



**Cell-free DNA screening (cfDNA)
(also known as non-invasive prenatal screening (NIPS) or
Non-invasive prenatal testing (NIPT))**

What is cfDNA Screening?

Cell-free DNA (cfDNA) screening is an optional blood test to determine if a pregnancy has a higher chance of having Down syndrome (trisomy 21), trisomy 18, or trisomy 13. These conditions usually happen by chance, do not run in families, and do not happen because of anything either parent does or does not do.

What is Down syndrome?

Down syndrome is the most common genetic condition seen in newborns. It is sometimes called trisomy 21. Babies with Down syndrome have difficulty learning and different facial features. Babies with Down syndrome may also have problems with their heart and other organs.

What are trisomy 18 and trisomy 13?

Both of these conditions are less common than Down syndrome. Babies with trisomy 18 or trisomy 13 usually have multiple birth defects and often do not live very long after birth. Many pregnancies with trisomy 18 or trisomy 13 result in pregnancy loss.

How is cfDNA screening done?

This test involves taking a sample of a pregnant person's blood after 10 weeks of pregnancy. cfDNA screening looks at the small pieces of placental DNA in the pregnant person's blood to determine if a pregnancy is at high risk or low risk for these conditions. Results are usually available within 14 days of blood draw.

Do I need to have an ultrasound before having cfDNA screening?

An ultrasound is recommended before the screening is completed, but does not need to occur on the same day. The ultrasound is needed to confirm the due date, as this screening can only be done after 10 weeks of pregnancy. It is also important to know if the woman is carrying more than one baby.

How accurate is cfDNA screening?

For singleton pregnancies, cfDNA screening will detect 99% of pregnancies with Down syndrome. This means that the test detects 99 out of 100 pregnancies with Down syndrome, so there is a small chance that the test will be normal when the pregnancy has Down syndrome. For trisomy 13 and trisomy 18, detection rate is >91%. There is also a small chance of a "false positive" which means the pregnancy shows a high chance for Down syndrome and the pregnancy does not have Down syndrome.

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How do test results come back?

- A “negative” (low chance) result does not rule-out these conditions in the pregnancy. However, this test will pick up >99% of pregnancies with Down syndrome and 91-96% of pregnancies with trisomy 13 or trisomy 18.
- A “positive” (high chance) result **does not** mean your pregnancy has Down syndrome or another health problem. In this case, we would offer further testing that can get us closer to a yes or no answer.
- Sometimes the testing fails for a variety of reasons. This can indicate a health problem in the pregnancy. In this case, further testing should be offered to you.

Is cfDNA screening covered by insurance?

Some insurances only cover this test when the pregnant person is over 35 or otherwise at increased risk for these conditions. Other insurances may cover in all singleton pregnancies. Self-pay options and financial assistance are available. The CPT code for cfDNA screening in singleton pregnancies is 81420 and the CPT code for the testing our office sends in twin pregnancies is 81507.

What other testing options are available?

- **First trimester screening.** A first trimester screen is a test that combines blood work and ultrasound. Detection rate for Down syndrome is ~85% and detection rate for trisomy 13 and 18 is ~90%.
- **Quad screening or second trimester maternal serum screen:** A quad screen is a blood test that is usually done between 15-20 weeks gestation. Detection rate for Down syndrome is ~80%, detection rate for trisomy 18 is ~60%, and detection rate for open spina bifida is ~80%.
- **CVS and amniocentesis** can test for Down syndrome, trisomy 13, and trisomy 18 with more accuracy. They are considered diagnostic. They can also test for more conditions. These tests involve taking a small sample from the placenta (CVS) or a small sample of the amniotic fluid (amniocentesis). Both CVS and amniocentesis have small risks for pregnancy complications or miscarriage.
- **A targeted ultrasound** in the second trimester can also detect most pregnancies with open spina bifida and may see differences in some pregnancies with Down syndrome, trisomy 18, or trisomy 13.

What if I have more questions?

If you have more questions or are not sure if you want screening, you can request an appointment to meet with a genetic counselor early in your pregnancy to review all of your screening and testing options.

There are educational videos available at mombaby.org/resources/prenatal-screening-options-cfdna/ and at mombaby.org/all-resources.

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