Genetic Screening

Aneuploidy is the term used to describe having an abnormal number of chromosomes. When it comes to evaluating your risk of having a child with aneuploidy, your options include:

- No screening,
- Non-invasive screening which involves maternal-fetal medicine consultation for bloodwork and ultrasound, or
- If necessary, diagnostic testing performed by our maternal-fetal medicine partners.

As part of your genetic counseling, we will discuss all of your options within your first few appointments so you feel equipped to make the best choice for yourself. We support our patients in any options that they choose.

First Trimester

All pregnant women 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. In terms of non-invasive testing, all patients will have a late first trimester ultrasound between 10-14 weeks. The first trimester ultrasound cannot detect all abnormalities but can screen for major structural anomalies.

Coupled with the first trimester ultrasound, we offer bloodwork as well. You may have heard of the first trimester screen, nuchal translucency, cell-free fetal DNA, or NIPT testing (which has many brand names). 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
First trimester screen and nuchal translucency	• Maternal blood draw and ultrasound between 12-13 weeks • 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 10 weeks (depending on the test) • 99% detection rate for Down Syndrome	• Cost will be dependent on insurance* • This is a test that we refer out to our maternal-fetal medicine partners.

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. AFP (alpha-fetoprotein): A maternal blood test drawn between 15-20 weeks which will help screen for neural tube defects such as anencephaly and spina bifida
2. 18-22 week ultrasound to visualize fetal anatomy

Diagnostic Testing

While screening tests can tell you if you are at risk, we use diagnostic tests to confirm the abnormality. There are two commonly used diagnostic tests: Chorionic villi sampling (CVS) and Amniocentesis. If after discussion with your provider we determine these tests may apply to you, we will refer you to a high-risk specialist who performs this testing.

Carrier Screening for Specific Genetic Conditions

We offer expanded screening to test if you or your partner(s) are carriers for a genetic disorder that you may pass to your baby. For example, we can test for cystic fibrosis, spinal muscle atrophy, fragile X, or sickle cell disease.

Many patients are unsure of their ethnicity or family history, therefore, we recommend genetic screening of the diseases listed below.

Heritage or History	Recommended Carrier Screening
All patients, regardless of race or ethnicity	• Cystic fibrosis and spinal muscle atrophy
African, Mediterranean, Middle Eastern, Southeast Asian, West Indian	• Sickle cell disease, thalassemia, and other hemoglobinopathies
Ashkenazi Jewish	• Tay–Sachs disease, Canavan disease, and Gaucher disease
Cajun, French Canadian	• Tay–Sachs disease
Women with a family history of mental retardation or premature ovarian failure	• Fragile X

This is a test that we refer out to our maternal-fetal medicine partners.