THE QUADRUPLE SCREEN

Maternal Serum Screening

Information for Pregnant Individuals

ARUP LABORATORIES

As an expectant mother, you have the option of having your blood drawn for a screening test to determine whether your baby has an increased chance of having an open neural tube defect (such as spina bifida), Down syndrome, or trisomy 18.

What is a maternal serum quadruple (quad) screen?

The quad screen is a blood test that measures four proteins produced by the pregnancy, alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), estriol (uE3), and dimeric inhibin A (DIA), 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 of gestation, but it may be performed as early as 14 weeks and as late as the end of the 24th week. If the levels of the above proteins 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.

Which types of birth defects are identified by the quad 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 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. 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 quad screen at finding birth defects?

BIRTH DEFECT	DETECTION RATE
Down syndrome	81%
Trisomy 18	~80%
Open neural tube defects	80%

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 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 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 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% 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.

A nonprofit enterprise of the University of 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

BD-PP-008, Rev 7, July 2024