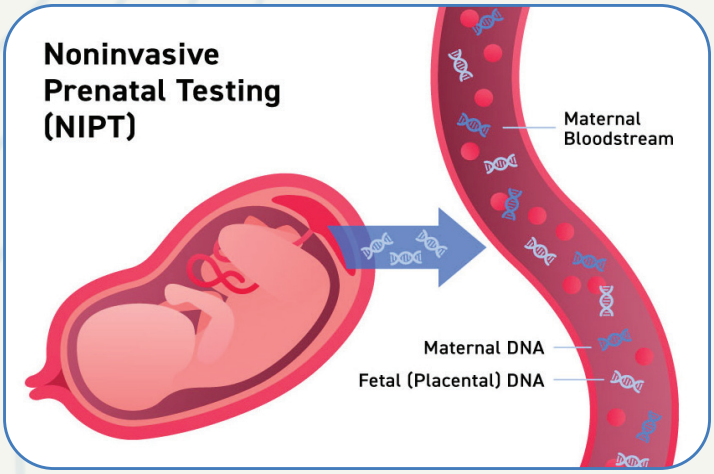
Prenatal cfDNA *screening*

Washington State Department of HEALTH

Patient Fact Sheet

WHAT is Prenatal cfDNA?



Prenatal cell-free DNA screening, also called cfDNA screening, is a blood test often done after the 10th week of pregnancy to examine placental DNA in the pregnant person's bloodstream.

Prenatal cfDNA screening can identify if your pregnancy is at an increased risk to have certain chromosomal disorders called aneuploidies (extra or missing a copy of a chromosome).

Prenatal cfDNA is DNA from the placenta that is present in the pregnant person's bloodstream during pregnancy. Placental DNA is usually identical to the baby's DNA, and moves around in the pregnant person's blood when they are pregnant.

This screen goes by different names such as non-invasive prenatal screening or testing. But no matter the name, it's a way for providers to learn more about the baby. This screen can also predict the sex of the baby.

WHO should be offered Prenatal CfDNA screening?

Prenatal screening should be discussed and offered by the care provider to all pregnant patients regardless of age or risk factors. However, check with your insurance provider if they cover prenatal cfDNA screening.

For pregnancies with more than one baby, discuss with your provider which screening option is right for you.

Patients with active cancer should speak with a specialist on any limitations for the screening.

WHEN should they be screened?

Pregnant patients, regardless of their age, who are at least 10 weeks pregnant are routinely offered this screening.

WHAT will you learn from the screening results?

cfDNA screens for aneuploidies or chromosomal conditions in your pregnancy. The chromosomal conditions it screens for are:

- **Down syndrome (trisomy 21)**
- **Patau syndrome (trisomy 13)**
- **Edwards syndrome (trisomy 18)**

The screen also identifies an abnormal number of sex chromosomes known as aneuploidy and predicts the sex of the baby.

It is important to note that while a typical screening result may be comforting, it does not ensure a healthy pregnancy. If there are any unexpected results, your provider will discuss this with you.

Additionally, not all genetic disorders or birth defects are screened for by cfDNA.

Prenatal screening options:

Which one is best for me?

There are many prenatal genetic screening options available today that can provide information about the possibility of a chromosomal or genetic condition in the pregnancy.

Choosing which set of screening tests to undergo, or whether to undergo any of these tests at all in pregnancy is your choice. One of the most important questions to consider when deciding about prenatal testing is: What will this information mean for you? Knowing the answer to this question can help you make the choices that are right for you.

Questions to consider regarding prenatal cfDNA screening include:

- How would you feel if results indicated a higher chance for a genetic condition or birth defect?
- Would you consider a diagnostic test, such as through amniocentesis if a screening test indicated an increased chance for a genetic condition?
  - If not, would you be ok waiting until the baby is born to know for sure if the condition is present?
- Do you think this information would help you feel more prepared?
- Does more information with the possibility of uncertainty make you anxious?

Video Resources:

Prenatal Genetic Testing: Making an Informed Decision

(PLAY ALL)

- How to decide about prenatal testing
- What conditions are commonly tested for during pregnancy?
- Prenatal cell-free DNA testing
- Prenatal ultrasound
- Amniocentesis
- Chorionic Villus Sampling (CVS)
- Genetic Carrier Screening

Some people feel that prenatal genetic screening is not right for them. Consider these questions:

- Would you prefer not to face the decision of whether or not to have a diagnostic test if your screening test came back with a result showing your baby has a higher chance of having a genetic condition?
- Are you confident that even if the baby did have a genetic condition, it wouldn't change your pregnancy plans?

