

AS AN EXPECTANT MOTHER,

you have the option of having your blood drawn for a screening test to determine whether your pregnancy has an increased chance of having an open neural tube defect (e.g., spina bifida), Down syndrome, or trisomy 18.

MATERNAL SERUM SCREENING

The Quadruple Screen

Information for Pregnant Women



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UTAH AND ITS DEPARTMENT OF PATHOLOGY

aruplab.com

ARUP LABORATORIES

500 Chipeta Way
Salt Lake City, UT 84108-1221
Phone: 800-522-2787
Fax: 801-583-2712

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What is a maternal serum quadruple (quad) screen?

The quad screen is a blood test that measures four proteins, (AFP, hCG, uE3, and inhibin-A (DIA)), produced by the pregnancy in order to determine if the baby has an increased chance of having spina bifida, Down syndrome (DS), or trisomy 18 (T18).

Typically, the test is performed between 15 and 20 weeks gestation, but may be performed as early as 14 weeks and as late as the end of the 24th week. If the levels of the above substances indicate that the pregnancy is at increased risk for one of these disorders, further tests, such as an ultrasound examination or amniocentesis, may be needed to clarify the test results.

What types of birth defects can be found by the quad screen?

Down syndrome

Babies with Down syndrome are born with an extra 21st chromosome. 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

Babies with trisomy 18 have an extra 18th chromosome. 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 (ONTDs)

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. 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 quad screen at finding birth defects?

BIRTH DEFECT	DETECTION RATE
Down syndrome	80%
Trisomy 18	~80%
Neural tube defects	80%

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 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 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 normal test result guarantee that my pregnancy does not have a birth defect?

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

Additionally, 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 the maternal serum quad screen, please talk with your doctor, genetic counselor, or other healthcare provider.