

UNDERSTANDING CYSTIC FIBROSIS (CF) INSIDE AND OUT

STAY CF SMART



KENNEDY AND CARSON
Siblings living with CF



Explore CF with me, Eugene
the gene, in 2 editions of
[CF Behind the Scenes!](#)

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UNDERSTANDING THE IMPACT

Cystic fibrosis (CF) feels different for everyone, but every person with CF experiences progression.

This brochure can help you understand what causes CF and how it impacts the body.

What causes CF?

It all starts with a problem within the cells. Mutations found on both copies of a specific gene cause proteins throughout the body to be made incorrectly.

This results in a buildup of thick, sticky mucus in the lungs and beyond. The mucus buildup leads to inflammation and infections, which can cause damage even before it can be felt.

Learn even more
about how CF impacts
the body with me,
Eugene the gene!



THE ROLE OF GENES

Genes carry the instructions for every part of the body. An error or mistake in these instructions can cause certain diseases.

A specific gene is responsible for CF. It's called the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene. This gene contains the instructions for producing CFTR proteins, which do an important job on the surface of certain cells throughout the body.

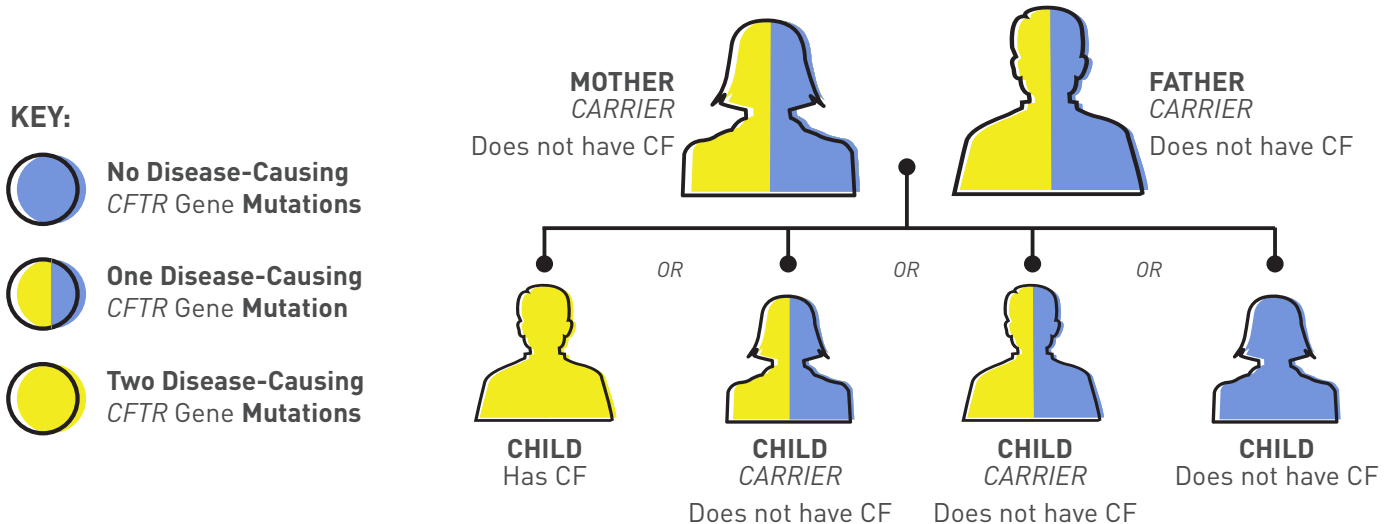
But when someone has CF, a change (called a mutation) in each of the 2 copies of this gene results in CFTR proteins that aren't able to do their job.



Of the many possible mutations in the *CFTR* gene, not all mutations can cause the protein defects that result in CF.

Just like other genes, the *CFTR* gene is inherited from parents.

Every person has 2 copies of this gene—not just people with CF—and gets 1 copy from each parent. A person with CF has 2 abnormal copies of the *CFTR* gene; the abnormality is a change called a mutation.



If both parents are carriers of a disease-causing *CFTR* gene mutation, each of their children has a 25% chance of having CF and a 50% chance of being a carrier.

CF AND YOUR GENOTYPE

Different mutations affect CFTR proteins in various ways. As a result, CF symptoms and when they occur can be different for everyone.

