

Screening for Chromosome Abnormalities/Genetic Conditions

Babies with chromosomal conditions have a problem in one or more of their chromosomes. Each person has 23 pairs of chromosomes or 46 in all. For each pair, you get one chromosome from your mother and one chromosome from your father. About 1 in 150 babies is born with a chromosomal condition. Down syndrome is an example of a chromosomal condition. Because chromosomes and genes are so closely related, chromosomal conditions are also called genetic conditions.

What problems can chromosomal conditions cause?

Sometimes chromosomal conditions can cause miscarriage. This is when a pregnancy stops growing before 20 weeks. More than half of miscarriages are caused by chromosomal conditions. These conditions also can cause stillbirth, which is when a baby dies in the womb before birth but after 20 weeks of pregnancy. Each child born with a chromosomal condition is different. Some children with chromosomal conditions have intellectual disabilities or birth defects, or both. Some children with these conditions don't have any serious problems. The problems depend on which chromosomes are affected and how.

How do you know if your baby has a chromosomal condition?

Screening versus Diagnostic Tests

The American Congress of Obstetricians and Gynecologists (ACOG) recommends that all pregnant women be offered prenatal tests for Down syndrome and other chromosomal conditions. A screening test is a test to see if you or your baby is more likely than others to have a certain health condition.

You can have screening tests in the first or second trimester of pregnancy. First trimester screening is done at 11 to 13 weeks of pregnancy. Along with a blood test, you get a special ultrasound that checks the back of your baby's neck. Testing in the second trimester is called maternal blood screening. You can get this blood test between 15 and 20 weeks of pregnancy.

If a screening test shows that your baby may have a problem, your provider gives you a diagnostic test. This is a medical test to see if you do or don't have a certain health condition. Diagnostic tests include amniocentesis or chorionic villus sampling. Your provider also can check your baby's blood for chromosomal conditions after he/she is born.

What screening tests are available?

There are a variety of approaches to screening. Some tests are carried out in the first trimester and some in the second. They may include blood tests, an ultrasound scan, or, ideally, a combination of the two.

Different tests have different detection rates. The detection rate tells you how good the test is at identifying babies who are truly affected as being high risk.

First trimester screening or Early Risk Assessment (ERA): This includes a blood test that measures the levels of two proteins in your blood and a special ultrasound scan called a nuchal translucency (NT) test.

- The blood test measures two proteins that are produced by your placenta: free Beta-hCG and PAPP-A. A woman who is carrying a baby with Down syndrome is more likely to have abnormal levels of these two proteins in her blood. The blood test can be done from 10 weeks through the end of 13 weeks.
- The NT test measures the clear (translucent) space in the tissue at the back of your baby's neck. Babies with chromosomal abnormalities tend to accumulate more fluid at the back of their neck during the first trimester, causing this clear space to be larger. The NT screening can only be done between 11 weeks of pregnancy through the end of 13 weeks. (Because this test requires special training and equipment, it may not be available in many smaller institutions.)

Low Risk does not mean zero risk: together, these tests are known as the first-trimester combined screening. They will identify as high risk about 79 to 90 percent of babies with Down syndrome (depending on which study you look at). This means that up to 21 percent of babies with Down syndrome will not be identified by this test.

The advantage of first trimester screening is that you learn about your baby's risk for chromosomal problems relatively early in the pregnancy. If the risk is low, the results will offer you some reassurance. If the risk is high, you may have the opportunity to consider CVS, which will allow you to find out whether your baby has a problem while you're still in your first trimester. (Keep in mind that CVS is usually done no later than 13 weeks, 6 days, so to keep all of your options open, it's best to complete your first trimester screening sooner rather than later.)

Second trimester screening involves a blood test, commonly known as the quadruple screen. This test is usually done between 15 and 18 weeks of pregnancy. The test measures the levels of four substances in your blood: AFP, hCG, uE3, and inhibin A. Having abnormal levels of these substances in your blood is associated with an increased likelihood of carrying a baby with Down syndrome. The test detects about 80 percent of babies with Down syndrome.

Second trimester screening is generally offered in conjunction with first trimester screening as part of what's known as integrated or sequential screening (see below).

- Integrated and sequential screening: Integrated and sequential screening involves both first- and second-trimester screening tests. Integrated screening looks at the results of the first trimester blood test, the NT measurement, and the second-trimester quadruple screen. It gives you a single assessment of your risk after all of these screening tests are completed.
 - Integrated screening detects 90 to 96 percent of Down syndrome cases. If the NT scan is not available, you may have serum integrated screening, which looks at the results of the blood tests from the first and second trimesters. This approach detects about 85 to 88 percent of Down syndrome cases. Integrated screening is the most accurate screening method currently available. Unfortunately, you don't get any results until the second trimester.
- Your caregiver may offer you an alternate strategy, known as sequential screening. With this approach, you get a first trimester risk assessment after the NT screening and the first blood test. If the result indicates that your baby is at increased risk of having a chromosomal abnormality, you'll be offered diagnostic testing at that time, with the opportunity to consider CVS.