







Screening for Cystic Fibrosis

Cystic Fibrosis (CF) is a genetic (inherited) disorder that affects the lungs and digestive system of about 30,000 children and adults in the United States (70,000 worldwide). More than 10 million Americans are symptomless carriers of the defective CF gene. The disease is the most common, deadly, inherited disorder affecting Caucasians in the United States. An estimated 1 in 29 Caucasian Americans have the CF gene. It is most common among those of Northern or Central European descent. The risk for carrying the CF gene is somewhat lower in non-Caucasian groups - 1-in-46 for people of Latino background, 1-in-65 for African-Americans, and 1-in-90 for Asian-Americans.

People with Cystic Fibrosis produce thick mucus in their lungs causing difficult breathing. This mucus also may promote infections that can be life-threatening. CF secretions also can cause problems with food absorption by blocking pancreatic enzymes. Males with Cystic Fibrosis are generally sterile. Because there are more than 1,000 mutations of the CF gene, symptoms and their severity differ from person to person. Currently, the median age of survival is 36.9 years; with the median age of survival significantly higher in males than in females.

Cystic Fibrosis is a recessive genetic disorder. Both parents must carry a copy of the abnormal gene for the problem to occur in their child. A person who has one copy of an abnormal gene for a recessive disorder is a *carrier* for that disorder, even though he or she may show no signs of it. If both parents are carriers, each of their children has a 25% or a 1-in 4 chance of having the disorder. The diagram to the right illustrates the possible offspring of two parents who are carriers of the CF gene.

Possibility of a Child with CF When Both Parents are Carriers of the Gene			
Father is a CF Carrier		Mother is a CF Carrier	
			
Child Has CF – 25%	Child is a Carrier of the CF Gene, but does not have CF – 50%	Child is a Carrier of the CF Gene, but does not have CF – 50%	Child Does not have CF and is NOT a carrier – 25%

When both parents carry the CF gene, there is a 25% chance (1 in 4) that their child will have CF, a 50% chance that the child will be a carrier just like the parents, a 25% chance that the baby will not have the gene - not a carrier and not have the disease.

CF Carrier Screening is a blood or saliva test that checks whether or not parents-to-be have (carry) the abnormal gene that causes CF. This test can help determine your risk for having a child with CF. Couples can choose to test one partner, and then the other if one is a carrier; or both partners may be tested at the same time. During pregnancy, screening both partners at the same time can be helpful if the couple thinks they might be at high risk and would consider further prenatal testing to see if the fetus has CF.

If your test results are normal, the chance that you are a CF carrier is small. *There are some rare CF gene defects that the test does not detect. For this reason, you could be told your test result is normal, and you could still be a carrier.* The likelihood of this situation is very small.

Deciding whether or not to have the test is your own personal choice. Talk it over with your partner and with your health care provider and get as much information as you need to decide what's right for you and your baby.

Other References

www.cff.org – Cystic Fibrosis Foundation

www.acog.org – American College of Obstetricians and Gynecologists

www.marchofdimes.com – March of Dimes