Genetic Testing

There are several options available for pregnant women regarding genetic testing. Some methods require a simple blood test, while others are more invasive and associated with some risk. Genetic testing cannot detect the majority of birth defects that could affect a fetus, and no test can guarantee a perfect baby.

The following is a brief summary of what testing is available along with some of the advantages and disadvantages. These tests are all considered optional; however, approximately 90% of patients pursue testing. Keep in mind, there are ***noninvasive*** tests which are only screening tests and ***invasive*** tests which determine a diagnosis.

1st Trimester

Currently, the standard of prenatal care is to offer second trimester testing. The American College of Ob/Gyn feels first trimester screening for Down syndrome is an option. Until recently, no testing was available to detect chromosomal abnormalities like Down syndrome in the first trimester that was not invasive. Currently, there are three options.

1) First Trimester Screen: this *non*invasive test combines an ultrasound marker known as a Nuchal Translucency (NT) and blood work to calculate the risk of having a baby with Down syndrome (DS) or Trisomy 18 (an often fatal chromosomal abnormality). This test will detect approximately 83% of babies with DS. However, this test gives no information about neural tube defects (NTDs). (A NTD is a problem with the spinal cord. An example of a common NTD is spina bifida).

The advantage of the First Trimester Screen is that it is noninvasive and offers information about DS and Trisomy 18 as early as 12 to 14 weeks. Because it is not as accurate at detecting chromosomal problems as the **Integrated Screen**, this screen would be recommended to patients who want the earliest possible information to consider a Chorionic Villus Sampling.

2) Chorionic Villus Sampling (CVS): this is an invasive test requiring sampling of placental tissue from within the uterus. Generally, this is reserved for patients at high risk for chromosomal abnormalities or those with an abnormal First Trimester Screen. Risks associated with the procedure include the possibility of miscarriage and rarely, limb defects. Patients who desire this form of testing are referred to a specialist trained in the technique.

3) Integrated Screen: This noninvasive test combines the Nuchal Translucency (NT) at 12 - 14 weeks with two blood samples obtained at 12-14 weeks and 16-20 weeks. This test has a 94-96% detection rate for DS, 80% for NTD's, and 90% for Trisomy 18. The false positive rate is 5% (this implies that 5% of normal fetuses will have an abnormal screen).

For our low risk patients, we recommend the Integrated Screen as it has the highest detection rate for chromosome abnormalities and is noninvasive.

4) NIPD - Noninvasive Prenatal Diagnosis: The MaterniT21 PLUS laboratory developed test is a noninvasive blood test that is available for women with increased risk indicators for fetal chromosome variations at 10 weeks of pregnancy and beyond. This test detects an increased amount of chromosomal 21, 18, 13 material that is circulating in maternal blood. The increased risk factors include one or more of the following: Advanced maternal age, Positive serum screening test, Fetal Ultrasound abnormality suggestive aneuploidy, and/or Personal/family history of chromosomal abnormalities.

2nd Trimester

The following tests are routinely offered in the second trimester:

1) Quad Screen: This is a *non*invasive blood test offered between 16 and 20 weeks which screens for DS, NTDs, and trisomy 18. The test will detect approximately 80% of babies with DS and 80-90% of babies with NTDs. The detection rate is lower than the **Integrated Screen** and therefore is recommended for patients who present for care in the second trimester.

2) Amniocentesis: This is an invasive test that involves placing a needle in the uterus and withdrawing amniotic fluid for analysis. The procedure is routinely offered to women over 35 who are at an increased risk for genetic abnormalities, such as DS. It is also offered when a woman has an abnormal genetic screening test or abnormal ultrasound. The advantage of this test is that it gives definitive information regarding genetic abnormalities with a detection rate of nearly 100%. The risk associated with this procedure is the possibility of miscarriage, which occurs in approximately 1/250 amniocentesis procedures.

We recommend this invasive test for patients of moderate risk who want a diagnosis rather than a screening test and who are aware of the risk of miscarriage.

3) Ultrasound: This is a *non*invasive test routinely performed at 20 weeks which allows visualization of the baby. Ultrasound can detect many fetal abnormalities including those associated with DS and NTDs. However, the detection rate is still approximately 80% when used in combination with the Quad Screen.