

# CYSTIC FIBROSIS

Everything you  
need  
to know about  
being a carrier

## What Does It Mean To Be A Carrier of Cystic Fibrosis?

You've recently learned that you are a carrier for cystic fibrosis. You should know that being a carrier is not an illness and does not affect your health. In fact, you would probably not have found out that you are a carrier without having a special blood test. Being a carrier simply means that one copy of the gene that can cause cystic fibrosis has a change that keeps it from working properly. You have a second copy of the cystic fibrosis gene that is working normally.

What is a gene and what does it do? Genes are our body's "instructions" that determine our physical characteristics, such as blood type and hair color. They are located on chromosomes, which are found in almost every cell of our bodies. We inherit one set of chromosomes from each of our parents; therefore, our chromosomes, and the thousands of genes located on each of them, come in pairs.

The cystic fibrosis gene pair tells our bodies how to make a specific protein, called cystic fibrosis transmembrane regulator (CFTR). This protein affects the movement of fluid in and out of cells found in the lungs, the digestive tract and other organs in the body. If a person has one copy of the cystic fibrosis gene that does not work properly, he or she is called a **carrier** of cystic fibrosis. As long as a person has one working copy of this gene, the body has enough CFTR to do its job which is why that person does not have cystic fibrosis and never will have cystic fibrosis.

What is cystic fibrosis?

When babies inherit two non-working copies of the cystic fibrosis gene, the lungs will begin to produce large amounts of abnormally thick mucus. This mucus collects in the lungs leading to congestion, coughing and frequent infections which can lead to permanent and fatal lung damage. The digestive tract may not work normally which can lead to trouble gaining weight and growing. Intelligence and appearance are not affected except that affected individuals are often thin and may be slightly shorter than expected.

Although present from birth, symptoms of cystic fibrosis may first appear at different ages. The disease worsens as people get older and most will die in their 30's although some do not survive past childhood and others live longer.

How does one become a carrier?

Since all of our genes are inherited from our parents on their chromosomes, individuals who are cystic fibrosis carriers have inherited their non-working gene from one of their parents. This means that brothers and sisters of a carrier have a 50% chance to be a carrier.

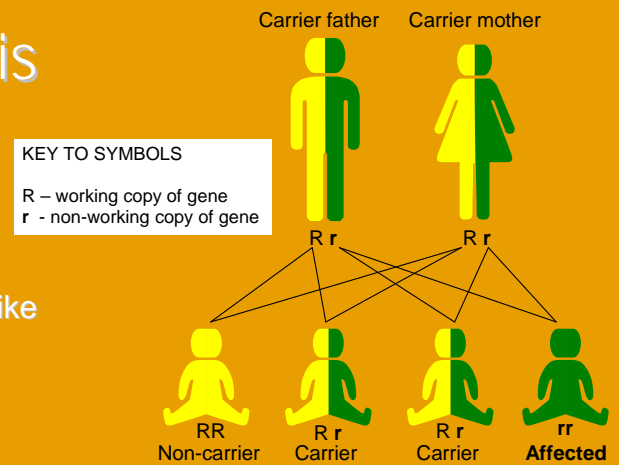
Can anyone be a carrier for cystic fibrosis?

Yes. Cystic fibrosis carriers are found in all nationalities and ethnic groups but are much more commonly found in Caucasian individuals whose ancestors came from Europe. In the European Caucasian population, about one in every 25 individuals carries a change in the cystic fibrosis gene.

# Inheritance of Cystic Fibrosis

When both parents are cystic fibrosis carriers there is:

- ❖ A 25% chance that the baby is not a cystic fibrosis carrier and does not have cystic fibrosis.
- ❖ A 50% chance that the baby is a cystic fibrosis carrier just like his or her parents.
- ❖ A 25% chance that the baby has cystic fibrosis



Is there a cure for cystic fibrosis?

No. Although research is being conducted, there is nothing that can be done to cure the disease at this time. Affected individuals receive daily chest therapy and medications and require periodic hospitalizations. Progress in the treatment of cystic fibrosis has made it possible for affected individuals to live longer than in the past, when children typically died at a very young age.

Can being a carrier lead to having cystic fibrosis?

No. Carriers of cystic fibrosis will never develop the disease because they have one working copy of the CFTR gene. Their bodies make enough of the protein to be healthy.

Can my children have cystic fibrosis?

Since children receive half of their genetic information, like eye and hair color, from their mother and half from their father, the answer to this question depends on whether or not your partner is a carrier.

If your partner is not identified as a cystic fibrosis carrier, then we would not expect that your children will have cystic fibrosis. With each pregnancy, you will have a 50% chance of having a child who is a cystic fibrosis carrier just like you and a 50% chance of having a child with two working copies of the cystic fibrosis gene. None of these children will have cystic fibrosis.

If your partner is also a cystic fibrosis carrier, then there are three possible pregnancy outcomes, including the possibility of a child with cystic fibrosis (see diagram above).

To find out the chance of having a baby with cystic fibrosis, your partner should be tested. The accuracy of the results of this test varies depending on ethnic background.

What choices do I have if my partner is also a carrier of cystic fibrosis? There are several options available if both you and your partner are carriers. In order to find out before birth if a baby has cystic fibrosis, prenatal testing can be performed as early as the 10<sup>th</sup> week of pregnancy. If the results are normal, the parents can be reassured. If the results show that the baby will be affected, the parents can be better prepared, and they can make informed decisions regarding continuation or termination of the pregnancy.

Will my child be tested for cystic fibrosis at birth?

All babies born in California have newborn screening for cystic fibrosis. This screening will detect most babies with cystic fibrosis and will identify some babies who are cystic fibrosis carriers.

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