

AS AN EXPECTANT MOTHER,

you have the option of
having the integrated test
to determine whether your
baby has an increased
chance of having certain
birth defects.



A NONPROFIT ENTERPRISE OF THE UNIVERSITY OF
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MATERNAL SERUM SCREENING

The Integrated Test

Information for Pregnant Women



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What is the integrated test?

The integrated test is a combination of first and second trimester testing. An ultrasound of the pregnancy and a maternal blood draw are done between the middle of the 10th week and the end of the 13th week of pregnancy. A second blood sample is drawn between 15 and 22 weeks gestation.

The ultrasound dates the pregnancy and provides a nuchal translucency (NT) measurement. This measures the thickness of the tissue at the back of the baby's neck. If the NT measurement cannot be obtained for any reason, you can still have an integrated screen, but it will be slightly less accurate.

The laboratory performs testing on the two blood samples: PAPP-A in the first trimester, and AFP, hCG, uE3, and DIA in the second trimester. The results of these tests, along with the NT measurement (if done), are combined to provide the screen results.

What types of birth defects can be found using the integrated test?

Down syndrome (DS)

Babies with Down syndrome are born with an extra copy of chromosome 21. This causes mild to moderate mental retardation, specific facial features, and sometimes physical problems, such as heart defects. About half of all babies born with Down syndrome will live to at least age 50.

Trisomy 18 (T18)

Babies with trisomy 18 have an extra copy of chromosome 18. This causes multiple physical problems and severe mental retardation. Most babies with trisomy 18 do not survive the first year of life.

Open neural tube defects (ONTD)

Spina bifida and anencephaly are the most common ONTDs. When a baby is born with spina bifida, part of the bone covering the spinal cord does not form correctly and the spinal cord is exposed. Surgery is needed to close the opening. Even with surgery, spina bifida can cause problems ranging from bowel and bladder control difficulties to paralysis of the legs, hydrocephalus (fluid on the brain), and learning disabilities.

Anencephaly occurs when the fetal skull and brain do not develop. Babies with anencephaly cannot survive.

How reliable is the integrated test at finding birth defects?

The integrated test has the highest DS detection rate of all the available screening tests. The real advantage of the integrated test is that this high detection rate is paired with a low screen positive rate. This means that of all screening tests, the integrated test is least likely to come back abnormal when the baby does not have DS.

BIRTH DEFECT	DETECTION RATE	SCREEN POSITIVE RATE
Down syndrome	85–87%	1%*
Trisomy 18	90%	<1%
Neural tube defects	80%	1.5%

*If nuchal translucency cannot be measured, the risk of a chromosome disorder is still calculated, but the screen positive rate is slightly higher.

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. 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 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 is expensive and has a small risk for miscarriage, the decision to have this test 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 and/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 normal test result guarantee that my pregnancy does not have a birth defect?

No. The integrated test is not a diagnostic test and does not detect every case of DS, T18, or an ONTD.

All pregnancies have a 2–3 percent risk of having a birth defect. This test screens for the three most common birth defects, but not for all birth defects.

If you would like to learn more about the integrated test, please talk with your physician, genetic counselor, or other healthcare provider.