

North Carolina Women's Hospital
First Trimester Serum and Ultrasound Combined Screen: (FIRST)

What is FIRST?

FIRST is a screening test offered to all pregnant women between 11 and 14 weeks of pregnancy. It is a screening test that can tell you if you are at an increased risk for having a baby with certain birth defects or problems. After learning about the FIRST screen, you can decide if you want the test or not.

What birth defects are screened for by the FIRST test?

FIRST screens for two genetic problems. One is called Down syndrome and the other is called trisomy 18. It can also help to find if a baby is at risk for certain heart problems.

What is Down syndrome?

Down syndrome is the most common genetic problem seen in newborns. It is caused by an extra chromosome 21. Babies with Down syndrome have mental retardation and unusual physical features common in children with Down syndrome. Babies with Down syndrome also may have problems with their heart and other organs.

What is Trisomy 18?

Trisomy 18 is a genetic problem caused by an extra chromosome 18. Trisomy 18 is less common than Down syndrome, but more serious. Babies with trisomy 18 have severe birth defects and usually do not live very long after birth.

How is FIRST screening done?

To start, you will meet with a genetic counselor to answer questions about the test. Then, if you decide the test will be helpful for you, a sample of your blood is taken and an ultrasound of the baby is done. The results of these two tests are combined with your age to see if you are at risk for having a baby with Down syndrome or trisomy 18. It takes about 5 days to get the results of the test. Your genetic counselor will call you with your result.

Why is an ultrasound done?

To be accurate, the test must be done at a certain time in pregnancy. The ultrasound measures the baby to make sure that the pregnancy is between 11 and 14 weeks. Fluid found at the back of the baby's neck is also measured. This is called the nuchal translucency (NT). A large amount of fluid can mean a higher risk for Down syndrome, trisomy 18, heart defects, and other problems.

Why is a blood test done?

The blood test measures proteins in the mother's blood which are made by the growing pregnancy. If levels of these proteins are different than the average pregnancy, it can mean an increased risk for Down syndrome or trisomy 18.

What if my test results are screen negative?

A screen negative result means that your risk for having a baby with Down syndrome or trisomy 18 is not higher than a 35 year old woman's risk.

What if my test results are screen positive?

A screen positive result means that there is a higher risk for having a baby with Down syndrome or trisomy 18, but it does not mean your baby has either of these problems. If your test results are screen positive, you will be offered another test to look for these genetic problems. This test can be done from a sample of the placenta (CVS) or from the fluid around your baby (amniocentesis).

What are CVS and amniocentesis?

CVS (chorionic villus sampling) and amniocentesis are tests which look directly at the baby's cells. CVS is done between 10-14 weeks of pregnancy and amniocentesis is done after 15 weeks of pregnancy. These tests are used to diagnose genetic problems. Both of these tests have a risk for pregnancy complications or miscarriage. This risk for CVS is about 1 in 200 pregnancies (0.5%). The risk for amniocentesis is about 1/300-1/500 pregnancies (0.2-0.3%).

How accurate is FIRST?

FIRST identifies 85% of pregnancies with Down syndrome and over 90% of pregnancies with trisomy 18. Since FIRST is a screening test, it does not give final answers. It will let you know if you are more or less likely to have a baby with one of these problems. The only way to know for sure if your baby is affected is to do a test, such as a CVS or amniocentesis that will diagnose the problem specifically.

Is FIRST covered by insurance?

FIRST is covered by many insurance companies. If you want the test, we suggest you contact your insurance company to discuss if it will be covered. If it is not covered by your insurance company, you will be expected to pay for the test. Financial counselors are available to discuss payment plans.

How do I make sure I have the FIRST testing?

If you decide you want the FIRST test, tell your healthcare provider or contact the UNC Prenatal Diagnosis Program at (919)966-2229. Remember, FIRST testing must be done early in pregnancy so if you are interested it is important to let your healthcare provider know as soon as possible.

Will I be offered other screening tests in my pregnancy?

If you choose to have FIRST, you will be offered another screening test around 18 weeks of pregnancy to see if your baby is at increased risk for spina bifida or another type of neural tube defect (ONTD). Neural tube defects result in an opening in the baby's spine or skull. This testing is sometimes done on a sample of the mother's blood. The blood test is called AFP, or alpha-fetoprotein. This testing can also be done by a targeted ultrasound.

If you do not have FIRST, you will be offered a different screening test around 18 weeks of pregnancy that screens for spina bifida, trisomy 18 and Down syndrome. This test is called the triple screen or quad screen. It is also done on a sample of the mother's blood. If you have FIRST, you will not need a triple or quad screen, because you will have already had screening for Down syndrome and trisomy 18.