

Prenatal testing (also called genetic testing) can provide information about your baby's health and development. Testing can provide reassurance that your baby is healthy. It can also alert you and your provider to a concern or help prepare you for a child with special needs.

It's your choice whether to have any of these tests. Some people want as much information as possible, while others do not. Your provider can talk with you about testing options and recommend tests that might best fit your needs.

There are two types of prenatal risk tests:

Screening tests predict the chance that your baby has a certain health condition. These tests are done with an ultrasound or blood test, so the test itself doesn't pose any risk to your baby.

Diagnostic tests show if your baby does or does not have certain health conditions. These tests are invasive and there is a risk that you or your baby could be harmed by having the test. You might choose a screening test first and do a diagnostic test only if the screening test shows your chance of a problem is high.

Information about common tests used during pregnancy are included in this brochure. Please talk with your provider if you have any questions about prenatal testing.

FIRST TRIMESTER

Nuchal translucency (NT) ultrasound This screening test may be done at 11-14 weeks of pregnancy. An NT ultrasound measures the thickness of the fluid at the base of the baby's neck to check for chromosomal problems and other conditions and structural defects.

Combined screen

This screening test may be done at 11-14 weeks of pregnancy, and includes results from an NT ultrasound along with a blood test. The blood test looks at different hormone levels and tells you the chance that your baby has Down syndrome or trisomy 18.

Integrated or sequential screen

Both of these screening tests have two parts that include two blood tests and an NT ultrasound.

The first part is a blood test and NT ultrasound done at 11-14 weeks. The second part is a blood test done at 15-22 weeks. The complete screening test uses the results of both blood tests and the ultrasound to tell you the chance that your baby has Down syndrome, trisomy 18, or spina bifida.

Cell-free DNA test*

This screening test can be done between 10-22 weeks and is currently the most accurate blood screen available to detect for Down syndrome and other chromosome problems.

It checks the fetal DNA found in mom's blood during pregnancy. It can also tell the gender of your baby and screen for sex chromosome abnormalities. It can detect a problem in 99 out of 100 women. If you have a positive result, we recommend a diagnostic test such as CVS or amniocentesis to confirm the diagnosis.

If you have cell-free DNA testing, an AFP blood test at 15-22 weeks is still recommended to screen for spina bifida.

*Only women at high risk of having a baby with a chromosome problem can have this test. High-risk women are those who will be 35 years or older at the time of delivery, have already had a child with a chromosomal problem, or had another type of screening test with abnormal results.

Chorionic villus sampling (CVS)

This is a diagnostic test done between 10-13 weeks that can tell you for certain if your baby has a chromosomal problem. This is an invasive procedure and usually only done if you've had a positive screening test or already had a child with a chromosomal problem. It involves placing a needle through the cervix and taking a small sample of placental tissue. The risk of miscarriage after CVS is 1 to 2 in 100 women.

SECOND TRIMESTER

Alpha-fetoprotein (AFP)

This screening test is done between 15-22 weeks of pregnancy. It is done along with a combined screen or cell-free DNA test (it is included in the integrated, sequential, and quad screens). It is a blood test that looks for a protein that is normally produced by the baby. The level of AFP in the blood can help identify neural tube problems in the baby such as spina bifida.

Quad screen

This screening test involves one blood sample taken between 15-22 weeks. A quad screen tells you the chance that your baby has Down syndrome, trisomy 18, or spina bifida.

Amniocentesis

This is a diagnostic test done between 15-22 weeks that can tell for certain if your baby has a chromosomal problem. It is done if you've had a positive screening test, if you are age 35 or older, or if you have a family history of birth defects.

With the help of an ultrasound to guide the provider, a needle is placed into your abdomen. A small amount of amniotic fluid from the sac surrounding your baby is taken. The risk of miscarriage after amniocentesis is about 1 in 400 women.

Other tests during your pregnancy

Anatomy ultrasound

This test is done around 20 weeks. The ultrasound checks to see that your baby is growing and all major organs are formed. At this stage in your pregnancy, an ultrasound may find problems with your baby's development, such as a heart defect, spina bifida, kidney problem, or cleft lip. Although this test will not diagnose chromosome problems, it may show signs of them or other health conditions.

Genetic disease carrier testing

This is a simple blood test that looks for the presence of common genetic conditions such as cystic fibrosis, Tay Sachs disease, and sickle cell anemia. Conditions are usually tested based on family history and ethnicity. If you and your partner are both carriers for the same genetic condition, your baby could be born with that condition.

Both parents will need to give a small blood sample. If tests show both parents are carriers, an amniocentesis or CVS can be done to tell if the baby has the condition.

Coverage for prenatal tests

Coverage for prenatal tests and exams varies by health plan. Certain prenatal tests, even if recommended, may not be covered depending on your health plan. If you are not at high risk and would like to have certain tests for reassurance, you may need to pay out of pocket.

Check your benefit booklet or call Member Services at 1-888-901-4636 for information about your coverage for prenatal testing.

For more information

Visit the Women's Health website for more information about prenatal testing: <http://www.womenshealth.gov/pregnancy/you-are-pregnant/prenatal-care-tests.html>

Prenatal risk screening

- Screening tests
- Diagnostic tests