North Clinic
~Where caring is a tradition~

Prenatal Testing: Carrier Screening

Carrier Screening:

A "carrier" is someone with mild or no symptoms of a disorder but carries an abnormal gene for a disorder which can be passed on to their child. Carrier screening is performed through a blood test. If you are found to be a carrier, your partner is then screened for the disorder. If you are not a carrier, no additional testing is done. Carrier screening can be done before or during pregnancy. People of certain ethnic groups are more likely to be carriers of different diseases; below you will find information on the most commonly screened for disorders. All carrier screening has limitations; if you decide to pursue carrier screening you will meet with a genetic counselor to learn more about these limitations. Results are usually available within 2-3 weeks.

Cystic Fibrosis

Cystic Fibrosis (CF) is a progressive disease that primarily affects the lungs, pancreas and gastrointestinal systems; the average life span of an individual with CF is 37 years. CF is more common in the non-Hispanic white population. One in 25 non-Hispanic white individuals (without a family history of CF) are carriers of an abnormal gene for Cystic Fibrosis. For people of Ashkenazi Jewish descent the carrier rate is 1 in 24; Hispanic white 1 in 58; African American 1 in 61; Asian American 1 in 94. It is becoming increasingly difficult to assign a single ethnicity to individuals; therefore, all women of reproductive age are offered CF screening.

Fragile X Syndrome

Fragile X syndrome is the most common inherited form of mental retardation. Mental retardation or impairment can range from mild to severe and is the most common known cause of autism. 1 in 250 women without a family history of mental retardation are carriers; the carrier frequency is 1 in 85 for women with a family history of mental retardation. Carrier screening and genetic counseling is offered to all women with a family history of fragile X-related disorders, unexplained mental retardation or developmental delay, autism or premature ovarian insufficiency.

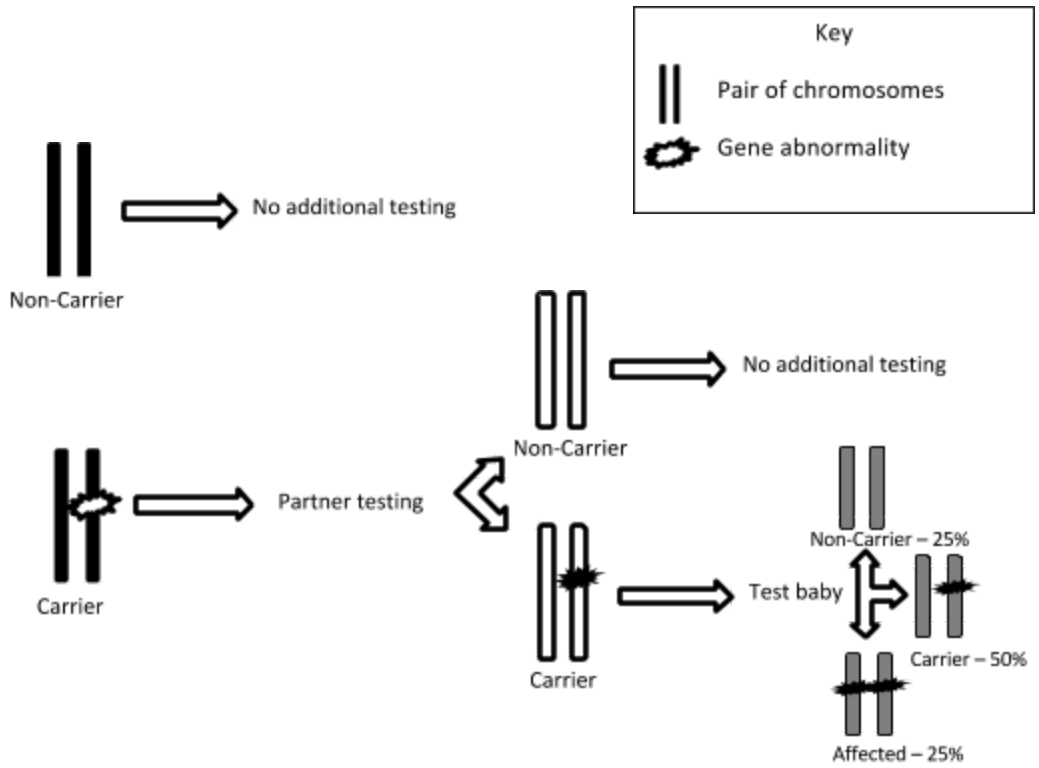
Spinal Muscular Atrophy:

Spinal Muscular Atrophy (SMA) is a disease that leads to wasting of muscles and overall weakness. It is estimated that 1 in 40 to 1 in 60 people are carriers for this disease. Patients with a family history of SMA are offered carrier screening.

Tay-Sachs Disease:

Tay-Sachs disease is a progressive neurologic disease that causes death in early childhood. 1 in 30 people of Ashkenazi Jewish descent is a carrier while the carrier frequency for non-Jewish individuals is 1 in 300. Persons of Cajun or French-Canadian descent have a higher carrier frequency than the general population; therefore, carrier testing is offered to people of Ashkenazi Jewish, Cajun or French-Canadian descent.

Hemaglobinopathies:
Hemaglobinopathies (Sickle Cell Disease and thalassemia) are caused by abnormalities in hemoglobin (the oxygen carrying component in red blood cells) and can lead to disruption of blood supply to vital organs. One in 12 African Americans carry an abnormal hemoglobin. Other populations with high frequencies of abnormal hemoglobin are: Greeks, Sicilians, Turks, Arabs, Southern Iranians and Asian Indians. A hemoglobin electrophoresis (a simple blood test) may be ordered to help diagnose diseases, called hemoglobinopathies, involving abnormal hemoglobin production, such as sickle cell disease and thalassemia.



Insurance Codes:

Please ask your insurance company if these tests are covered prior to moving forward with testing:

- Cystic Fibrosis Carrier Screening: 81220 (North Clinic Fee: $900.00)
- Fragile X Carrier Screening: 81243 (LabCorp Fee: $391.00)
- Spinal Muscular Atrophy Carrier Screening: 81401 (North Clinic Fee: $408.00)
- Tay Sachs Carrier Screening: 81255 (LabCorp Fee: $440.00)
- Hemoglobin Electrophoresis: 83021, 85660 (LabCorp Fee: $200.00)

-With reflex for alpha-thalassemia: 81257, 82728, 83021, 85027, 85660 (LabCorp Fee: $306.00)