Knowing their genotype can help someone with CF:

- Better understand CF symptoms and how CF can progress
- Build a personalized and effective CF care plan with the healthcare provider

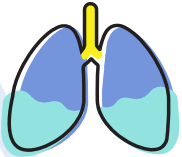
Unsure about your child's genotype? Here are important things to ask the healthcare provider at your next visit:

- What steps should I take to find out my child's genotype?
- How does the genotype play a role in developing a CF care plan?
- How do CFTR mutations affect the way the body works?

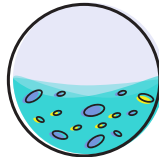
THE UNDERLYING IMPACT OF CF

CF AFFECTS DIFFERENT ORGANS IN DIFFERENT WAYS

Lungs: Thick, sticky mucus causes infection and lung damage



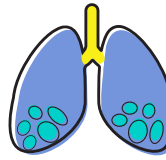
Mucus builds up in the lungs
and can't be cleared from the
airways easily



Germs build up in the mucus
causing infection, inflammation,
and scarring



Symptoms may develop such as
wheezing, shortness of breath,
and persistent, phlegmy cough



**Buildup of mucus and germs can also
lead to pulmonary exacerbations.**

Most people with CF are familiar with pulmonary exacerbations as periods of time when symptoms and/or lung function worsen. Pulmonary exacerbations may require treatment with antibiotics (by mouth, inhaled, or intravenous [IV]) and/or a hospital stay.

Digestive system: Thicker digestive fluids damage organs



In the pancreas

- Thick fluids block ducts (small tubes) within the pancreas, an organ that makes enzymes that break down food
- Blocked ducts make it harder for digestive enzymes to reach the small intestine, where they do their job
- Without these enzymes, the body has trouble breaking down food and absorbing nutrients. Not having these enzymes leads to poor growth and slow weight gain



In the liver

- Thicker bile blocks bile ducts and can cause irritation or inflammation in the liver



In the intestines

- Thicker digestive fluids can also block the intestines. In most people with CF, blockage of the intestines mainly leads to chronic constipation. It can also lead to acute conditions such as meconium ileus and distal intestinal obstruction, also known as DIOS



CF can also affect other parts of the body, which can lead to problems like sinusitis and infertility. To learn more about these conditions, visit [CFSource.com](https://www.cffsource.com).

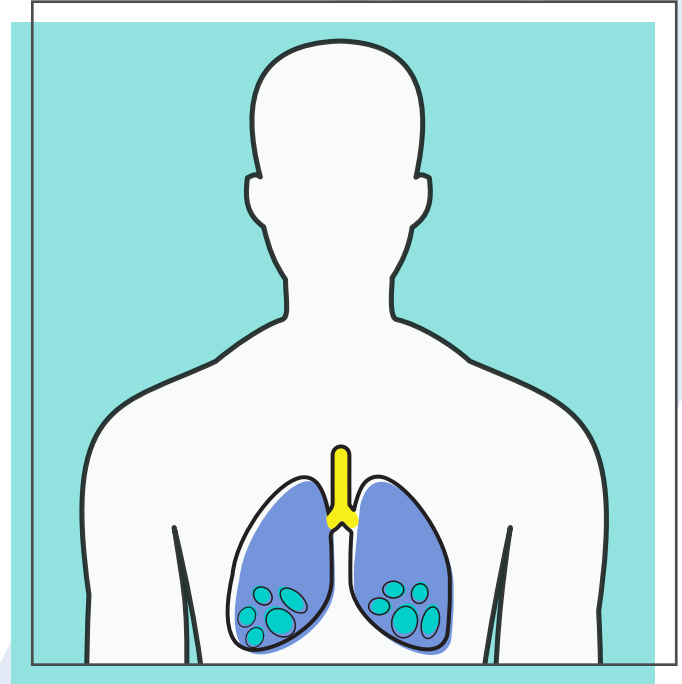


Follow me, *Eugene the gene*, to see what else happens when CFTR proteins don't work like they should.

PROGRESSION IN THE LUNGS

Beginning early, the buildup of thick, sticky mucus in the lungs results in a cycle of infection, inflammation, and more mucus buildup. Pulmonary exacerbations are often a key part of this cycle.

This is especially serious because a pulmonary exacerbation means more than just a hospital stay or an extra round of antibiotics. It may cause permanent lung damage that advances disease progression.



Bronchiectasis causes loss of lung function

It's important to promptly treat pulmonary exacerbations to prevent and slow down the progression of **bronchiectasis**, a permanent reshaping of the airways that causes them to become loose and scarred.

