



Genetic Testing Options

- When you meet with the doctor or genetic counselor, you may be offered different tests. These include ***prenatal screening***, ***prenatal diagnosis***, and/or ***genetic carrier testing***.
- Having these tests performed on you is ***optional***. They are ***not required*** for you to deliver your baby. However, if you are interested in additional information about your pregnancy, you may want to think about having some of these tests done.

PRENATAL SCREENING

- Prenatal screening checks your pregnancy for certain chromosomal disorders, including Down syndrome and other conditions like Trisomy 18, Trisomy 13, and spina bifida.
- The purpose of screening is to put your pregnancy into a ***risk group*** (either ***high risk*** or ***low risk***).
- There are times when someone's results put them in the "wrong group". For example, someone may be classified as high risk for a certain disorder, but the baby does ***NOT*** have the disorder. Or, the pregnancy is classified as being at low risk for a specific condition, when that condition is ***actually present***. The chance of this happening depends on what screening test you choose.
- A negative test result does not guarantee the birth of a healthy baby, and a positive test result does not guarantee the baby has that specific condition.
- ***Screening does not put the pregnancy at risk for miscarriage.*** It is non-invasive. It is a blood test for the patient. Level 2 ultrasound (sonogram) is also a screening tool that does not put the pregnancy at an increased risk for miscarriage.

PRENATAL DIAGNOSIS

- Diagnostic testing is able to diagnose genetic and chromosomal disorders with 99.9% accuracy.
- These tests are invasive, meaning they put the pregnancy at a ***small risk*** for miscarriage, depending on the procedure, gestation, etc.
- These are ***NOT*** blood tests. They usually consist of a CVS (chorionic villus sampling) or amniocentesis.
- Diagnostic testing includes the most accurate tests available prior to delivery.
- The purpose of testing is for ***information only***; not for prenatal cure, therapy, or treatment. However, someone may choose to deliver in a high-risk center, where there are advanced medical staff and equipment in case there is a complication at delivery, or shortly after.
- The American Congress of Obstetricians and Gynecologists (ACOG) recommends all pregnant women be ***offered*** prenatal diagnosis (ACOG Practice Bulletin 88, Dec. 2007).

GENETIC CARRIER TESTING

- These are blood tests that **do not** put the pregnancy at a risk for miscarriage.
- These are diseases where mom and dad can be carriers and **have no symptoms**, even if nobody in the family has these diseases.
- If mom and dad are carriers of the same genetic disease, they have a 25% risk of having a child with that disease.
- Specific diseases offered for testing for will vary depending on your interest, family history, and ethnic background (Caucasian, African American, Hispanic, Asian, etc.).
- Most of these tests are covered by insurance.

I, (print name) have read the information above regarding prenatal screening, prenatal testing, and genetic carrier testing. If I have questions, I will discuss them with the genetic counselor or doctor. THIS IS NOT A CONSENT FORM. SIGNING IT DOES NOT COMMIT YOU TO ACCEPT OR DECLINE ANY TESTS.

Signature: _____ Date: _____