Quest Diagnostics®

May, 2015 • Members

Cystic Fibrosis

What is CF?

Cystic fibrosis (CF) is one of the most common genetic diseases. It's caused by changes (mutations) in DNA that are passed down from a person's parent. These changes indirectly affect the flow of water into and out of cells. Some tissues don't get enough water. This causes mucus in the lining of tissues to be thick and sticky. This mucus clogs airways in the lungs and ducts in the pancreas. And that leads to most of the symptoms of CF.

There is no cure for CF. It's a condition that people have for life. And the symptoms get worse as time goes on.

How CF is inherited

CF is an autosomal recessive disease. This means that a child must inherit 2 mutations to get it. One is passed down from the mother and the other is passed down from the father. Most of the time, the mother and father don't have CF. They are called carriers because they "carry" a mutation in their DNA. If both parents are carriers and don't have CF, each of their children has a:

- 25% chance of getting a mutated gene from both parents. In this case they have CF.
- 25% chance of getting a normal gene from both parents. In this case they do not have CF, and they are not a carrier. They can't pass a CF mutation to their children.
- 50% chance of getting a normal gene from one parent and a mutated gene from the other. In this case they don't have CF, but they are a carrier. They can pass the CF mutation to their children.

CF and ethnicity

People of any ethnic background can have CF. But it occurs more often in some groups than in others.

Race/Ethnicity	Birth rate[1]
Ashkenazi Jewish	1 in 2,270
Non-Hispanic Caucasian	1 in 2,500
Hispanic	1 in 13,500
African American	1 in 15,100
Asian	1 in 35,100

Symptoms of CF

Most symptoms are caused by mucus that is too thick and sticky. Typical CF symptoms include:

- Persistent coughing, at times with phlegm
- Wheezing or shortness of breath
- Frequent lung infections
- Malnutrition
- Slow growth and weight gain, even with a good appetite
- Smelly, greasy stools
- Male infertility
- Salty-tasting skin

Not all people with CF have the same symptoms. Some people have mild symptoms; others have severe symptoms. This depends in part on which changes a person has in their DNA. Sometimes symptoms appear in infancy. Other times they don't appear until years later.

Cystic Fibrosis

Members

Screening and diagnosis of CF

Carrier screening is done to find out whether a person is a CF carrier. If a woman isn't a carrier, she and her partner can't have a child with CF. If she is a carrier, her partner can be screened next. If both of them are carriers, they could have a child with CF. They can then talk with their doctor to learn about their options. Or they could talk with a genetic counselor. Knowing their options will help the couple plan their family.

Doctors should offer this screening to all women who are thinking about getting pregnant. Couples who get screened before they get pregnant have the most options. For example, they could have in vitro fertilization. This can be followed by genetic testing of the embryo. Or they could have fetal diagnosis done once they get pregnant.

If not screened before, couples can be screened early in pregnancy. This is not optimal. There is only one option if both parents are carriers. That option is fetal diagnosis.

CF screening is another type of screening. It's done on the baby as part of newborn screening. If the baby screens positive, more testing will be done. This is done to find out if the baby actually has CF.

People who show CF symptoms later in life can be tested too. The sweat test is often used for this. It measures the amount of salt in a person's sweat. A positive result can be confirmed by a second sweat test. Or it can be confirmed with a genetic test that can detect CF mutations.

How doctors treat CF

There is no cure for CF. But treatment can help patients feel better and live longer. The treatments used depend on the person's symptoms. But most people with CF regularly:

- Have some kind of chest therapy to help loosen and get rid of the thick mucus in the lungs.
- Take medicine that thins the mucus and helps the person breathe easier.
- Take medicine that helps fight lung infections.
- Take medicine that helps the digestive system get nutrients from food.
- Take a multivitamin. This helps make sure the person gets enough vitamins and minerals.

How long do people with CF live?

People with CF are living much longer than they used to. This is mainly due to earlier diagnosis and treatment. Research has helped too. In the last 60 years, survival has increased greatly[2]:

- In the 1950s, most people with CF didn't live to go to the first grade.
- In 1985, people with CF lived about 25 years.
- In 2007, people with CF were expected to live about 37 years.

Quick facts about CF in the United States

- About 1 in 31 people are CF carriers.[2]
- About 30,000 people have CF.[3]
- Nearly half the people with CF are age 18 or older.[4]
- About 1 in 3,700 people are born with CF.[3]
- About 1,000 new cases are diagnosed each year.[3]
- More than 75% of people with CF are diagnosed by age 2.[2]

Additional information

You can find more information about CF at these Web sites:

- Cystic Fibrosis Foundation: cff.org/AboutCF
- CF Living: cfliving.com
- National Institutes of Health: ghr.nlm.nih.gov/condition/cystic-fibrosis

References

1. Palomaki GE, FitzSimmons SC, Haddow JE. Clinical sensitivity of prenatal screening for cystic fibrosis via CFTR carrier testing in a United States panethnic population. *Genet Med*. 2004;6:405-414.
2. Cystic Fibrosis Foundation. Frequently asked questions. cff.org/AboutCF/Faqs. Updated March 12, 2014. Accessed March 19, 2015.
3. American Lung Association. State of lung disease in diverse communities. Cystic fibrosis. lung.org/assets/documents/publications/solddc-chapters/cf.pdf. Published 2010. Accessed March 17, 2015.
4. Cystic Fibrosis Foundation. About cystic fibrosis. cff.org/AboutCF. Updated May 2014. Accessed March 17, 2015.