitively determine** whether or not the baby has a particular genetic condition, and can look for many more conditions than cfDNA screening.
- These tests are more invasive than a blood draw and carry a small (<0.5%) risk of miscarriage. The decision of whether or not to pursue diagnostic testing would be up to you.



Chorionic villus sampling tests the tissue from the placenta for chromosome and genetic conditions, and can be performed between 11 and 14 weeks.

Amniocentesis tests the fluid from around the baby for chromosome and genetic conditions, and can be performed between 15 and 22+ weeks.

Pregnancy Options

After receiving a prenatal diagnosis, there are several different pregnancy options.

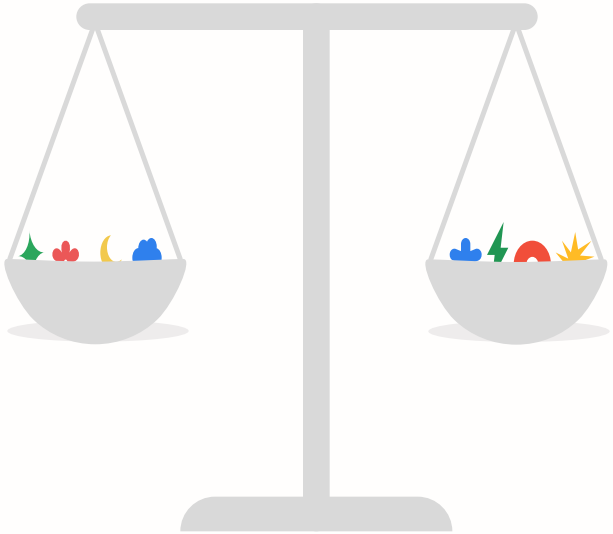
- Some decide to **continue the pregnancy** and raise the baby. Knowing about a diagnosis ahead of time can allow them to gather more information, connect with other families, and prepare for their baby's care.
- Others feel they are not able to care for a child with complex medical needs, and choose to look into support for **adoption** or pregnancy **termination/abortion**.

How do I decide if cfDNA screening is right for me?

The decision of whether or not to pursue cfDNA screening is **entirely up to you**, and your providers are here to offer information and support you regardless of your decision.

Reasons cfDNA screening may be right for you:

- Negative results would make you feel reassured and less anxious throughout your pregnancy.
- You would want to pursue diagnostic testing if there were a positive result.
- You would want to know about a chromosome condition ahead of time to learn about the condition, identify support resources, and prepare for your baby's care.
- You would want to change your pregnancy plans if you knew about a chromosome condition ahead of time.



Reasons cfDNA screening may not be right for you:

- You feel that non-definitive results would be unhelpful to you or cause significant anxiety.
- You would not want to pursue diagnostic testing if there were a positive result.
- You would not want to know about a chromosome condition ahead of time, and would prefer to find out after the baby is born.
- You would not change your pregnancy plans if you knew about a chromosome condition ahead of time.

Resources for more information:

Visit **geneticsupportfoundation.org/pregnancy-genetics** for more information on:

- cfDNA screening
- Diagnostic testing
- Chromosome conditions
- Genetic testing decisions

Talk with a genetic counselor:

Visit **geneticsupportfoundation.org/appointments** to schedule a secure video appointment with a certified genetic counselor.

I WANT TO SCHEDULE