THE INTEGRATED TEST

Maternal Serum Screening

Information for Pregnant Individuals

ARUP® LABORATORIES

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.

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 of 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 specific tests on the two blood samples: a pregnancy-associated plasma protein A (PAPP-A) test in the first trimester, and alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), estriol (uE3), and dimeric inhibin A (DIA) tests in the second trimester. The results of these tests, along with the NT measurement (if done), are combined to provide the screen results.

Which types of birth defects can be detected using the integrated test?

Down syndrome

Babies with Down syndrome (DS) are born with an extra copy of chromosome 21. This causes mild to moderate intellectual disability, 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 50 years of age.

Trisomy 18

Babies with trisomy 18 (T18) 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

Spina bifida and anencephaly are the most common open neural tube defects (ONTDs). When a baby is born with spina bifida, part of the bone covering the spinal cord does not form correctly, leaving the spinal cord 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 of 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 of the screening tests, the integrated test is least likely to yield an incorrect high-risk result.

BIRTH DEFECT	DETECTION RATE	SCREEN POSITIVE RATE
Down syndrome	87%	1%*
Trisomy 18	90%	<1%
Open neural tube defects	80%	1–2%

** If NT 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 healthy pregnancies (the baby does not have DS, T18, or an ONTD). False-positive results occur because screening tests are designed to identify women who are at increased risk of having 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, but only that your baby 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 options to determine if your baby does or does not have a birth defect. A detailed ultrasound is

recommended. Prenatal cfDNA screening, previously referred to as noninvasive prenatal testing (NIPT), chorionic villus sampling (CVS), or amniocentesis may be offered. Prenatal cfDNA screening is also a screening test, but it is more accurate and requires only a blood draw. In CVS, a small piece of the placenta is tested. Amniocentesis involves testing a small amount of the amniotic fluid (the fluid that surrounds 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. Because CVS and amniocentesis are expensive and carry a small risk for miscarriage, the decision to have either of these tests is yours. Prenatal cfDNA screening is not diagnostic like CVS and amniocentesis, but it 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 during 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 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% 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.

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A nonprofit enterprise of the University of Utah and its Department of Pathology

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BD-PP-010, Rev 5, July 2024