

OPTIONS FOR GENETIC TESTING

A variety of tests are available to detect chromosomal abnormalities in your fetus.

FIRST TRIMESTER SCREENING:

All patients tested in the 1st trimester will receive an AFP only blood test at 16 weeks, which screens for spina bifida with a 95% accuracy rate. Patients who have abnormal 1st trimester testing may have to wait until 16 weeks to get a final diagnosis

Nuchal Lucency and Sequesntial Screening: This is a non-invasive test that includes an ultrasound and blood testing to tell you if your fetus is at a higher risk of Down's syndrome. This test also can tell you if your fetus is at higher risk for heart defects. The screening is 90% accurate.

Chorionic villus sampling: This is an invasive test that actually tells you the chromosomal makeup of your baby. A perinatologist does the procedure and the placenta is sampled. This carries a 1% risk of miscarriage.

Cystic Fibrosis: This is a genetic disease causing pulmonary and digestive disorders. These children are often very sick with many hospitalizations and a shortened life expectancy. You may now get a blood test to see if you are a carrier. There is a 1 in 22 carrier incidence and the test is offered to all our patients

Spinal Muscular Atrophy: This is a group of inherited muscle diseases that cause progressive muscle degeneration and weakness. Approximately 4 out of every 100,000 have the condition.

Fragile X syndrome: A genetic condition that causes a range of developmental problems including learning disabilities and cognitive impairment. Usually, males are more severely affected by this disorder. Approximately 1 in 4,000 males and 1 in 8,000 females are affected

SECOND TRIMESTER SCREENING:

Quad-AFP: this is a non-invasive blood test that tells you if your baby is at higher risk for Down's syndrome, trisomy 18 (a specific genetic disorder) or spinal cord abnormalities. This screening is 75% accurate and false positive results can occur.

Amniocentesis: This is an invasive test done by your physician who samples the amniotic fluid. This test is 100% accurate in determining if your baby has a chromosomal abnormality. This carries a 0.5% risk of miscarriage.

Level II Ultrasound: This is a comprehensive ultrasound performed by a perinatologist at 20-22 weeks. This screening looks for any congenital anomalies.