

First Trimester Screening

All women are offered certain screening tests during pregnancy. Diagnostic tests will be offered to women who have abnormal screening test results.

If a woman is already at increased risk of having a baby with a birth defect, she may be offered a diagnostic test first rather than having a screening test. These risk factors may include:

- Age 35 years or older when the baby is due
- Family or personal history of birth defects
- Previous child with a birth defect
- Use of certain medicines around the time of conception
- Diabetes before getting pregnant

First trimester screening is a test that combines the results of a special ultrasound test and blood tests and can be done between 10 and 14 weeks of pregnancy. This test detects signs of Down syndrome, trisomy 18, and heart defects. This type of screening is fairly new and is not done everywhere.

The ultrasound test is called nuchal translucency screening. This test uses ultrasound to measure the thickness at the back of the neck of the fetus. An increase may be a sign of Down syndrome.

The blood test measures the level of two substances in the mother's blood: pregnancy-associated plasma protein-A (PAPP-A) and free-beta human chorionic gonadotropin (hCG). In cases of Down syndrome, hCG levels are higher than expected and PAPP-A levels are lower.

The combined results of the nuchal translucency screening and the blood tests show if the fetus might have a defect. If the results show there is an increased risk of Down syndrome, diagnostic testing can be done to get more information.

If the nuchal translucency is increased, but the combine screening does not show a risk of Down syndrome or trisomy 18 (the hCG and PAPP-A test results are normal), a heart defect could be present. To check on this, a detailed exam of the fetal heart can be done later in pregnancy.

Maternal Serum Screening

With maternal serum (blood) screening, several tests often are performed together. This is known as "multiple marker screening." These tests measure the level of three or four of the following substances in your blood:

- *Alpha-fetoprotein (AFP)* – A substance made by a growing fetus. It is present in amniotic fluid, fetal blood, and, in smaller amounts, in the mother's blood.
- *Estriol* – A hormone made by the placenta and the liver of the fetus.
- *Human chorionic gonadotropin* – A hormone made by the placenta.
- *Inhibin-A* – A hormone produced by the placenta.

The test is called a triple screen or a quad screen depending on the items tested. Tests results can be combined to determine the risk of certain disorders.

For a maternal serum screening test, a small amount of blood is drawn from a vein in your arm and is sent to a lab to be studied. The test results usually are ready within a week after the blood sample is taken.

These tests usually are done around 15-20 weeks of pregnancy. The timing of the test is important because some levels can only provide an accurate reading at a certain point in pregnancy. For instance, a high AFP level can simply mean you are further along in pregnancy than you thought or you are carrying more than one baby.

The test results show if there is an increased risk for neural tube defects, Down syndrome, trisomy 18, or abdominal wall defects. If your results are not in the normal range, your doctor will offer you further testing. In some cases, abnormal levels can be explained with an ultrasound exam.