



## Genetic Blood Tests

### **Cystic Fibrosis**

#### **What is cystic fibrosis?**

Cystic fibrosis (CF) is one of the most common genetic disorders in the Caucasian population, affecting approximately 1 in 3,000 people. The most common problems are chronic lung infection and poor absorption of food due to the accumulation of thick mucus in the lungs and pancreas of patients with CF. While much progress has been made in the understanding and treatment of the disease, there is no cure. At the present time, the median life expectancy is about 30 years.

#### **What causes cystic fibrosis?**

CF is caused by mutations in the CFTR gene. CF is an autosomal recessive disorder. For an individual to be affected with CF, he or she must inherit one copy of the mutated CF gene from each parent. Individuals having one copy of the mutated gene and one copy of the normal gene are known as carriers. Carriers do not have any symptoms of the disorder. The CF carrier frequency differs among different ethnic groups. The frequency is approximately 1 in 25-30 in individuals of Northern European or Ashkenazi Jewish ancestry, 1 in 50 in Hispanics, 1 in 65 in African Americans and 1 in 50 in Asians. When both parents are carriers for a mutation, there is a 1 in 4 chance that each pregnancy will be affected with CF.

#### **How can cystic fibrosis be detected?**

A DNA laboratory test for the mutations causing CF is available. This is a blood test. Results are usually ready within a week. The test can be performed on blood specimens to detect carriers or affected individuals. It can also be performed on prenatal amniotic fluid specimens to detect affected fetuses. Since there are over 900 different mutations within the CF gene, this test cannot detect all the mutations. The detection rate varies among different ethnic groups, with 97% for Ashkenazi Jews, 90% for Caucasians, 68% for Hispanics, 45% for African Americans and 30% for Asians.

#### **Who should be tested for cystic fibrosis?**

CF carrier testing should be considered for individuals with a family history of CF, spouses of CF carriers and pregnant couples who are of Northern European or Ashkenazi Jewish ancestry. Prenatal diagnosis is recommended when both parents have been found to be carriers, there is a family history of CF and one parent is found to be a carrier, a previous child has been diagnosed with CF or certain ultrasound abnormalities are seen in the fetus.

#### **What if the test does not show a CF mutation?**

If your test does not show a mutation in the CFTR gene, the chance that you are a CF carrier is low. That chance will depend on your ethnic background and family history. However, no CF test can find all the mutations of the CFTR gene.

### **What if the test shows a CF mutation?**

If your test shows a mutation in the CFTR gene, then you are a CF carrier. The test has 99% accuracy. Being a CF carrier will not affect your own health. If your test is positive, your partner should then be tested. Special counseling and testing should be considered if both you and your partner are carriers of CF mutation.

## **Ashkenazi Jewish Genetic Screening**

### **What is an Ashkenazi Jewish Disease?**

Ashkenazi is the term used to describe Jewish individuals who have ancestors from Eastern Europe. Roughly 90% of the six million Jewish individuals in the United States are of Ashkenazi descent. Similar to most ethnic populations, the Ashkenazi Jewish population has a higher prevalence of certain genetic disorders. Individuals of Jewish descent should be screened for Tay-Sachs disease, Canavan disease and Gaucher's disease.

### **What is Tay-Sachs disease?**

Tay-Sachs disease is a fatal genetic disorder that occurs more frequently in the Ashkenazi (Eastern European) Jewish population. Approximately 1 in 27 Ashkenazi Jewish individuals are carriers of this disease. A baby with Tay-Sachs disease appears normal at birth, but after six months of age, the child progressively develops mental retardation followed by paralysis, blindness, and seizures. Death usually occurs by the age of five. Tay-Sachs disease is caused by a deficiency of an enzyme called Hex-A. As a result of this deficiency, there is an accumulation of certain substances, which damage the nervous system.

