

[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 13](#) are all caused by an extra chromosome.

Screening for [sex chromosomes](#) can help predict the sex of the baby, but it can also screen for [extra or missing sex chromosomes](#). These [sex chromosome](#) differences can vary widely from mild with no notable physical or developmental differences to severe and life-limiting in rare cases. There are four more common sex chromosome differences that are screened for: [Monosomy X/Turner syndrome \(45,X\)](#), [Triple X \(47,XXX\)](#), [Klinefelter syndrome \(47,XXY\)](#), and [Jacobs syndrome \(47,XYY\)](#).

Some labs have recently started offering screening for a set of genetic conditions called [microdeletion syndromes](#). [Microdeletions](#) are when a small piece of a chromosome is missing, rather than an entire chromosome. There are currently no medical guidelines in place to support using [cfDNA](#) to screen for [microdeletion syndromes](#), but there may be instances where this information could be helpful. Some [microdeletion conditions](#) that labs are screening for are:

- [22q11 deletion syndrome](#)
- 1p36 deletion syndrome
- Angelman syndrome
- Prader-Willi syndrome
- [Cri-du-chat](#)
- [Wolf-Hirschhorn syndrome](#)
- [Jacobsen syndrome](#)
- [Langer-Giedion syndrome](#)

Because there is great variation in the conditions that may be screened for, it is important to discuss these options with your provider to make a decision that is best for you and your family.

Click [here](#) to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Related Articles

- [cfDNA Testing: How Does it Work?](#)

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...

- [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...

- [cfDNA Testing: How to Decide](#)

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...