Bronchiectasis eventually affects almost all people with CF and can make it harder to clear mucus from the lungs and move air in and out of the airways. As it worsens, the lungs become more damaged, leading to a loss of lung function.

If left untreated, this cycle of infection and inflammation causes bronchiectasis, and can result in a permanent loss of lung function.



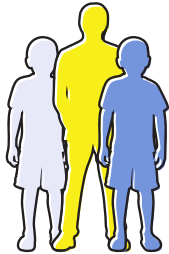
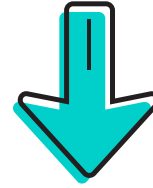
In CF, permanent lung damage may occur even before it can be detected by lung function tests.



Starting from a young age, people with CF may experience a lung function decline of 1 to 3 percentage points each year on average.

It's never too early to talk to a healthcare provider about potential lung damage

Part of what makes lung function decline so serious is that some people don't always feel the change. So someone may lose a lot of lung function before they begin to feel like something is wrong.



CF is different for everyone. But all cases of CF progress over time. The first step toward managing CF is to take a proactive approach to care.



IMPACT ON OTHER PARTS OF THE BODY

CF impacts other parts of the body as well. This can be caused by the progression of the disease or the effects of necessary medicines used to treat it.

Impact on the Pancreas



The pancreas does 2 important jobs. It makes enzymes that break down the fats and proteins in food. It also makes hormones, such as insulin, which help regulate blood sugar levels. As CF progresses, it can limit the pancreas' ability to do these jobs.

Pancreatic insufficiency

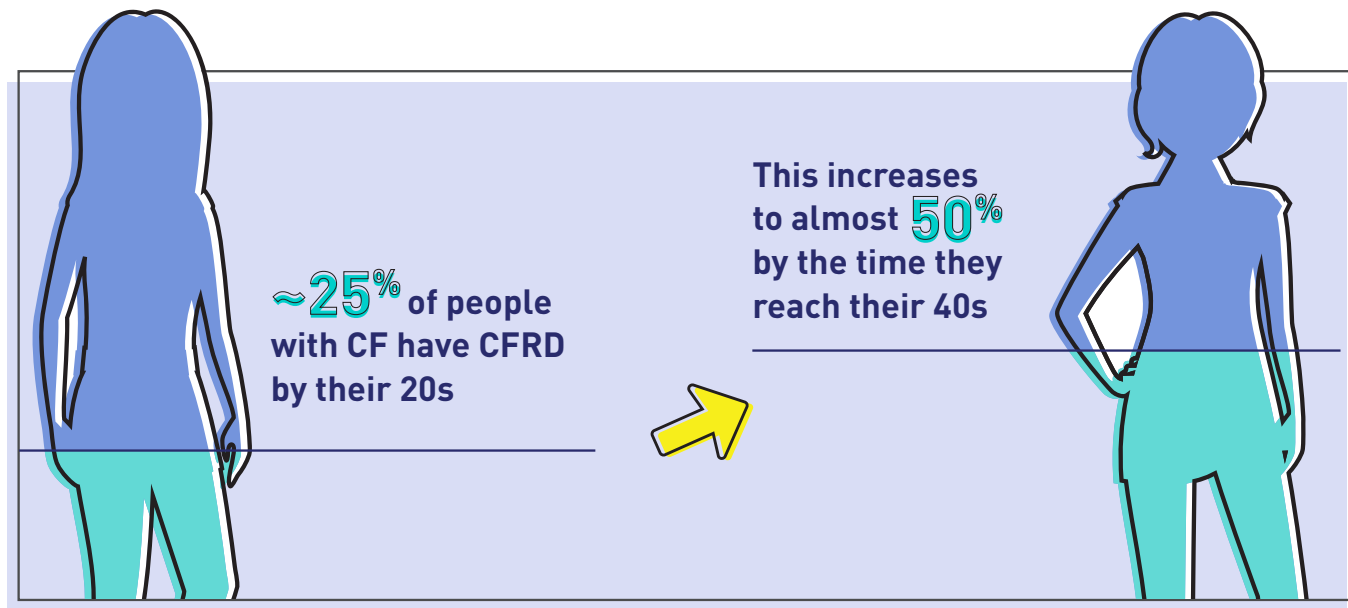
In people with CF, thick fluids block small tubes in the pancreas called ducts. This blockage makes it harder for the enzymes to reach the small intestine where they help digest food. Not having a sufficient amount of these digestive enzymes in the small intestine is called **pancreatic insufficiency**.

