What is carrier screening?

Carrier screening is used to assess an individual's, and ultimately a couple's, risk of having a child with a serious genetic condition. Both the American Congress of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) publish recommendations regarding carrier screening, and recommend that patients who are considering pregnancy be offered carrier screening for conditions such as cystic fibrosis, spinal muscular atrophy, and thalassemia / sickle cell disease. You can also opt to be screened for a larger number of genetic conditions through an expanded carrier panel.

Everyone has some abnormal genes in our genetic make up, usually with no effect on our health. The more genes that are tested the more likely it is that we will find an abnormal gene. Typically both parents must carry an abnormal cope of the gene for the same condition to have an affected child. If one member of the couple tests positive for a condition, the other member of the couple must be tested also before we can proceed with therapy. If both members of the couple are positive further genetic counseling will be offered.

Genetic carrier screening only tests for specific genetic conditions. A normal genetic screen markedly decreases but does not eliminate the chance of the specific condition that is being tested for. Genetic screening does not rule out all birth defect.

The following options are available for carrier screening:

Cystic Fibrosis: Cystic fibrosis (CF) is a medical condition which results in excessive mucus production in the lungs, digestive tract, and pancreas. It is a life-long genetic disorder associated with digestion and breathing problems that may lead to severe infections and infertility in affected males. Some cases are more severe than others. Respiratory failure is the most common cause of death making the average lifespan 37 years.

Spinal Muscular Atrophy: Spinal muscular atrophy (SMA) is a genetic disease that results in progressive muscle weakness and paralysis. There are three types of SMA. The most severe type is usually diagnosed within the first few months of life. Affected children have severe muscle weakness and typically do not survive past the age of 2. The other two types, which are less common than the severe type, involve a smaller degree of muscle weakness. Most affected people need to use wheelchairs or need assistance with walking. Life expectancy for the less severe types ranges from the teenage years to adulthood. Those with the mildest form of SMA are expected to have a normal lifespan.

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Thalassemia and Sickle Cell Disease: Sickle cell disease is an inherited disorder in which red blood cells have a crescent shape, which causes chronic anemia and episodes of pain. Thalassemia is a blood disorder in which individuals have reduced production of hemoglobin, the molecule that carries oxygen in red blood cells. There are different types of thalassemia. In severe forms, infants can be stillborn or die shortly after birth. In less severe forms, individuals may experience anemia, weakness, fatigue, and enlargement of the liver and spleen. Some forms of thalassemia may require regular blood transfusions.

Expanded carrier panel: The expanded carrier panel (Carrier Map) screens for CF, SMA, thalassemia and sickle cell disease, as well as over 300 other genetic conditions in one test. The conditions that are tested for range from minor inconvenience to fatal diseases. Genetic counselling will be offered with any abnormal results. As such a large number of diseases are being screening there is a high likelihood of finding a positive result.

Please note that genetic screening may or may not be covered by your insurance plan.

I acknowledge that I have received information regarding my options for genetic carrier screening and I:

_____ **Decline all genetic screening**

_____ **I wish to consider options: I decline today and will inform the practice if I want testing in the future.**

_____ **I wish carrier testing for:**

____ cystic fibrosis

_____ spinal muscular atrophy

_____thalassemia /sickle cell disease

_____ expanded carrier panel with the Carrier Map Screen

_____ _____

Signature **Date**

_____ _____

Witness **Date**

Follow up_____ **Date:**_____