



## **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.