

If I am "screen positive" what additional testing is available?

If a screening test is abnormal, it does not necessarily mean that the baby has one of these birth defects. In fact, most women who have abnormal screening results will have normal, healthy babies. If you "screen positive", your doctor will offer you one of the following diagnostic procedures:

- **Chorionic villi sampling (CVS)** is a procedure that takes a small amount of tissue from the developing placenta. The tissue is then sent to a laboratory for chromosome analysis. CVS is performed between 10 and 12 weeks of pregnancy. CVS is associated with a small risk of miscarriage.
- **Amniocentesis** is a procedure that withdraws a small amount of fluid that surrounds the fetus. The fluid is then sent to a laboratory to test for chromosome abnormalities. An amniocentesis is usually performed around the 16th week of pregnancy. Amniocentesis is also associated with a small risk of miscarriage; however, the risk is lower than that for CVS.

FirstScreen does not screen for open neural tube defects. Open neural tube defects occur when the baby's neural tube does not close completely and an opening remains along part of the baby's spine or head. Open neural tube defects occur in about 1 out of every 1,500 live births. A second trimester blood test called MS-AFP, or an ultrasound, is required to detect open neural tube defects.

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FirstScreen

When a woman finds she is pregnant, she faces many choices. One important choice is whether to have a maternal serum screening test, such as FirstScreen, to determine if she is at increased risk of having a baby with certain birth defects.

The good news is that most babies are born healthy and that FirstScreen is a non-invasive test. This brochure provides some information to help you decide whether to have this test. If you have any additional questions, please speak with your doctor.

What is FirstScreen?

FirstScreen is a blood test which shows if you are at increased risk of having a baby with Down syndrome or trisomy 18. It requires a sample of your blood and an ultrasound measurement performed in the first trimester of pregnancy.

What is Down syndrome?

Down syndrome is caused by the presence of an extra chromosome #21 and results in both mental and physical abnormalities. Approximately 1 in 800 babies is born with Down syndrome. The risk of having a child with Down syndrome gradually increases with the age of the mother, but can occur at any maternal age.

What is trisomy 18?

Trisomy 18 is caused by the presence of an extra chromosome #18 and results in serious mental retardation and physical deformities, including major heart defects. Approximately 1 in 6500 babies is born with trisomy 18. Only 1 out of 10 babies affected with trisomy 18 lives past the first year of life. As with Down syndrome, the risk of having an affected child gradually increases with the age of the mother.

How is FirstScreen performed?

Between the 10th and 14th week of your pregnancy, a small amount of blood is drawn from your arm and the levels of two proteins, PAPP-A and hCG, are measured in a laboratory. These proteins are made by the developing baby and the placenta, and are found in every pregnant woman's blood. However, when a fetus is at risk for Down syndrome or trisomy 18, the amount of these circulating proteins may be abnormal. The result of an ultrasound measurement, called nuchal translucency, is combined with your blood test result to yield the final screening assessment.

Your specific test result is affected by:

- Exactly how far along you are in your pregnancy on the day that the blood sample is taken and the ultrasound is performed.
- Your weight, ethnic background, and age.
- Whether you are an insulin-dependent diabetic or take certain types of medications.
- Whether a close relative has Down syndrome.

What is nuchal translucency?

Nuchal translucency (NT) is a measurement of the fluid filled space at the back of the developing fetus' neck. Extra fluid in this space indicates that the fetus is at a higher risk for certain birth defects.

What do FirstScreen results mean?

It is important to understand that a screening test does not provide a diagnosis; rather it predicts the likelihood of a defect occurring. FirstScreen can only tell you if there is a greater chance of your baby having Down syndrome or trisomy 18. There are two types of screening test results:

Screen Negative

If the results show measurements within normal range, the chance of having a baby with Down syndrome or trisomy 18 is low. This is called a "screen negative" result. In rare instances, screening will not detect these birth defects as it cannot detect all high-risk pregnancies.

Screen Positive

If the results show abnormal measurements, there is an increased chance of having a baby with Down syndrome or trisomy 18. This is called a "screen positive" result. If your result is screen positive, your doctor will offer diagnostic testing to determine if your baby is affected with one of these birth defects.

First trimester screening leads to the detection of approximately 83% of Down syndrome cases and 80% of trisomy 18 cases.*

*According to several large, multi-center studies