Cystic Fibrosis

Information for Caregivers

Arkansas Children's

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Arkansas Children's Hospital is an accredited Cystic Fibrosis Care Center by the National Cystic Fibrosis Foundation

Cystic Fibrosis: Information for Caregivers

We know parents are worried and have many questions when they find out their child is diagnosed with cystic fibrosis (CF). Its important parents know they aren't alone and there is a team of people to help them. There are things that can be done to help their child's health right away. One of the first steps is to learn more about CF.

What is Cystic Fibrosis?

Cystic fibrosis (CF) is a permanent condition. People with CF look like everyone else, but have health problems. Cystic fibrosis does not affect the brain or how smart a person is. Children with CF go to school, play sports, and get their driver's license. Adults go to college, have successful jobs, live on their own, and get married. CF does not affect every person the same way so it's hard to say how mild or severe the condition may be. There is not yet a cure, but there are many treatments. People with CF are living longer, better-quality lives than ever before.

CF is caused by genes that do not work properly. These genes are inherited from each parent. When healthy, the CF gene makes a protein called CFTR. This protein is found in the cells of many organs, like the lungs and the pancreas. CFTR stands for "cystic fibrosis transmembrane regulator". This protein helps the flow of chloride and sodium (salt and water) in and out of cells. When the CF gene is changed (called a mutation), the CFTR protein in the cells does not work properly. This causes the salt balance in the body to be unequal and leads to very thick mucus. This mucus then blocks the passages in certain parts of the body.

- Lungs/Sinuses: The CF lung is dry and the thick mucus can't easily be moved or coughed out. In time, the thick mucus builds up and clogs the airways. This can lead to infections and inflammation (swelling) in the lungs. Thick mucus in the sinuses can also cause infections.

- Digestion/Growth: Thick mucus blocks the digestive tract and pancreas. This stops digestive enzymes from getting into the intestines. Enzymes are needed to break down food, which provides nutrients to help people grow and stay healthy.

- Liver, bone health, and reproductive systems: Liver disease, CF-related diabetes, bone mineral issues, and low vitamin levels can also occur. Most men with CF are unable to naturally conceive a child. However, methods now exist to help this problem. Women with CF may become pregnant, but have special health concerns. They should talk to their doctor if thinking about having a baby.

