Congratulations on your pregnancy! We look forward to taking care of you during this exciting time. At your first maternity visit, several options for prenatal genetic testing were reviewed for you to consider. Here is a summary, as it can be confusing to understand all of the different options, what is best for you and how to schedule the tests.

SCREENING TESTS

These three tests calculate the risk of certain genetic conditions for your pregnancy. As the testing is done by bloodwork and ultrasound, they do not pose a risk of miscarriage to the pregnancy. Only one option is chosen.

SEQUENTIAL SCREEN

This test measures hormone levels in your blood twice, first at 10 - 13 weeks and second at 16 - 18 weeks. The first blood draw is done in conjunction with an ultrasound. The results of these tests are then analyzed to calculate the risk of Trisomy 18, Trisomy 21 (Down Syndrome) and Neural Tube Defects (Spina Bifida) for the pregnancy.

Insurance does not always cover this screening, so we ask that you confirm coverage. You will need to call our office to schedule if you decide to pursue this testing.

NON-INVASIVE PRENATAL TESTING (NIPT)

This test checks the DNA of the fetus via a single blood test drawn after 10 weeks of pregnancy in women 35 years or older. The results will figure out the risk of Trisomy 13, Trisomy 18 and Trisomy 21 (Down Syndrome). The gender of the fetus can also be determined, if desired. NIPT can also be utilized as follow up to previously abnormal test results from either the Sequential Screen or the Quad Screen.

Insurance does not always cover this screening, so we ask that you confirm coverage.

QUAD SCREEN

This test measures four hormone levels in the mother's blood at 16 - 19 weeks pregnancy. The results of these tests are then analyzed to calculate the risk of Trisomy 18, Trisomy 21 (Down Syndrome) and Neural Tube Defects (Spina Bifida) for the pregnancy. This test has been available for many years and is usually covered by insurance. It is less accurate than the sequential screen and NIPT, but is a perfectly acceptable alternative if you don't have insurance coverage for the other options.

DIAGNOSTIC TESTS

These tests will diagnose chromosomal abnormalities. They are invasive tests, in which fluid or tissue is removed from the pregnancy. There is a risk of miscarriage as a result of these tests.

AMNIOCENTESIS

This test evaluates the fetal chromosomes after a small amount of amniotic fluid is removed using a sterile needle from the amniotic sac after 16 weeks. The results are diagnostic, meaning that they diagnose or confirm a genetic problem. It is now usually performed if there is a specific genetic history or an abnormal screening test result. It also is a test that poses a risk of miscarriage because of the invasive nature of how the amniotic fluid is removed.

CHORIONIC VILLUS SAMPLING

This test evaluates fetal chromosomes by performing a biopsy of the placenta using a sterile needle between 10 – 13 weeks. The results are diagnostic, meaning that they diagnose or confirm a genetic problem. It is performed earlier than an amniocentesis, and is usually considered if there is a family history of a specific genetic condition. It is also a test that poses a risk of miscarriage because of the invasive nature of the placental sampling.

Please consider all of these options carefully. Some of these tests may need to be scheduled before your next appointment. You may certainly choose to decline all of the testing options.

If you have any questions, feel free to call the office during regular business hours to discuss with either a nurse, your midwife or your physician.