### **What is Canavan Disease?**

Canavan disease is a progressive disorder in which the brain and nervous system degenerate. Symptoms of Canavan disease include brain damage, mental retardation, feeding difficulties, blindness, and a large head. There is no treatment, and death usually occurs in the first decade of life.

### **What is Gaucher's Disease?**

Gaucher's Disease is an inborn error of metabolism that results from a specific malfunction in one of the body's individual chemical processes. Although there are at least 34 mutations known to cause Gaucher's Disease, there are 4 genetic mutations which account for 95% of the Gaucher Disease in the Ashkenazi Jewish population. The carrier rate is 1 in 14 Jewish people of Eastern European ancestry and 1 in 100 of the general population.

### **How are these diseases inherited?**

All three diseases are inherited in an autosomal recessive pattern. For an individual to be affected, he/she must inherit one copy of the abnormal (mutated) gene from each parent. Individuals having one copy of the particular disease-causing gene and one copy of the normal gene are known as carriers. Carriers usually do not have any symptoms of the disorder. If both parents carry the same mutated gene, their child has a 25% chance of having the disease. If only one parent carries the disease gene, their child is not at risk for having that disease but has a 50% chance of being a carrier. If both parents are carriers, the couple should undergo prenatal genetic counseling.

### **How do I get tested?**

A simple blood test can be performed from either parent to determine if he/she is a carrier of these diseases. If both parents are carriers, then prenatal testing can be performed to determine whether or not the fetus is affected.

## **Sickle Cell Anemia**

### **What is sickle cell anemia?**

Sickle cell anemia is an inherited disorder that affects hemoglobin, a protein that enables red blood cells to carry oxygen to all parts of the body. The disorder produces abnormal hemoglobin, which causes the red blood cells to become crescent or sickle shaped. Normal red blood cells are round and move through blood vessels in the body to deliver oxygen. Sickle red blood cells become hard, sticky and have difficulty passing through the small blood vessels. When these hard, pointed red cells go through capillaries, they clog the flow and break apart. This causes pain, damage and anemia.

### **What is sickle cell trait?**

Sickle cell trait is a person who carries one sickle hemoglobin producing gene inherited from their parents and one normal hemoglobin gene. Normal hemoglobin is called type A. Sickle hemoglobin, called sickle cell trait, is the presence of hemoglobin AS on the hemoglobin electrophoresis. This will NOT cause sickle cell disease.

### **How do you get sickle cell anemia or trait?**

You inherit the abnormal hemoglobin from your parents, who may be carriers with sickle cell trait or parents with sickle cell disease. You can not catch it. You are born with the sickle cell hemoglobin and it is present for life. If you inherit only one sickle gene, you have sickle cell trait. If you inherit two sickle cell genes you have sickle cell disease.

### **How common is sickle cell anemia?**

It is most common in people whose ancestors come from sub-Saharan Africa, Spanish-speaking regions of Central and South America, Saudi Arabia, India and the Mediterranean. The disease occurs in approximately 1 in every 500 African-American births and 1 in every 1,200 Hispanic-American births. One in 12 African Americans carries the sickle cell trait.

### **How can I be tested?**

A simple blood test called the hemoglobin electrophoresis can be requested by your doctor. If you are found to have sickle cell trait, your partner should also be tested to determine if the baby is at risk for sickle cell disease.

## **Fragile X Syndrome**

### **What is Fragile X Syndrome?**

It is the most common form of inherited mental retardation and accounts for approximately 40% of cases with X-linked mental retardation. It is recommended that any person with unexplained mental retardation, developmental delay or autism be tested. The American College of Medical Genetics also recommended carrier testing on the basis of a family history of unexplained mental retardation. It is not currently recommended to test all women who are pregnant. For more information see: <http://www.fragilex.org/>, or [http://www.cdc.gov/genomics/hugenet/factsheets/FS\\_FragileX.htm](http://www.cdc.gov/genomics/hugenet/factsheets/FS_FragileX.htm).