See Figure 1 showing a normal CFTR protein versus non-working CFTR protein

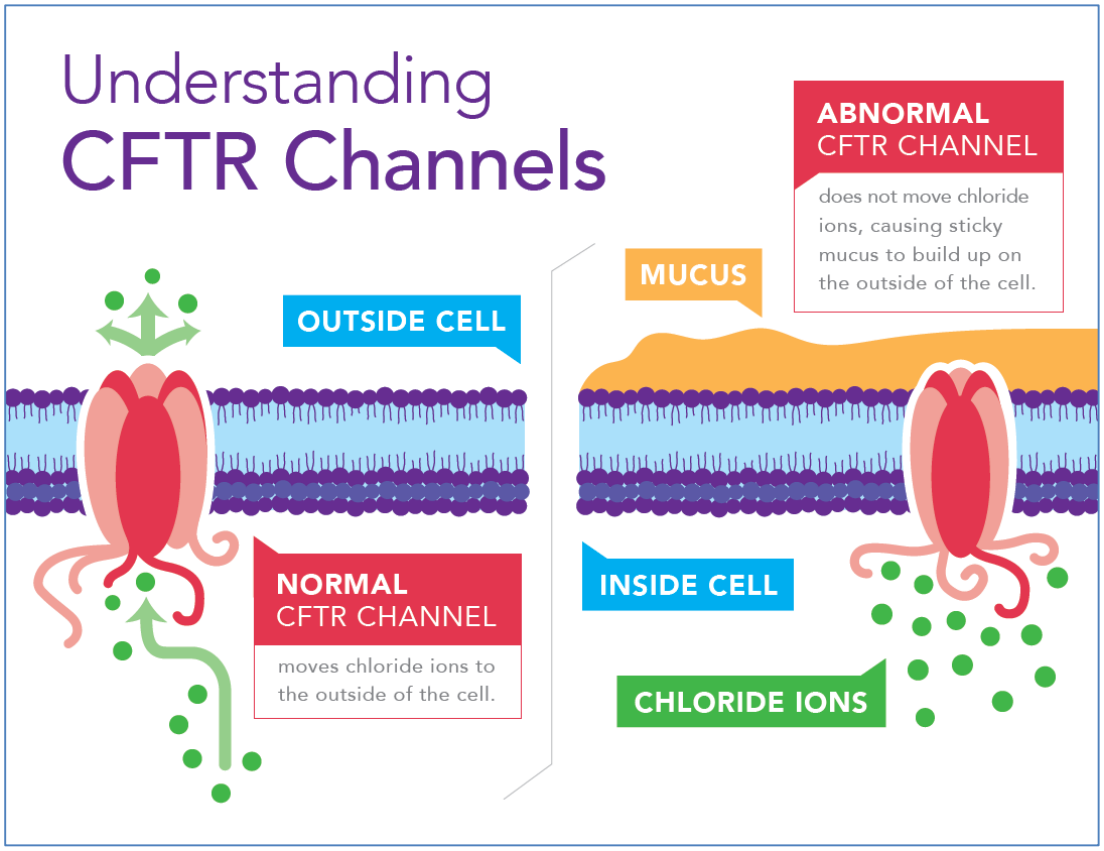

Figure 1

Who gets CF?

Around 30,000 people in the United States have CF. It is most common in Caucasians, but it can affect all races. The CF symptoms are different from person to person. Research shows that the severity of CF symptoms is partly based on the types of CF gene mutations. Some mutations are known to cause more severe problems than others. Scientists have found more than 1,900 different mutations of the CF gene.

How do people get cystic fibrosis?

The root cause of CF is genetic. This means people inherit CF from their parents through genes (or DNA). Genes are found in the cells of the body and provide instructions to make proteins. Proteins define how the body looks, grows, and develops.

In CF, cells in parts of the body aren't working normally due to a change in the CF gene, called mutation. The CF gene mutation then gives wrong instructions to the protein (called CFTR) about how to work. This protein controls the flow of chloride and sodium (elements of salt) in and out of these cells. When the protein doesn't work properly, as in cystic fibrosis, the salt balance in the body is disturbed. Some changes in the CF gene can cause mild problems, others cause more severe symptoms.

To have cystic fibrosis, a person must inherit two copies of the non-working CF gene - one copy from each parent. Both parents of a child with cystic fibrosis are CF carriers. Being a CF carrier means that someone carries one CF gene mutation, but the other copy of the gene is healthy. A CF carrier will not have CF. A CF gene mutation can be passed down in the family for many years without someone having the disease. This is why receiving a CF diagnosis often surprises some families. Many, if not most people, do not know they are carriers until they have a child with CF.

Each time two carriers of a CF gene change have a child, the chances are:

- 25% (1 in 4) the child will have CF;
- 50% (1 in 2) the child will carry the CF gene but not have CF; or
- 25% (1 in 4) the child will not carry the gene and not have CF

See the Figure 2 below for an example of two carrier parents and how that may affect their children.

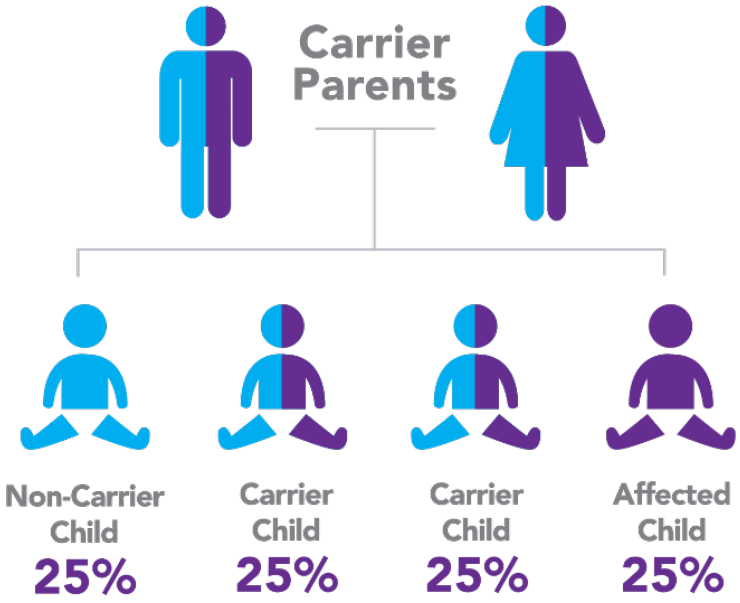

Figure 2

In a family with four children, it is possible for none or all of them to have CF. Each child has the same chance to inherit CF gene mutations from both parents, no matter if any of the other siblings are carriers or have CF.

Among Caucasian Americans, about one in 29 people are CF carriers. In other races, one in 46 Hispanic Americans, one in 65 African Americans and one in 90 Asian Americans are CF carriers.

Who can be tested for CF?

Arkansas screens all newborns for CF. This is important because catching the condition early leads to prompt treatment and better health in the long run. Adults and older children are not routinely screened for CF, but can choose to be tested. This happens sometimes when a child in the family is born with CF and other relatives wish to know if they could have children with CF or if they are a CF carrier. It is advised that all siblings of babies born with CF also be tested.

