Prenatal Testing: Carrier Screening

Carrier Screening:

What is carrier screening?

Carrier screening is a type of genetic test that can tell you whether you carry a gene for certain genetic disorders. As a carrier, you may have mild or no symptoms of the disorder, but can pass the gene associated with a disorder onto your child. When this screening is done before or during pregnancy, it allows you to find out your chances of having a child with a genetic disorder. It involves testing a sample of blood, saliva or tissue from the inside of the cheek and can be done before or during pregnancy.

Who should have carrier screening?

All women who are thinking about getting pregnant or who are already pregnant are offered carrier screening. Screening is available for a limited number of diseases, including those listed below. You can have additional screening for other disorders as well.

People of certain ethnic groups are more likely to be carriers of different diseases. All carrier screening has limitations. If you decide to pursue carrier screening you will be referred to a genetic counselor to learn more about disorders for which screening is available, limitations of this screening, and estimated cost. Results are usually available within 2-3 weeks.

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.

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 commonly 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:

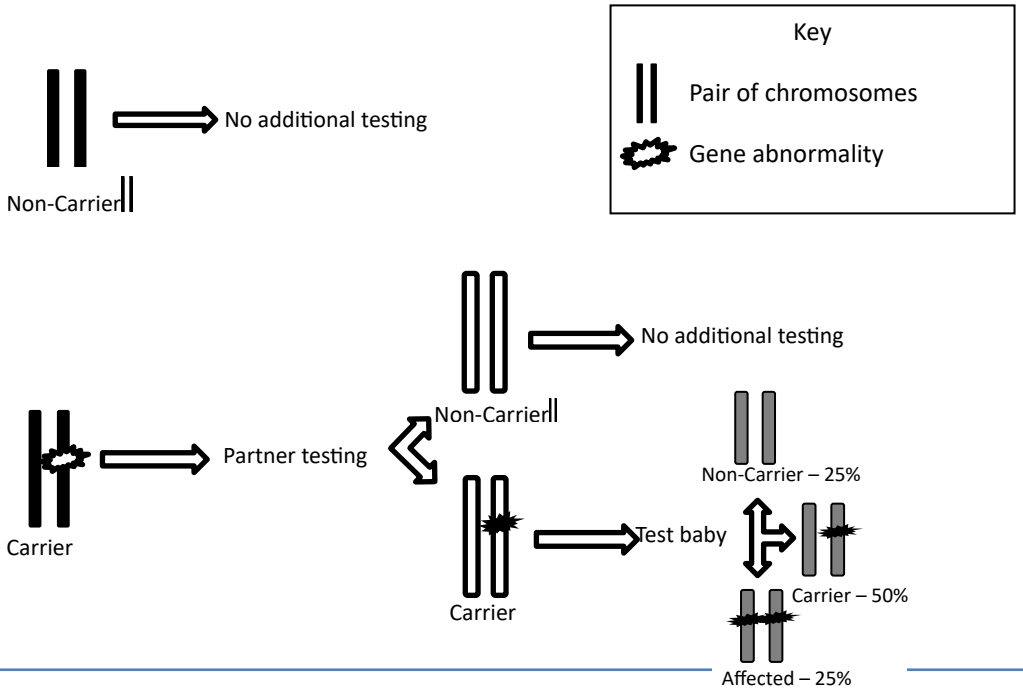
Spinal Muscular Atrophy (SMA) is a disease that leads to wasting of muscles and overall weakness. It is estimated that 1 in 40 to 1 in 60 people are carriers for this disease. Patients with a family history of SMA are offered carrier screening.

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.

Hemoglobinopathies:
Hemoglobinopathies (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 blood test) may be ordered to help diagnose these diseases.



Insurance Codes:

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

- Cystic Fibrosis Carrier Screening: 81220
- Fragile X Carrier Screening: 81243
- Spinal Muscular Atrophy Carrier Screening: 81401
- Tay Sachs Carrier Screening: 81255
- Hemoglobin Electrophoresis: 83021, 85660

-With reflex for alpha-thalassemia: 81257, 82728, 83021, 85027, 85660