There are many prenatal screening tests available, and your healthcare professional will be able to determine, based on a number of factors, which prenatal testing options are the most appropriate to offer you. The decision to undergo prenatal genetic screening is personal and should reflect your values, personality, beliefs, and needs.

To learn more about prenatal genetic screening and additional resources, visit:

Prenatal Genetic Testing – Washington State Department of Health

(doh.wa.gov/prenatal-genetics-for-patients)

What is a normal result?

A normal, or “low risk”, result means there is a low chance that the pregnancy has

- Trisomy 21 (Down Syndrome)
- Trisomy 18
- Trisomy 13

Other conditions may also have been included on your screening and would also have a lowered risk with this result.

These low-risk results do not guarantee that the pregnancy does not have these conditions, as no prenatal screening can detect all birth defects or genetic conditions.

Speak with your provider for additional details and other testing that should be offered, even with low-risk results.

What is an abnormal result?

An abnormal, positive, or “high-risk”, result means there is a higher chance that this pregnancy has the condition, and additional testing should be discussed with your healthcare provider.

These results do not guarantee this pregnancy has the condition. No irreversible decisions should be made based on this screening alone.

Talk with your healthcare provider about next step options, including possible additional imaging or diagnostic testing.

Prenatal Terms Defined

Amniocentesis: A procedure by which a sample of amniotic fluid is withdrawn from the amniotic sac. This is usually done by inserting a long needle through the abdominal and uterine walls with the guidance of ultrasound.

Aneuploidy: Also called a chromosomal condition/ disorder, it is the occurrence of an extra or missing copy of a chromosome. See Trisomy and Monosomy.

Chorionic Villus Sampling (CVS): A procedure during pregnancy in which a sample of cells from the placenta is removed to check for possible genetic abnormalities.

Down syndrome (trisomy 21): A genetic condition in which there are three copies of chromosome number 21 instead of two.

Edwards Syndrome (trisomy 18): A genetic condition in which there are three copies of chromosome number 18, instead of two.

Microdeletions: Chromosomal deletions that are too small to be detected by light microscopy using conventional cytogenetic (a branch of genetics) methods. Also known as submicroscopic deletions

Monosomy: Refers to the condition in which only one chromosome from a pair is present in cells rather than the two copies usually found in each cell. See Aneuploidy.

Prenatal Terms Defined (Continued)

Patau syndrome (trisomy 13): A genetic condition in which there are three copies of chromosome number 13 instead of two.

Sex Chromosomes: A sex chromosome is a chromosome that determines the sex of the individual. There are two sex chromosomes, X and Y, that in combination determine the sex of an individual. Males are XY and females are XX.

Trisomy: Refers to the condition in which there are three copies of a chromosomes instead of the usual complement of two of each chromosome found in diploid cells. See Aneuploidy.

Please refer to the ***Prenatal Genetics Glossary*** listed below for a more comprehensive resource.

Options for Follow-Up

Your provider will talk with you about your results. If the results show a high risk for aneuploidy, you can choose to do further testing during the pregnancy. You can also choose to do no further testing or to test the baby after delivery.

During pregnancy, there are two tests you can do to confirm or rule out this condition. Your provider will talk to you about the risks and benefits of these tests. Your provider may also refer you to a specialist to discuss these options.

- In the first trimester, you can choose to have chorionic villus sampling (CVS). During the test, a trained high risk OB specialist (a doctor specializing in high-risk pregnancies) can obtain a small sample of placental tissue using ultrasound guidance. The placenta contains chorionic villi (which are tiny projections of placental tissue), which is used for genetic testing.
- In the second trimester, you can choose to have amniocentesis. During the test, a small sample of amniotic fluid is taken. Amniotic fluid contains cells that have been shed from the baby, which is a normal process.

After delivery, blood can be taken from the umbilical cord and sent for genetic testing.

These tests are all optional and the decision to have more testing is a personal one.

Some pregnant people feel that having more information about the baby will help them make decisions about pregnancy. Having a diagnosis allows people to have choices, such as preparing for special delivery or newborn medical needs, choosing an adoption plan, or not continuing the pregnancy.

The decision to have further testing is complicated, and in part, depends on how one feels about the risk of complications from a procedure versus wanting more information to make decisions and/or prepare for a baby with immediate and/or long-term special health needs. Some families feel they can prepare for the birth of a child with special health needs without knowing a diagnosis.

Washington State Department of HEALTH

DOH 344-086 November 2023 CS

To request this document in another format, call 1-800-525-0127. Deaf or hard of hearing customers, please call 711 (Washington Relay) or email doh.information@doh.wa.gov.