YOU HAVE THE OPTION

of having the first trimester screening test to determine whether your pregnancy has an increased risk of certain birth defects.

A NONPROFIT ENTERPRISE OF THE UNIVERSITY OF UTAH AND ITS DEPARTMENT OF PATHOLOGY

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MATERNAL SERUM SCREENING

The First Trimester Test

Information for Pregnant Women

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What is the first trimester test?

The first trimester test is a combination of measurements of certain serum markers present in maternal blood and results of an early pregnancy ultrasound. Both the ultrasound and maternal blood draw are done between the beginning of the 11th and the end of the 13th week of pregnancy. The ultrasound is used to date the pregnancy and to measure the nuchal translucency (NT), which is the thickness of the tissue at the back of the fetal neck. The blood tests measure two proteins: pregnancy-associated plasma protein-A (PAPP-A) and human chorionic gonadotropin (hCG).

What type of birth defects can be detected by the first trimester test?

1. **Down syndrome (DS):** Also called trisomy 21, Down syndrome is caused by the baby having an extra copy of chromosome 21. This results in mental retardation and physical problems, the most common of which is a heart defect. About half of all persons with Down syndrome will live to at least age 50.

2. **Trisomy 18 (T18):** Babies with trisomy 18 have an extra copy of chromosome 18. This causes multiple physical problems and severe intellectual disability. Most babies with trisomy 18 do not survive the first year of life.

Open neural tube defects (ONTD) are NOT detected by this test. A separate test, AFP, will be needed later in the pregnancy to screen for these disorders. Blood for AFP testing can be drawn between the beginning of the 14th and the end of the 24th week of gestation. The best time to draw is between 16 and 18 weeks.

How reliable in the first trimester test at

BIRTH DEFECT	DETECTION RATE	SCREEN POSITIVE RATE
Down syndrome	85–90%	6%
Trisomy 18	80%	<1%

My screen came back as "abnormal." What does this mean?

Most pregnancies that have abnormal test results are actually normal pregnancies (the baby does not have DS, T18, or an ONTD). False positives occur because screening tests are designed to identify women who are at increased risk to have a baby with certain birth defects. These screening tests are not diagnostic tests. A positive screening test result does NOT mean that your baby has a birth defect, only that he/she is at increased risk of having one.

What is recommended when a test result is abnormal?

Your doctor or genetic counselor will discuss additional testing that can be done to determine if your baby does or does not have a birth defect. Most often, a detailed ultrasound is recommended. Non-invasive prenatal testing (NIPT), chorionic villus sampling (CVS), or amniocentesis may be offered. NIPT is also a screening test, but one that is more sensitive and has a very low false positive rate. It involves only a blood draw. In CVS, a small piece of the placenta is tested. Amniocentesis involves testing a small amount of the fluid surrounding the baby. Both CVS and amniocentesis allow the laboratory to directly examine the baby's chromosomes to accurately identify DS and T18. Amniocentesis, especially when paired with an ultrasound, can also test for ONTDs. Since CVS and amniocentesis are expensive and have a small risk for miscarriage, the decision to have either of these tests is yours. NIPT is also expensive, and is not diagnostic like CVS and amniocentesis, but does not put the pregnancy at risk and may provide reassurance that the baby does not have DS or T18.

What happens if the follow-up tests show that the fetus has a birth defect?

If a birth defect is detected, you will be given as much information as possible about the condition.

Several options may be available, including increased surveillance of the pregnancy, arrangements for special care at delivery or after the baby is born, or discontinuation of the pregnancy. Your doctor or genetic counselor can discuss your test results and options with you.

Does a negative test result guarantee that my baby does not have a birth defect?

No. The first trimester test is not a diagnostic test and cannot guarantee that a baby does not have a birth defect. It screens for two of the more common birth defects, but a negative test does not mean that the baby has no chance of having T18 or DS, only that it is at a lower risk for these particular birth defects. All pregnancies have a 2–3 percent risk for birth defects, including many for which this test does not screen.

If you would like to learn more about the first trimester test, please speak with your physician or healthcare provider.