# Cell-free DNA Screening: what you need to know

*also known as non-invasive prenatal testing (NIPT) or non-invasive prenatal screening (NIPS)*

## What is cell-free DNA screening, and what conditions does it test for?

Cell-free DNA screening is a blood test offered to pregnant individuals starting from 10 weeks of pregnancy to screen for certain chromosomal conditions. The most common conditions screened for include Down syndrome, Trisomy 18, Trisomy 13, as well as sex chromosomal conditions like Turner syndrome and Klinefelter syndrome. In some cases, deletion syndromes such as DiGeorge syndrome and Cri-du-chat syndrome can also be screened for.

## Understanding chromosomes:

Chromosomes are structures that contain our genes. Genes, around 20,000 in number, need to fit inside each of our cells. They package themselves into chromosomes, like a tightly wound string, allowing them to fit within our cells. If we were to unravel a chromosome, it would be an extremely long string of genes. Normally, there are a total of 46 chromosomes, each containing approximately 500-1000 genes. Missing or extra chromosomes can lead to birth defects, intellectual disabilities, or other health complications.

## How does the screening work?

During pregnancy, a pregnant individual's blood contains fragments of their DNA, as well as DNA from the placenta. The laboratory analyzes these DNA fragments to determine which chromosomes they come from. Each chromosome should have a specific amount of DNA. If there is an abnormal amount of DNA from a particular chromosome, it increases the chance of the pregnancy having the associated condition. Screening results may not be 100% accurate, and there is a small chance of missed or misdiagnosed conditions.

## Understanding positive and negative results:

If the screening test is positive, it means there is an increased chance that the pregnancy could have the condition being screened for. However, a positive result does not confirm the presence of the condition. Additional diagnostic testing, such as amniocentesis or chorionic villi sampling during pregnancy or after birth, is necessary to confirm the diagnosis. Ultrasound can provide further information but cannot diagnose chromosomal conditions. It is important to remember that a positive result only increases the chance of the condition, but it does not guarantee the presence of the condition.

On the other hand, if the screening test is negative, it reduces the chance of the tested conditions being present. However, it does not completely eliminate the possibility. It is essential to understand that regardless of age or family history, every person has a chance of having a pregnancy with a chromosomal condition, which is why screening is offered.

## Who should consider cell-free DNA screening?

Cell-free DNA screening can be considered by any pregnant individual who wants to gain information about these conditions. However, it is important to note that this screening is optional, and individuals should discuss it further with their healthcare provider to make an informed decision. The decision to undergo cell-free DNA screening is a personal choice, and healthcare providers are available to provide guidance and answer any questions you may have.

For more information, please speak to your healthcare provider.

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