



First trimester screening

What is first trimester screening?

First trimester screening is an optional test to determine if a pregnancy has an increased chance for Down syndrome, 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?

Trisomy 18 and trisomy 13 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 first trimester screening done?

First trimester screening involves a blood test and ultrasound. The ultrasound measures the amount of fluid found at the back of the baby's neck. This is called the nuchal translucency (NT). An increased amount of fluid can mean a higher chance of Down syndrome, trisomy 18, trisomy 13, and other problems.

Why am I scheduled to speak with a genetic counselor?

You will meet with a genetic counselor before the ultrasound to discuss other testing options, review family history, fill out paperwork, and have your blood collected. Your genetic counselor will call you with your result about five days later.

How will the results of my screening come back?

The results will either be reported as "within range" (low chance) or "increased risk" (high chance).

- A normal (low chance) result does not mean you do not have one of these conditions. In the pregnancy. This test will pick up 85% of pregnancies with Down syndrome and 90% of pregnancies with trisomy 18 or trisomy 13.
- An "increased risk" (high chance) result **does not** mean your pregnancy has Down syndrome or another health problem. In this case, we would discuss further testing options that can get us closer to a yes or no answer.

What other testing options are available?

- **Quad screening or second trimester maternal serum screen**: A quad screen is a blood test that is very similar to first trimester screening, but done later in pregnancy. It is usually done between 15-20 weeks gestation.
- **(CVS) and amniocentesis**: CVS and amniocentesis test for Down syndrome, trisomy 18, and trisomy 13 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.
- **Cell-free DNA screening (cfDNA) or non-invasive prenatal testing (NIPT)**: This is a different blood test that screens for Down syndrome, trisomy 18, and trisomy 13. It has been shown to be more accurate at detecting Down syndrome than first trimester screening. This test may miss some other health problems that first trimester screening can pick up. Insurance often does not cover cfDNA or NIPT in women unless they are over 35 years old or at high risk.

How do I schedule first trimester screening?

Tell your healthcare provider as soon as possible if you are interested in first trimester screening.

Will I be offered other screening tests in my pregnancy?

You will be offered a blood test called AFP, or alpha-fetoprotein in the second trimester. to see if the chance for spina bifida or another type of neural tube defect (NTD) is increased. A targeted ultrasound in the second trimester can also detect most neural tube defects.

Is first trimester screening covered by insurance?

First trimester screening is covered by many insurance companies. The billing code (CPT code) for ultrasounds (76801 and 76813) and blood work (84704, 84163, 82105) can be provided to your insurance company to check on coverage. Financial counselors are available at UNC healthcare if you have further questions

What if I have more questions?

If you have more questions or are not sure if you would like to have first trimester 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.

