

Optional Testing

You will have the decision to test for the potential of fetal genetic conditions. If you are interested in any optional tests, please check with your insurance plan to see if these tests are covered. High risk or abnormal results may require additional testing.

Questions you may have regarding these optional tests are usually discussed at your first appointment.

Carrier Screening – This blood test performed in our office will determine if you carry a gene that could cause a genetic condition in your child. Further testing is always required to confirm whether or not there is a problem. Carrier status for Cystic Fibrosis (CF), Spinal Muscular Atrophy (SMA), Duchenne Muscular Dystrophy (DMD), and Fragile X are commonly tested.

Cell Free Fetal DNA – This blood test is performed in our office after 8 weeks gestation to screen for whether your pregnancy is at high or low risk for Down syndrome (Trisomy 21), Trisomy 13 and Trisomy 18. If desired, this test can also determine the sex of the baby.

Early screen/nuchal translucency – This ultrasound and blood test is performed between 11-13 weeks gestation at a high risk obstetric specialist's office. The test determines whether your pregnancy is at high or low risk for Down Syndrome (Trisomy 21), Trisomy 13 and Trisomy 18.

AFP-4 (Quad screen) – This blood test is performed between 15-20 weeks gestation at our office. The test determines whether your pregnancy is at high or low risk for Down Syndrome (Trisomy 21), Trisomy 18 and birth defects of the spinal cord and skull.

Amniocentesis – This procedure is performed by a high-risk OB specialist after 16 weeks gestation. The test can determine the presence of abnormal genes or chromosomes associated with Down Syndrome or other genetic conditions. Under ultrasound guidance, a needle is inserted through the mother's abdomen into the amniotic sac, and a small sample of amniotic fluid is removed for genetic testing. This test is typically only recommended in special high risk situations.