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Cystic Fibrosis Carrier Testing

Cystic Fibrosis is the most common inherited disease among Caucasians, affecting approximately one in every 2500 newborns. However, cystic fibrosis may affect individuals of any ethnic background.

Symptoms of cystic fibrosis include viscous mucous formation and frequent infections in the respiratory system. The digestive system may also be affected due to disorders of the pancreas. The average life expectancy of an individual with cystic fibrosis is 20-30 years. Males are almost always, and females are frequently, infertile.

Cystic fibrosis is autosomal recessive in inheritance. This means that an individual must have two copies of the gene for cystic fibrosis, one copy inherited from each parent, to have the disease. Individuals with only one copy of the gene for cystic fibrosis are carriers of the disease. These individuals usually have no clinical symptoms, but have a 50% chance of passing the gene onto their children. Thus, if both parents are carriers, they have a 25% chance of having a child with cystic fibrosis.

The National Institutes of Health consensus conference on genetic testing for cystic fibrosis recommends that all couples expecting a baby or planning a pregnancy, as well as individuals with other indications, such as a family history of cystic fibrosis, be offered carrier testing. Carrier testing may not be conclusive and may only reduce the individual's risk for carrying a gene mutation for cystic fibrosis. If both parents are found to be known carriers, further prenatal testing is available.

<u>Ethnic Group</u>	<u>Carrier Frequency</u>	<u>Detection Rate</u>
Caucasian, non-Jewish	1/29	90%
Ashkenazzi Jewish	1/29	97%
Hispanic	1/46	57%
African-American	1/60-65	75%
Asian-American	1/90	30%

A negative result for cystic fibrosis carrier testing does not rule out the possibility of being a carrier of a cystic fibrosis mutation. The cost of the test will be billed to your insurance. However, you may ultimately be responsible for the cost of the test. Although every effort will be made to ensure the confidentiality of your medical and genetic data, absolute confidentiality cannot be guaranteed.

The cost is approximately \$250.00 which may or may not be covered by your insurance carrier.