It is also possible for a baby to be tested for CF while in the womb. This is not a routine test. It may be suggested when the baby has a higher chance of having CF, such as family history or if parents know they are carriers. Couples can also consider using the in vitro fertilization process to learn if an embryo has CF before it is implanted. To learn more, contact your doctor, obstetrician, or genetic counselor.

What is the treatment for CF?

The CF Foundation created treatment guidelines for CF care. CF Centers around the country follow these guidelines as they are based on research and facts. There are many treatments available, but not everyone may use the same ones. The CF Team will create a specific plan for each person based on test results and needs. Common treatments include:

- Monthly clinic visits from birth to 1 year of age
- At least four clinic visits each year throughout life (after 1st birthday)
- Yearly tests to be done at one of the clinic visits (chest x-ray, blood work, etc.)
- Daily airway clearance therapy to remove mucus from lungs
- Daily inhaled medicine (bronchodilators, mucus thinners, antibiotics)
- Taking enzymes with meals and snacks to help digest food
- Taking extra vitamins special for CF
- Eating more food, calories, and fats than normal
- Being active or exercise regularly

Most CF care is done in the home as part of daily routine. Care is done by caregivers when children are young, but the long-term goal is for children to learn to do their own treatments to prepare for adult life. Sometimes it may be necessary for someone with CF to come into the hospital. There are several reasons this could happen, but one might be to get medicine through a tube in the arm (called an IV) to fight an infection.

New treatments are now available to correct certain CF gene mutations. These new drugs help correct the flow of salt and water in and out of the cells. This restores the balance in the affected areas of the body. These medicines can't be used for all the CF gene mutations yet, but research is ongoing to create new drugs that will help all people with CF**.**

Staying Healthy:

Besides treatments, there are other suggestions to help reduce the chance of people with CF getting sick, such as stopping the spread of germs. People with CF can spread germs to each other that do not harm someone without CF. Also, if they get a cold or flu, it's often harder for them to get well and can lead to lung infections. To prevent spread of germs, the CF Foundation recommends the following:

- Regular immunizations
- Yearly flu vaccine (for caregivers and the person with CF)
- Learn how to cough or sneeze into a tissue and throw it away
- Wash hands regularly (after coughing or sneezing, enter and leaving the clinic or hospital, after using the restroom, and after doing treatments)
- Keep at least 6 feet from other people with CF or others who have colds or infections

- Wear a mask in the hospital and clinic areas
- Disinfect medical equipment in the home as directed by CF Team
- Keep regular check-ups with primary care doctor

Your CF team will also wear gown and gloves when entering the clinic or hospital room. This is to protect from passing germs from one patient to another.

Everyone affected by CF (child and caregiver) should pay extra attention to self-care. This means doing things like taking breaks when you need them, getting enough rest, getting help from others, and finding healthy ways to deal with the emotions and concerns you have.

Where can people with CF get the best care?

People with CF should be seen at an accredited CF Care Center. This means the center is routinely evaluated by the CF Foundation to make sure it is providing the most current recommendations in the safest way. Arkansas Children's Hospital is the only accredited pediatric CF center in the state. The CF center uses a team of specialists to work together with caregivers to treat CF at every age and stage. Team members include:

- Pulmonary doctors
- Nurses
- Dieticians
- Respiratory therapists
- Pharmacists
- Social Workers
- Psychologists

What is the life expectancy?

There is no way to know how long each person with cystic fibrosis will live. Many different factors affect the health of someone with CF such as:

- severity of disease
- age at diagnosis
- adherence to treatments
- lifestyle choices
- inherited resilience traits

In 1955, people with CF were not expected to live out of childhood or attend grade school. However, data from the CF Foundation shows that more than 49% of people with CF living in the United States are 18 years of age or older. The average age of survival for people with CF

increases each year. In 2013, it was 41 years of age. These numbers show that advances in treatment and CF care continue to improve the lives of people with CF.

We hope this information has made you feel more informed about CF and what to expect. While it may seem overwhelming at first, be encouraged that many people with CF respond to challenges with great strength, humor, and zeal for getting the most out of life. There is a team of specialists around the world working very hard to make life better for people living with CF. There are many resources available where you can go to learn more. We hope you will consider our CF team your biggest resource of all. We look forward to getting to know your child and family.

Resources:

- Cystic Fibrosis Foundation: www.cff.org
- Arkansas Cystic Fibrosis Care Center: www.arkansasCF.com
- Genetics Home Reference summary of cystic fibrosis: http://ghr.nlm.nih.gov/condition/cystic-fibrosis
- Clinical and Functional Translation of CFTR: http://www.cftr2.org/

For more information or questions, please contact the Cystic Fibrosis Center or the Genetics department at Arkansas Children's Hospital.

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