Pancreatitis

The small group of patients that remain pancreatic sufficient may experience a condition called pancreatitis. Pancreatitis is inflammation of the pancreas, and it can result in severe symptoms such as abdomen and back pain, nausea, vomiting, fever, rapid pulse, and weight loss.



85% to 90% of people with CF have pancreatic insufficiency (PI), which is common even at a young age.



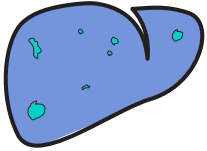
The pancreas

Over time, scarring in the pancreas, caused by thick digestive fluids, can damage the cells that produce insulin. Insulin is a hormone that is essential to regulating blood sugar.

Some people with CF can develop cystic fibrosis-related diabetes (CFRD), a form of diabetes seen in people with CF. CFRD is when the pancreas doesn't make enough insulin. When acutely ill, the body may not properly use the insulin it does produce. As a result, blood sugar levels will increase.

Note that CFRD is different from type 1 and type 2 diabetes and, therefore, is treated differently.

Impact on the Liver



Liver disease is considered to be one of the most serious health risks associated with CF. People with CF have thicker bile, and as a result, the tubes that drain bile—bile ducts—can become blocked. This often leads to irritation or inflammation in the liver.

Over time, thick bile can lead to scarring and damage of the liver, which can cause a condition called **cirrhosis**. Keep in mind that cirrhosis occurs in the late stages of CF-related liver disease and not everyone with CF-related liver disease experiences it.



Cirrhosis makes it difficult for blood to travel through the liver. It can cause many complications, including:

- Low blood counts
- Bleeding in the esophagus
- Fluid buildup in the abdomen
- Difficulty breathing



About 10%-20% of people with CF develop liver disease, which includes cirrhosis, a buildup of fat in the liver, hepatitis, and/or other complications.



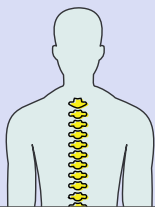
Care teams regularly monitor liver function in people with CF to check for progression in the liver.

Impact on the Bones



For people with CF, different factors contribute to **low bone density**, a condition where bones become less dense and are not as strong as they should be. Some people with CF may lose bone density more easily. Low bone density is sometime referred to as osteopenia, and when severe, osteoporosis.

For people with CF, bone health may be affected because they often have trouble absorbing enough vitamin D. Another factor contributing to low bone density is the increased inflammation throughout the body that is made worse by frequent lung infections.

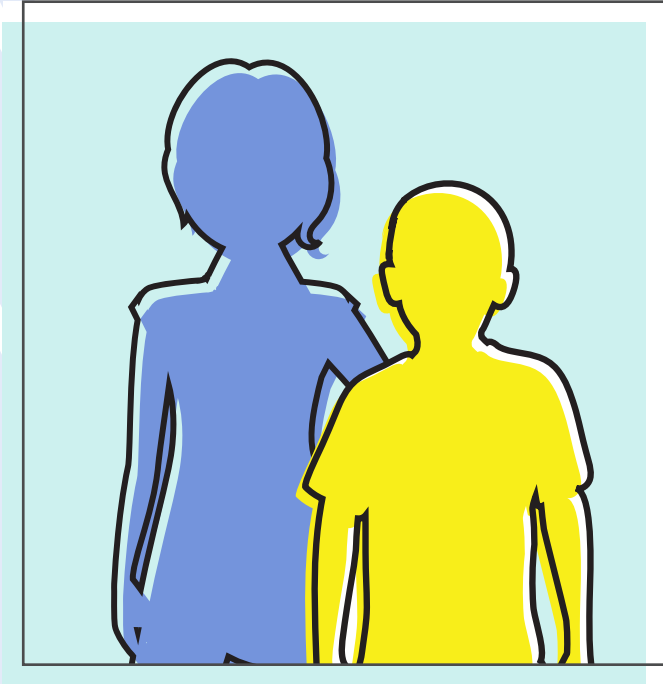


Up to 26% of adults with CF may have low bone density. Care teams begin screening people with CF for bone health at an early age.



To learn how CF affects other parts of the body, visit [CFSource.com](https://www.cffsource.com).

CARING FOR SOMEONE WITH CF



Caring for someone with CF as a parent or caregiver? There are ways to adapt your approach as they move through different stages of life.

Keep in mind that it's always a good idea to meet with the professionals at your child's CF Care Center to develop a care plan that fits your child's specific needs. Visiting your child's CF Care Center every 3 months, or 4 times per year, is a crucial part of managing their health.

