



**ADAMS
PATTERSON**
GYNECOLOGY
& OBSTETRICS
WOMEN'S CARE CENTER OF
MEMPHIS, M PLLC

FIRST SCREEN

First Trimester Screening for Down Syndrome & Trisomy 18

First screen is a blood test which shows if you are at increased risk of having a baby with Down syndrome or trisomy 18. It requires a sample of your blood and a special ultrasound measurement performed in the first trimester.

Your doctor wants you to know:

- This is an elective test.
- This screening does NOT evaluate for open neural tube defects.
- This screening does NOT evaluate for gene defects such as cystic fibrosis, sickle cell anemia, Tay-Sachs disease, thalassemia, familial dysautonomia or Canavan disease.
- If you are diabetic or have a multiple gestation and desire this screening you will be referred to a maternal fetal medicine specialist for evaluation.

What is Down syndrome?

Down syndrome is caused by an extra chromosome (#21) and results in both mental and physical abnormalities. Approximately 1 in 800 babies is born with Down syndrome, the most common cause of mental retardation. The risk of having a child with Down syndrome gradually increases with the age of the mother, but can occur at any age.

What is trisomy 18?

Trisomy 18 is caused by an extra chromosome (#18) and results in serious mental retardation and physical deformities, including major heart defects. Only 1 out of 10 babies affected with trisomy 18 lives past the first year of life. As with Down syndrome, the risk of having an affected child gradually increases with the age of the mother.

Each patient's specific result is affected by:

- Exactly how far along in your pregnancy you are on the day the blood sample and ultrasound are performed.
- Your weight, ethnic background and age.
- Whether a close relative has Down syndrome.

It is important to understand that a screening test does NOT provide a diagnosis; rather it predicts the likelihood of a problem to occur. First Screen can only tell you if there is a greater chance of your baby having Down syndrome or trisomy 18.

If your screening test is negative, the risk of you having a baby with Down syndrome or trisomy 18 is low. In rare instances, these birth defects will not be detected through screening, as it cannot detect all high-risk pregnancies.

If your screening test is positive, it does not necessarily mean that the baby has one of these birth defects. In fact, most women who have abnormal screening results will have normal babies. It does mean that your doctor will recommend diagnostic testing to determine if your baby is affected with one of these birth defects. Your doctor will recommend one of the following:

- **Chorionic villi sampling (CVS)**, which is the withdrawal of a small amount of chorionic tissue (tissue from the developing placenta), is performed between 10-12 weeks of pregnancy. Chromosome analysis will be done on this sample. This will be scheduled to be performed by another physician who specializes with this type of testing. CVS is associated with a small risk of miscarriage (about 1%).
- **Amniocentesis**, which is the withdrawal of a small amount of amniotic fluid from the uterus. The fluid can be tested for chromosome abnormalities, and also for open neural tube defects. Typically an amniocentesis is performed around the 16th week of pregnancy. This procedure is performed in our office by one of our physicians and the fluid is sent to a genetics laboratory for analysis. Complications from amniocentesis are uncommon but there is a slight chance of miscarriage (less than 1%).

Synopsis:

- First Screen provides you with information early in your pregnancy about your risk for having a baby with Down syndrome or trisomy 18.
- This test is performed on a blood sample that is taken between the 11th and 12th week of your pregnancy and sent for analysis.
- The test also includes the results of a specific ultrasound measurement of your baby that is taken on the same day as the blood sample.
- First trimester screening leads to the detection of approximately:
 - 83% of Down syndrome cases
 - 80% of trisomy 18 cases