

CYSTIC FIBROSIS FACT SHEET/CONSENT FORM

“WHAT IS CYSTIC FIBROSIS?”

Cystic Fibrosis (CF) is the most common inherited disorder usually affecting people of European white ancestry. It is a severe, chronic disease that especially affects the glands, which produce sweat, mucus, and digestive enzymes. Approximately one in every 2500 Caucasian newborns is afflicted with CF, and about 30,000 Americans now suffer from this disease. Affected individuals require intensive medical and respiratory therapy every day throughout their lives. In addition, men who have CF are sterile. The average life expectancy is 31 years.

Among all of our thousands of pairs of genes, only one gene pair is responsible for CF. The CF gene pair of a CF carrier includes one normal gene and one defective gene. The presence of one normal gene is sufficient to protect the carrier from ever developing CF. Again, carriers have no rush for developing CF. Approximately one in every 25 (4%) Caucasians carries one faulty CF gene, and the majority of these individuals remain unaware of their carrier status until they have a child with CF. If both members of a couple are carriers of CF, however, they have a 25% chance for having a child with CF, a 25% chance for having a child who is neither affected nor a carrier, and a 50% chance for having a child who is a carrier for CF (but unaffected).

“WHAT IS THE CARRIER DETECTION TEST?”

There are many different mutations which can render this specific gene “faulty.” Now, 86 mutations or “changes” in the CF gene can be identified, which allows for the detection of approximately 90% of carriers in the non-Jewish European Caucasian population living in North America. Screening for these same 86 mutations also allows us to detect approximately 97% of CF carriers in the Ashkenazic Jewish population.

SINCE WE ARE NOT ABLE TO IDENTIFY ALL CARRIERS WITH THIS TEST, A NEGATIVE TEST RESULT DOES NOT MEAN THIS PERSON TESTED IS NOT A CARRIER OF THE CF GENE. A NEGATIVE RESULT SIMPLY MEANS THAT THE LIKELIHOOD OF BEING A CF CARRIER IS SIGNIFICANTLY REDUCED.

“FOR WHO IS IT RECOMMENDED?”

We are offering CF carrier screening to everyone that is having prenatal testing or IVF at our center. CF is most common in the Northern European population but all ethnic groups have some risk, which will vary depending upon countries of origin. Listed below are current statistics we have regarding the risk reduction associated with a negative CF screen for various ethnic groups. Prenatal diagnosis for CF will be available if both individuals test positive for a CF mutation. If one partner screens positive, it will take at least two additional weeks to obtain results from the untested partner and/or fetus.

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(The statistics below assume that there is no family history of CF)

Prior Carrier Risk	Detection Rate (New carrier risk)	New Risk (One parent negative vs. both parents negative)
Northern European (i.e. English, Dutch, Swedish 1/25 (4%))	90% (1/250)	1/25,000 vs. 1/250,000
Ashkenazi Jewish (1/26 (3.9%))	97% (1/834)	1/86,736 vs. 1/2,782,224
African American 1/65 (1.5%)	75% (1/260)	1/67,600 vs. 1/250,400
Southern European (i.e. Greek, Italian, Portuguese) 1/25 (4%)	74% (1/83)	1/8,300 vs. 1/27,556

Other ethnic groups do not have available statistics.

Insurance may not cover this testing, but we can attempt to bill your insurance. Results will be available in approximately two weeks.

POINTS TO REMEMBER:

1. A positive result means you are a carrier. A negative test result means you are at a reduced risk to have a child with CF (NOT zero risk!).
2. If one partner tests positive, it will take another two weeks to obtain results on the spouse/partner and fetus.
3. There is approximately a 3% risk for birth defects, genetic conditions and mental retardation in the population. This testing will not guarantee that a fetus will be completely healthy.



I have had the opportunity to read the Cystic Fibrosis Fact sheet and choose the following option regarding testing:

CONSENT:

_____ I have read and understood the Cystic Fibrosis Fact Sheet information and I agree to have CF carrier testing**

_____ I have read and understood the Cystic Fibrosis Fact Sheet information and I decline CF carrier testing

Print Patient Name: _____

Signature: _____ Date _____

Print Partner Name: _____

Signature: _____ Date _____

****If you are an Advocate PHO Humana Capitated patient you will need to have this test performed at your PCP office**