Here are some age-specific tips to keep top of mind while providing care for your loved one.

Infants

Finding out about your child's CF can be overwhelming. You may be concerned about their future as you continue to learn about their condition and how to provide the best care you can. Here are a few things to keep in mind.



Some of the **procedures, tests, and scans** you can expect during routine appointments:

- Airway clearance techniques
- Respiratory cultures
- Chest x-rays



Know which **vitamins and nutrients** your child needs



Learn to address your child's **respiratory issues** and how to protect your child against infection



Influenza vaccination. Your child should have the seasonal flu vaccine at 6 months of age, then another at least 1 month after that dose. After reaching age 1, he or she should receive the vaccine annually.

- Make sure you, the members of your household, any out-of-home caregivers, and your child all get the annual influenza vaccine

Preschool-Aged Children

Children Ages 2–5

Continuing to care for your child in these early years brings a new set of challenges and opportunities. Here are some important things to keep in mind along the way:



- Stay up to date with vaccinations



- Monitor your child's respiratory health

- Perform airway clearance techniques daily and increase when ill as directed by your care team



- When your child experiences a pulmonary exacerbation, you may need to:
 - » Perform airway clearance techniques more frequently
 - » Give your child oral, inhaled, and/or intravenous (IV) antibiotics



- Check your child's weight regularly

School-Aged

Children Ages 6–12

As your child continues to grow and eventually reaches their pre-teen years, they'll become more aware of their CF and how it affects their life. Here are a few tips to help you navigate the changes you may see during this time.

- Help your child learn what causes CF and the importance of knowing about their disease and their genotype
- Begin to teach your child about how CF impacts the body
- Encourage your child to participate more actively in their care
- Help your child learn why it is important to perform airway clearance techniques

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Siblings living with CF



Adolescents

Children Ages 13–17

The teenage years are hard enough when you don't have CF; having CF only makes things harder. Here are a few tips to consider as you continue to provide your best care for a loved one on the brink of adulthood.



- **Encourage your teen to play an active role during appointments by:**
 - » Asking and answering questions on their own
 - » Knowing the reasons for yearly checkups and tests
 - » Learning the right food options from a nutritionist



- **Begin the steps for transitioning your teen from pediatric to adult care**



- Teach your teen to recognize when their symptoms get worse and how a **pulmonary exacerbation** can lead to lung damage



- **Help your teen keep up with their digestive health by:**
 - » Reminding them to take their digestive enzymes



- **Be on the lookout for diabetes**

Adults

As your loved one reaches adulthood, they might already have a good understanding of their CF. The key is to make sure they are equipped to manage their own care as they become fully independent.



- **Communicate with your loved one's care teams during the transition from pediatric to adult care**



- **Prepare your loved one for the transition to adult care**
- **Give your loved one more responsibility over time**



- **Make sure your loved one knows:**
 - » How to fit their treatments into their daily routine
 - » What their medications do, the potential side effects, and how to get refills
 - » About lung function and how to prevent infections
 - » The signs and symptoms of cystic fibrosis-related diabetes (CFRD)
 - » When they need to call the healthcare provider



- **Encourage your loved one to manage their CF by:**
 - » Getting sputum cultures every few months
 - » Having posterior/anterior and lateral chest X-rays every 2 to 4 years, or as needed (for example, during pulmonary exacerbations)
 - » Performing airway clearance and exercising regularly to keep their lungs healthy

START A CONVERSATION ABOUT CF PROGRESSION

Whether you have CF or are a parent or caregiver for someone who has CF, it's common to have questions about what progression looks and feels like, how it may affect you or your child, and how to measure and track it. Here are a few questions you may want to ask at your next care center visit.



How can I find out what genotype my child has?

What are some signs of disease progression?

How can someone with CF find out if they're experiencing progression?

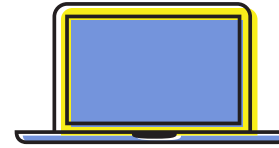
What are the signs of cystic fibrosis-related diabetes (CFRD)?

What are the signs of distal intestinal obstruction syndrome (DIOS)?

Have more questions? Add your own here:

MORE WAYS TO STAY CF SMART

Visit CFSource.com for helpful online resources, videos, and more



Learn More About CF With Eugene the Gene

It's important to get children familiar with CF from an early age. This educational series offers a fun and engaging way to learn all about CF.

Start exploring Eugene's world of interactive dictionaries, video series, and comic books.



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