

AS AN EXPECTANT MOTHER,

you have the option of
having the sequential
screen 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 Sequential Screen

Information for Pregnant Women



NATIONAL REFERENCE LABORATORY

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 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 cannot be done and your healthcare provider may discuss other testing options with you.

The laboratory performs the following tests on the two blood samples: PAPP-A and hCG in the first trimester, and AFP, hCG, uE3, and DIA in the second trimester. The results of these tests, along with the NT measurement, are combined to provide the test 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 defects (ONTD) 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 will be resulted out as "abnormal" in the first trimester and no second sample will be required. However, since 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 for a false-positive result
- Detects ONTDs
- Identifies pregnancies at highest risk for DS and T18 in the first trimester

Disadvantages:

- Most women will get their result in the second trimester.
- Most women will have two blood draws.

What types of birth defects can be found using the sequential screen?

Down syndrome (DS)

Babies with Down syndrome 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 age 50.

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)

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 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 sequential screen at finding birth defects?

BIRTH DEFECT	DETECTION RATE	SCREEN POSITIVE RATE
Down syndrome	86%	1.6%
Trisomy 18	90%	<1%
Neural tube defects	80%	1.5%

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 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, only that he/she 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 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 which 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 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 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 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 percent 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.