NORTHLAND WOMEN'S HEALTH CARE, P.C.
OBSTETRICS / GYNECOLOGY

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Screening Options in Pregnancy

Genetic (Aneuploidy) Screening Options:

The following are tests available to detect certain problems that may affect your pregnancy. These are all **screening** tests only and if abnormal should be followed up with a **diagnostic** test (for example an amniocentesis) to confirm the results.

Theses are all elective procedures and may not be covered by insurance.

First Trimester screen:
- This test includes a blood test and an ultrasound of the nuchal fold, an area at the back of the baby's neck. This screening looks for an increased risk of chromosomal disorders. This test will be done by a High Risk OB specialist (Maternal Fetal Medicine) but you do not have to be "High Risk" to have this test done.
- Detection rate for Downs Syndrome is 96%.

Second Trimester (Quad) Screen:
- This is a blood test offered between 15-20 weeks.
- This is a screening test for the increased risk of chromosomal disorders (for example Down syndrome and other trisomies) and neural tube defects (i.e. anencephaly and spina bifida).
Detection rate for Downs Syndrome is 81%.

Cell-Free DNA Testing:
- This is a blood test done any time after 10 weeks.
- This test assesses cell-free fetal DNA that is found in the mother's blood. This screens for some chromosomal disorders including Down Syndrome (Trisomy 21), Edwards Syndrome (Trisomy 18) and Patau Syndrome (Trisomy 13). It also screens for sex chromosome disorders including Turner Syndrome (XO), Klinefelter Syndrome (XXY), Jacobs Syndrome (XYY) and Triple X Syndrome.
- It does not screen for all birth defects or neural tube defects.
- Detection rate for Downs Syndrome is 99%
- There is the risk for false negative and false positive results. There is a higher risk for false positive results in low risk women (age less than 35).
    - *Therefore, we DO NOT recommend this as a primary screening test if your age is less than 35.*

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Hereditary Disease Screening Options:

The following are tests to see if you are a carrier for a hereditary disease. Theses tests only need to be done once in your life because your carrier status will not change over time. This testing can be done prior to or during pregnancy.

Theses are all elective tests and may not be covered by insurance.

Cystic Fibrosis:
This is the most common life threatening autosomal recessive condition in Caucasian populations. Approximately 1/25 non-Hispanic whites are a carrier of cystic fibrosis. Typically, both parents would have to be a carrier for the baby to become affected with the disease. It is recommended that all patients, regardless of ethnicity, be offered this test.

Spinal Muscular Atrophy:
This is an autosomal recessive disease that affects 1 in 10,000 births. Approximately 1/40 to 1/60 people are a carrier for the disease. There is no treatment available and these children often have very serious, sometimes fatal, developmental and physical problems. It is recommended that all patients, regardless of ethnicity, be offered this test.

Fragile X Syndrome:
This is the most common inherited form of mental retardation. Approximately 1 in 3,600 males are affected and 1 in 4,000-6,000 females are affected by the disease. Currently it is recommended for patients to be screened if they have a family history of Fragile X, undiagnosed mental retardation, autism or developmental delays.

We offer a **Trio Panel** that includes testing for all 3 of the above diseases.

There are also screening panels that test for 200+ hereditary diseases. If you have other hereditary diseases in your family please discuss this option further with your doctor.