Prenatal genetic screening tests

Some parents are interested in screening during pregnancy for genetic or inherited type disorders. A variety of genetic screening tests are available during pregnancy.

Genetic screening tests offered by this office include:

- Carrier screening for Cystic Fibrosis, Spinal Muscular Atrophy (SMA), and/or Fragile X Syndrome
- Prenatal genetic screening for Down Syndrome, Trisomy 18, and Open Neural Tube defects

Carrier screening involves a blood sample and can be performed at any time.

Prenatal genetic screening is offered via FIRST trimester screening, second trimester Maternal Serum Screening, and non-invasive prenatal testing by cell-free DNA (NIPT). Second Trimester Maternal Serum screening and NIPT involve taking a sample of the mother's blood, and these two tests are completed in the office. FIRST trimester screening is completed at UNC/Rex in Raleigh; our office is pleased to schedule an appointment for you if are interested in testing.

The genetic screening tests are optional: you choose if you would like to be tested. Many patients ask the question "Should I be tested?" The decision to have testing is a personal decision and is to be decided by you. Please feel free to discuss any questions you have regarding testing with your medical provider.

Additional tests may be appropriate based on your personal medical and/or family history. Your medical provider may make recommendations regarding additional genetic testing.

What is prenatal genetic testing?
Prenatal genetic testing gives parents-to-be information about whether their fetus has certain genetic disorders.

What are genetic disorders?
Genetic disorders are caused by changes in a person's genes or chromosomes. Some medical conditions are caused by missing or extra chromosomes. Inherited disorders are caused by changes in genes called mutations. Inherited disorders include sickle cell disease, cystic fibrosis, Tay–Sachs disease, and many others. In most cases, both parents must carry the same gene to have an affected child.

What are the two main types of prenatal genetic tests?
There are two general types of prenatal tests for genetic disorders:

- Prenatal screening tests can tell you the *chances* that your fetus has chromosomal disorder.
- Prenatal diagnostic tests can tell you whether your fetus *actually has* certain disorders. These tests are done on cells from the fetus or placenta obtained through diagnostic tests like an amniocentesis.

What is first-trimester screening?
First-trimester screening includes a blood test from the pregnant woman and an ultrasound exam. Both tests usually are performed together and are done between 10 weeks and 13 weeks of pregnancy.

What is second-trimester screening?

The second trimester maternal serum screen is a blood test that measures four different substances in your blood. The quad test screens for Down syndrome, trisomy 18, and neural tube defects. It is done between 15 weeks and 22 weeks of pregnancy.

What is cell-free DNA testing?
Cell-free DNA is the small amount of DNA that is released from the placenta into a pregnant woman's bloodstream. The cell-free DNA in a sample of a woman's blood can be screened for Down syndrome, trisomy 13, trisomy 18, and problems with the number of sex chromosomes. This test can be done starting at 9 weeks of pregnancy. It takes about 1 week to get the results. A positive cell-free DNA test result should be followed by a diagnostic test with amniocentesis or CVS.

The cell-free DNA screening test works best for women who already have an increased risk of having a baby with a chromosome disorder. For a woman at low risk of having a baby with a chromosome disorder, conventional screening remains the most appropriate choice. Cell-free DNA testing is not recommended for a woman carrying more than one fetus.

What do the different results of prenatal screening tests mean?
Results of blood screening tests for aneuploidy are reported as the level of risk that the disorder might be present:

- A positive screening test result for aneuploidy means that your fetus is at higher risk of having the disorder compared with the general population. It does not mean that your fetus definitely has the disorder.
- A negative result means that your fetus is at lower risk of having the disorder compared with the general population. It does not rule out the possibility that your fetus has the disorder.

You will be referred for genetic counseling and offered additional testing if your screening test is positive.

How accurate are prenatal genetic screening tests?
With any type of testing, there is a possibility of false-positive results and false-negative results. A screening test result that shows there is a problem when one does not exist is called a false-positive result. A screening test result that shows there is not a problem when one does exist is called a false-negative result. Your health care professional can give you information about the rates of false-positive and false-negative results for each test.

What should I consider when deciding whether to have prenatal genetic testing?
It is your choice whether to have prenatal testing. Your personal beliefs and values are important factors in the decision about prenatal testing.

It can be helpful to think about how you would use the results of prenatal screening tests in your pregnancy care. Remember that a positive screening test tells you only that you are at higher risk of having a baby with Down syndrome or another aneuploidy. A diagnostic test should be done if you want to know a more certain result. Some parents want to know beforehand that their baby will be born with a genetic disorder. This knowledge gives parents time to learn about the disorder and plan for the medical care that the child may need. Some parents may decide to end the pregnancy in certain situations.

Other parents do not want to know this information before the child is born. In this case, you may decide not to have follow-up diagnostic testing if a screening test result is positive. Or you may decide not to have any testing at all. There is no right or wrong answer.