

Prenatal Testing Options During Pregnancy

Prenatal Tests

There are a variety of prenatal tests that are available during pregnancy. These tests can provide information about whether there may be a higher than expected chance for certain problems such as chromosome conditions. These tests are done at different times during pregnancy.

The following information is an overview about prenatal testing. You are encouraged to talk with a genetic counselor or your primary care provider. He or she can review your risk factors and help you decide if prenatal testing is right for you.

Genetic Counseling

A genetic counselor will review your family history, talk about testing options and provide information about the risk of birth defects.

He or she will also explain what the potential results might mean and how this information will be used to care for you during your pregnancy.



First Trimester Screening (Nuchal Translucency)

The first trimester screening is usually done between 11 and 14 weeks of pregnancy. It uses ultrasound to measure the fluid area behind your baby's neck (nuchal translucency).

This information is combined with a blood test to measure two proteins made by your baby and placenta (PAPP-A and HCG).

This test can help identify if you are at an increased risk of carrying a baby with a chromosome condition such as Down syndrome or trisomy 18.

If you are at high risk, you will be offered a diagnostic test (chorionic villi sampling or amniocentesis) to determine if there is a chromosomal condition or rule it out.

Second Trimester Screening (Quadruple Test)

The second trimester screening is usually done between 15 and 21 weeks of pregnancy. It is a blood test to measure several proteins made by your baby and placenta.

This test can help identify if you are at an increased risk of carrying a baby with a chromosome condition such as Down syndrome or trisomy 18.

One of the proteins measured, called alpha-fetoprotein (AFP), helps identify most babies with openings in the skull, spine (spina bifida) or abdomen.

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If you are at an increased risk, you will be offered an amniocentesis to determine if there is a chromosomal condition or rule it out. If the results show an increased level of AFP, an ultrasound will be recommended.

Sequential Screening

Sequential screening includes the first trimester screening and two blood tests (the first between 11 and 14 weeks and the second between 15 and 21 weeks).

This test has a higher detection rate for Down syndrome and trisomy 18 compared to the first trimester screening or second trimester screening alone.

Initial results will be given to you after the first trimester screening. Final results will be given to you after the two blood tests.

If you are at an increased risk, you will be offered an amniocentesis to determine if there is a chromosomal condition or rule it out. If the results show an increased level of AFP, an ultrasound will be recommended.

Noninvasive Prenatal Testing

Noninvasive prenatal testing is a screening test. It measures the amount of DNA from the placenta in your blood.

This test has a high detection rate for the most common chromosome conditions including Down syndrome, trisomy 18, trisomy 13 and sometimes sex chromosome conditions.

This test can be done any time after 10 weeks of pregnancy.

While the detection rate is high, false-positive and false-negative results can occur. For this reason, a diagnostic test (chorionic villi sampling or amniocentesis) is recommended for confirmation of a high-risk result.

You and your health care provider will decide if this test is right for you.

Ultrasound

An ultrasound is recommended between 18 to 20 weeks of pregnancy. It can help find problems with the growth and development of your baby, including physical birth defects.

Some findings seen on an ultrasound may increase the chance that your baby could have Down syndrome or another chromosome condition. However, ultrasound alone cannot find genetic conditions. A normal ultrasound will reduce the risk for genetic conditions and other birth defects.

Chorionic Villi Sampling

Chorionic villi sampling (CVS) is a diagnostic test. It is usually done between 10 and 13 weeks of pregnancy.

The test uses ultrasound to guide a catheter through your vagina and cervix to your placenta. A small sample of chorionic villi (cells that form the placenta) will be removed and sent to the lab. The sample can also be taken using ultrasound to guide a needle through your abdomen.

CVS is more than 99 percent accurate in finding chromosome conditions such as Down syndrome.

There is a small chance of miscarriage (about 1 miscarriage in every 450 tests).

Amniocentesis

An amniocentesis is a diagnostic test. It is usually done after 15 weeks of pregnancy.

This test uses ultrasound to guide a thin needle into your lower abdomen and into the amniotic sac. A small sample of amniotic fluid is removed. The amniotic fluid will then be sent to the lab.

An amniocentesis is more than 99 percent accurate in finding chromosome conditions such as Down syndrome. It can also find neural tube defects such as spina bifida.

There is a small chance of miscarriage (about 1 miscarriage in every 900 tests).