

OPTIONS FOR GENETIC TESTING

As a woman ages, her eggs age. As a result, the risk of chromosomal abnormalities increases. This means that the DNA make-up of the baby could be abnormal. There are now tests available to see if your baby is at higher risk for these problems.

FIRST TRIMESTER SCREENING:

BUN STUDY: This is a non-invasive test that includes an ultrasound and blood testing to tell you if your fetus has a higher risk of DOWN syndrome. This test also can tell you if your fetus is at higher risk for heart defects. The screening is 90% accurate. It is still considered investigational.

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.

SECOND TRIMESTER SCREENING:

Quad-AFP: this is a non-invasive blood test that tells you if your baby is at higher risk for DOWN 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.

Genetic Ultrasound: This a targeted ultrasound performed at 18 weeks that looks for specific abnormalities associated with chromosomal abnormalities like DOWN syndrome.

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