Genetic Screening

All of our patients are offered some form of screening for Down Syndrome, Trisomy 13, and Trisomy 18. This can be accomplished by drawing your blood either alone or in combination with specific ultrasound measurements.

First Trimester
You may have heard of first trimester screen, nuchal translucency, cell-free fetal DNA, (including Counsyl, Harmony, Panorama, and others). With all the different genetic screening options, it can be confusing to understand which one is appropriate for you. The main differences between them are summarized below:

Test	Information	Cost
Nuchal translucency (NT)	Between 12-13 weeks of pregnancy, the thickness of the skin on the back of the baby's neck is measured. If the NT is 3mm or greater, more testing will be discussed. 64-70% detection rate for Down Syndrome. Carries a 5% false positive rate.	Covered by most insurances.
First trimester screen: bloodwork **and** nuchal translucency (NT)	Maternal blood draw **combined with** nuchal translucency. 82-87% detection rate for Down Syndrome. Carries a 5% false positive rate.	Covered by most insurances.
Cell-free fetal DNA (cfDNA), also called non-invasive prenatal testing (NIPT)	Maternal blood draw after 9-10 weeks (depending on the test) 99% detection rate for Down Syndrome.	Difficult to quote the cost, as it may depend on insurance, and each company offers different "Self-Pay" rates, which may change.

*"Advanced maternal age" (AMA) is defined as being 35 or older by the estimated date of delivery (your due date). At age 35, the risk of having a child with Down Syndrome, Trisomy 18, or Trisomy 13 is 1:200. For this reason, sometimes the cell-free fetal DNA tests are covered by insurance after age 35. However, we do recommend checking with your insurance regarding your individual policy.

Second Trimester
Regardless of the results of your first trimester testing, there are two tests we offer to all patients in the second trimester:

1. 20 week ultrasound to visualize fetal anatomy
2. AFP (alpha-fetoprotein): This is a maternal blood test drawn between 15-20 weeks which will help screen for neural tube defects such as anencephaly and spina bifida.

If you did not undergo first trimester screening, the Quad Screen can be performed between 15-20 weeks as well. The Quad Screen is a measure of 4 hormones in the maternal blood that can detect >80% of pregnancies with Down's Syndrome or neural tube defects, with a 5% false positive rate.

Specialized Invasive Testing
It is important to remember that a screening test can only tell you the RISK of having a baby with Trisomy 21 (Down's Syndrome), Trisomy 13, Trisomy 18, or other chromosomal disorders. For women or couples who either have an increased risk based on screening tests or for families that want a definitive diagnosis rather than a screening test, chorionic villi sampling (CVS) can be performed between 10-12 weeks or amniocentesis can be performed between 15-18 weeks. Chorionic villi sampling involves inserting a needle either through the abdomen or cervix to sample and analyze the placental tissue; amniocentesis involves inserting a needle into the abdomen to sample and analyze the amniotic fluid. These tests are usually performed at a high risk/perinatal obstetrics practice. You may also be referred to see a perinatologist for a level II or advanced ultrasound, which is a more detailed ultrasound performed by a high risk specialist.

Carrier Screening for Specific Genetic Conditions

You and your partner may also elect to be screened for specific genetic conditions. Ideally, this would be performed prior to pregnancy, although it can be performed during pregnancy as well. Screening for these specific conditions often is determined by an individual's ancestry, including family history of any of the below conditions, please discuss with your provider.

Heritage or History	Offered Screening
All patients	Cystic fibrosis, spinal muscle atrophy
African Mediterranean Middle Eastern Southeast Asian West Indian	Sickle cell anemia Thalassemia Other hemoglobinopathies
Ashkenazi Jewish heritage	Tay Sach's Canavan's Gaucher Syndrome Cystic Fibrosis
Cajun or French Canadian	Tay Sach's
Women with family history of mental retardation or premature ovarian failure	Fragile X