



Cystic Fibrosis Carrier Testing

Cystic Fibrosis (CF) is a chronic illness that affects approximately 1 in 3,000 newborns in America. It typically begins in the first years of life with digestive and breathing problems due to thickened mucus that:

- Clogs the lungs and leads to life- threatening lung infections
- Obstructs pancreas and stops natural enzymes that help the body break down and absorb food.

CF does not affect intelligence or appearance. Although progress has been made in understanding this condition, there is no cure for CF. Cystic Fibrosis is a life-long illness that is usually diagnosed in the first few years of life. The disorder causes problems with digestion and breathing. Medications and breathing treatments help treat symptoms. While in general people with CF have a shortened life span, some die in childhood, and others live into their 40s or even longer. Although there is no cure for CF, research on more effective treatments is under way.

The American College of Obstetrics and Gynecology recommends screening for CF to be considered by all:

- Individuals with family history of CF
- Partners of individuals who have CF
- Couples that are planning a pregnancy or beginning prenatal care.

What causes cystic fibrosis?

Cystic fibrosis is a genetic disorder. All genes come in pairs, so everyone has two copies of each gene. One copy comes from your mother and the other from your father. For some diseases- like CF- both genes of the pair have to be altered for a person to have the disease. If a person has one changed copy of a CF gene, that person is a carrier for CF. A carrier **does not** have CF. There are no known health problems associated with being a carrier. If a person has two changed copies of the CF gene, they will develop CF. When both partners in a couple are carriers, any child they have has a 1-in-4 (25%) chance to inherit a changed copy of the gene from each parent. A child with two changed copies of the CF gene will develop CF.

What is the purpose of cystic fibrosis testing?

The purpose of CF testing is to see if the couple is at increased risk for giving birth to a child who will have CF. Cystic fibrosis carrier testing is a laboratory test done on a sample of blood. If testing shows that a couple is at high risk, additional testing can be done on the developing baby to see whether or not it will have CF. Cystic fibrosis cannot be treated before birth. The purpose of having this information about your developing baby is so you can prepare yourself to care for a child with special health care needs. It is important to understand that CF carrier testing does not detect all carriers. If your test does not show a mutation on the CF gene, the chance that you could still be a CF carrier is very low. Diagnostic testing can be performed prenatally on amniotic fluid when both parents are found to be carriers.

Could I be the carrier of cystic fibrosis?

Yes. You could be a carrier of CF even if no one in your family has CF and even if you already have children without CF. About one of every 30 white people (about 3 in 100 or about 3%) carries the changed gene. See table below to evaluate your chance of being a carrier.

Ethnicity/ Race	Change of Being a CF carrier	Chance Both Partners Are CF Carriers
European Caucasian, Ashkenazi Jewish	1 in 29	1 in 841
Hispanic American	1 in 46	1 in 2,116
African American	1 in 65	1 in 4,225
Asian American	1 in 90	1 in 8,100

* If a relative of yours has CF, or is known to be a carrier of CF, your chance of being a carrier is greater based on your family history than your ethnic background.

You can have a child with a CF even if there is no history of it in your family, especially if your ancestry is Western European or Ashkenazi Jewish. Carriers do not have CF themselves. Both parents must be carriers of CF in order to have an affected child. CF screening can help determine if you are a carrier and at risk to have a child with CF.

Will my insurance cover the cost of testing?

Insurance coverage varies from policy to policy. In general, MediCal or Medicaid do not cover the cost of CF screening, HMO's will need prior authorization before CF screening can be performed, and PPO's will usually cover at 80% or more based on your policy provisions.