



## **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 nutrients 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. Symptoms of the disease range from mild to severe. Typical lifespan of an affected person is 37 years, though some may live longer.

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

CF is an autosomal recessive disorder. If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis. 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.

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

A DNA blood test for some of the mutations causing CF is available. The test can be performed on blood specimens or amniotic fluid to detect carriers or affected individuals. 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. If you are a carrier of CF and your partner has a negative test and no family history of CF, the chance that your baby will have CF is less than 1%.

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

Because it is becoming increasingly difficult to assign a single ethnicity, it is reasonable to offer cystic fibrosis carrier screening to all pregnant patients, provided that women are aware of their carrier risk and of the test limitations. CF carrier testing is strongly recommended 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.