THE SEQUENTIAL SCREEN

Maternal Serum Screening

Information for Pregnant Individuals

ARUP LABORATORIES

As an expectant mother, you have the option of a sequential screen to determine whether your baby has an increased chance of having certain birth defects.

What is the sequential screen?

The sequential screen is a two-part screening test for certain fetal disorders. 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 24 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, the sequential screen will not be completed and your healthcare provider may discuss other testing options with you.

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

The sequential screen is interpreted for Down syndrome (DS) and trisomy 18 (T18) risk after the first-trimester sample is received, and for DS, T18, and open neural tube defect (ONTD) risk after the second-trimester sample is received. If the risk for either DS or T18 is considered to be very high after the first blood draw, the test result will be "abnormal" in the first trimester, and no second sample will be required. However, because only a small percentage of screens will be called "abnormal" in the first trimester, most women will need to provide a sample in the second trimester, after which they will receive their final results.

Advantages

- Excellent detection of DS and T18
- Low chance of a false-positive result
- Detects ONTDs
- Identifies pregnancies at highest risk for DS and T18 in the first trimester

Disadvantages

- Most women will have two blood draws (in first and second trimesters).

Which types of birth defects can be detected using the sequential screen?

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 DS 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 T18 do not survive the first year of life.

Open neural tube defects

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, leaving the spinal cord exposed. Surgery is needed to close the opening. Even with surgery, spina bifida can cause problems such as issues with bowel and bladder control, 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 sequential screen at finding birth defects?

BIRTH DEFECT	DETECTION RATE	SCREEN POSITIVE RATE
Down syndrome	86%	1.6%
Trisomy 18	90%	<1%
Open neural tube defects	80%	1–2%

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 an increased risk of having 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, but only that your baby is at an 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, and a detailed ultrasound will be 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 amniotic fluid (the fluid that surrounds the baby). Both CVS and amniocentesis enable 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 baby 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 sequential screen is not a diagnostic test and does not detect every case of DS, T18, or an ONTD.

All pregnancies have a 2–3% background 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 maternal serum screening, please talk with your doctor, genetic counselor, or other healthcare provider.

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