TYPES OF RESULTS

Low Risk (negative):

- There is a minimal chance that the pregnancy has the conditions screened for.
- While reassuring, low risk results does not guarantee the fetus does not have a chromosome difference.

High Risk (positive):

- There is an increased chance that the pregnancy has one or more of the conditions screened for.
- This result does not tell us for sure if there is a chromosome difference. Diagnostic testing is needed to know for sure.

No call / Indeterminate (unknown result)

- The test was not able to provide a clear answer about the chance of certain chromosomal differences.
- This can happen for various reasons such as not enough fetal DNA in the blood sample.
- It does not mean something is wrong, but it might require a repeat test or other testing methods for more information.

OPTIONS BESIDES CFDNA SCREENING

- Ultrasound: can give information about fetal sex and some birth defects.
- Triple or Quad Screen: Blood screenings that can give information on the risk for trisomies, neural tube defects, and abdominal wall defects.
- Diagnostic Tests: Chorionic Villus Sampling or Amniocentesis .
  - Able to provide a diagnostic result (yes or no answer)
  - More comprehensive testing
  - More invasive

BENEFITS & LIMITATIONS

Having the screening done

Benefits	Limitations
• Non-invasive (does not need a sample from the pregnancy directly) • High accuracy for chromosomal trisomy 21, 18, and 13 • More information earlier in pregnancy; can help with decision making	• Not diagnostic • Not 100% accurate • Does not screen for all chromosomal differences • May not be covered by insurance

Not having the screening done

Benefits	Limitations
• No screening anxiety • Less testing • May result is less costs	• Less information earlier in pregnancy • May delay preparation for having an affected pregnancy

Making your own decision

Reasons for:

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Reasons against:

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THE UNIVERSITY OF ARIZONA
COLLEGE OF HEALTH SCIENCES
Genetic Counseling Graduate Program

Prenatal cell-free DNA screening:

Is it right for me?

This resource will provide a brief overview of prenatal cell-free DNA screening, what it screens for, the benefits and limitations, and other options.

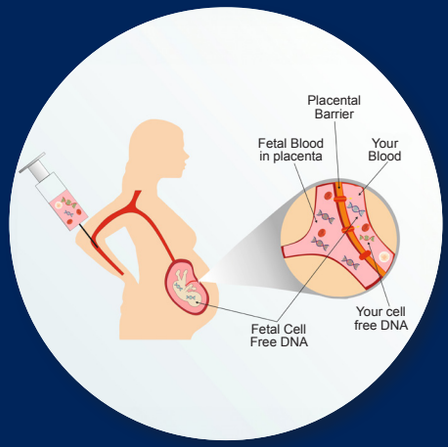
It is intended as a guide to open a discussion with your provider on what is best for you.

WHAT IS PRENATAL CELL-FREE DNA SCREENING?

Also called non-invasive prenatal screening (NIPS) or non-invasive prenatal test (NIPT), cell-free DNA screening (cfDNA) uses your own blood sample to screen for some chromosome differences.

During pregnancy, some of the DNA from the placenta passes into your bloodstream. The cfDNA screening will check this DNA to see if the pregnancy has an increased chance of a chromosome difference.

It is important to remember that this screening is not a diagnostic test. It will give information, but it will not tell you for certain if your pregnancy has a chromosome difference.



WHO IS IT FOR?

The screening offered to all people who are pregnant.

It can be performed anytime after 9-10 weeks of pregnancy.

QUESTIONS TO CONSIDER

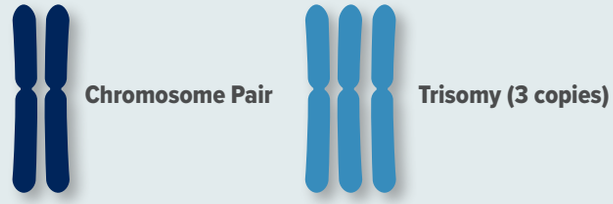
- Are you ready to possibly learn more than just the sex of the pregnancy?
- Will this screening cause anxiety while you wait for the results?
- Do you feel pressured to have the screening done?

WHAT DOES IT TYPICALLY SCREEN FOR?

Differences in the number of chromosomes

In each of our cells, we usually have 46 chromosomes, grouped into 23 pairs. These pairs are made up of one chromosome from the biological mother's egg and another from the biological father's sperm.

Chromosomes are structures that organize our DNA. Our DNA provides our body with instructions on how to develop and function.



When there is one extra copy of a chromosome it is called a trisomy. A trisomy changes how the body will develop and function.

Prenatal cfDNA screening will look at chromosomes 21, 18 or 13 for trisomies.

- Trisomy 21 (also known as Down syndrome) is the most common trisomy that infants are born with. People with Down syndrome typically have characteristic physical features, developmental delays, and intellectual disabilities.
- Trisomy 18 (also known as Edwards syndrome) and trisomy 13 (also known as Patau syndrome) are rarer. Many pregnancies that have one of these conditions will miscarry. Infants who have trisomy 18 or trisomy 13 typically die within the first year of life.

~ This screening does not look for all chromosome differences or other health conditions. Talk with your healthcare provider about your screening/testing options.

Biological sex & more!

The screening can give information about the biological sex of the pregnancy. It will also count the number of sex chromosomes.

The 23rd pair of chromosomes are also known as the sex chromosomes.

- Biological females typically have two X chromosomes.
- Biological males typically have one X and one Y chromosome.

Differences in sex chromosomes can include an extra or missing X or Y chromosome.

When this happens, it leads to differences in development and can affect overall health. A person with differences in the number of their sex chromosomes may have distinct physical features, learning differences, mental health conditions, and other health conditions.

Missing or extra pieces of DNA

Sometimes small pieces of the chromosome are missing (microdeletion) or there may be extra parts of it (microduplication).

This affects how much DNA there is. Sometimes if there is too much or not enough DNA it can lead to differences in development and overall health.

~ Depending on the specific lab your provider is using, prenatal cfDNA screening may or may not be looking for these types of changes.