What is prenatal screening?

Prenatal screening is testing offered to all parents who want to learn information about the health of their pregnancy. Most pregnancies have a low risk for birth defects and genetic conditions. Some women have a family history or medical history that identifies their pregnancy as "high risk". Even in "low risk" pregnancies, 2-3 out of every 100 babies are born with a birth defect. Prenatal screening tests are used to find pregnancies that may be at high risk for certain conditions and need special care.

Two screening tests that are commonly offered during pregnancy are the first trimester screen and second trimester screen (quad screen). It is up to you to decide if you would like to have these screening tests.

First Trimester Screening

The first trimester screen involves combining results from a blood test and an ultrasound. Blood is usually drawn from the mother between 9 and 13 weeks to measure two normal substances (hCG and PAPP-A). An ultrasound is done between 11 and 13 weeks to measure the amount of fluid behind the baby's neck, also called the nuchal translucency (NT). This combined result shows which pregnancies have an increased risk for Down syndrome, trisomy 13, or trisomy 18.

Second Trimester (Quad) Screening

The quad screen involves taking a blood sample from the mother and measuring the amount of four normal substances (AFP, hCG, uE3, and DIA). This test is done between 15 and 22 weeks. This result shows which pregnancies have an increased risk for a neural tube defect, Down syndrome, or trisomy 18.

What do the results mean?

Most women will have a negative prenatal screening test result. These women need regular prenatal care. A negative test does not mean the baby will not have a genetic condition or birth defect. There is no prenatal test that can detect all possible problems.

About 5% of women will have a positive prenatal screening result. Additional prenatal care may be needed. However, a positive screening result does not mean that the baby has a condition.

What do these tests look for?

Down syndrome is caused by an extra copy of chromosome 21. Individuals with Down syndrome have distinctive physical features and an increased risk for birth defects such as heart and bowel problems. People with Down syndrome also have mild to moderate intellectual disability. First trimester screening detects up to 90% of pregnancies with Down syndrome. A quad screen detects 70-80% of pregnancies with Down syndrome. The chance of having a baby with Down syndrome increases with maternal age, so it is more common for women who are 35 or older to have a positive screening test.

Trisomy 13 and **Trisomy 18** are more severe chromosome disorders. They are caused by an extra copy of chromosome 13 or 18. These are not as common as Down syndrome. Babies with either condition usually have many birth defects, severe developmental problems and few survive the first year of life. First trimester screening detects up to 95% of pregnancies with trisomy 13 or 18. A quad screen will detect about 60% of pregnancies with trisomy 18.

Neural tube defects (NTDs), such as spina bifida and anencephaly, are birth defects that involve the spine or brain. Individuals with spina bifida require surgery and may have lifelong physical disabilities. Babies with anencephaly do not survive long past birth. First trimester screening will not detect NTDs. The AFP measurement in a quad screen will detect about 85% of open NTDs.

Will I need other testing?

With a positive screening test, you should be referred to a "high risk" Maternal-Fetal Medicine physician and a genetic counselor. These specialists will review your results and help you make a plan for your pregnancy. You may be offered additional testing like ultrasound, non-invasive prenatal testing, or diagnostic testing.

Ultrasound can be used to look for some birth defects, especially NTDs. The accuracy of ultrasound may depend on the age of the pregnancy and other factors, so additional testing may still be recommended.

Non-invasive prenatal testing is a newer blood test often used for "high risk" patients. It measures DNA from the pregnancy to determine the risk for certain chromosome conditions. It has higher detection rates and lower false positive rates than standard screening.

Chorionic villus sampling (CVS) and amniocentesis are diagnostic tests that can most accurately detect chromosome conditions. Amniocentesis can also confirm NTDs. These two tests do involve removal of tissue or fluid from the pregnancy and have a slight risk of miscarriage.

How can I get more information?

The Greenwood Genetic Center provides genetic services throughout South Carolina. Contact the office nearest you for more information.

Greenville
14 Edgewood Drive
Greenville, SC 29605
1-866-478-4363 (toll free)
(864)250-7944
(864) 455-1600 (prenatal counselors)

Greenwood
106 Gregor Mendel Circle
Greenwood, SC 29646
1-888-442-4363 (toll free)
(864) 941-8100

Charleston
3520 W. Montague Avenue, Suite 104
North Charleston, SC 29418
1-866-588-4363 (toll free)
(843)746-1001

Columbia
1911 Thurmond Mall
Columbia, SC 29201
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Prenatal Screening and Testing

This brochure is published by the Greenwood Genetic Center, a nonprofit institute organized to provide clinical genetic services, diagnostic laboratory testing, educational programs and materials and research in the field of medical genetics.

Greenwood Genetic Center