

CF BEHIND THE SCENES WITH EUGENE

CAUSES AND IMPACTS

Hi there!

Welcome to CF Behind the Scenes with me, Eugene. I'm here to share some of my knowledge of cystic fibrosis, better known as CF. I'm going to take a look at what happens inside the body that causes CF.

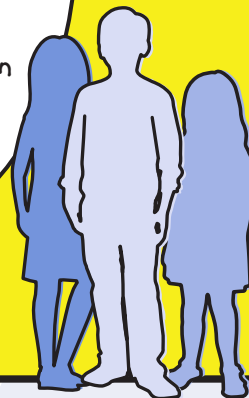
Let's go!



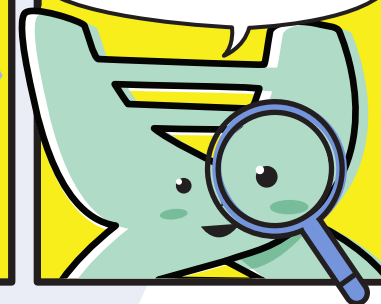
**Eugene
THE GENE**

Start here

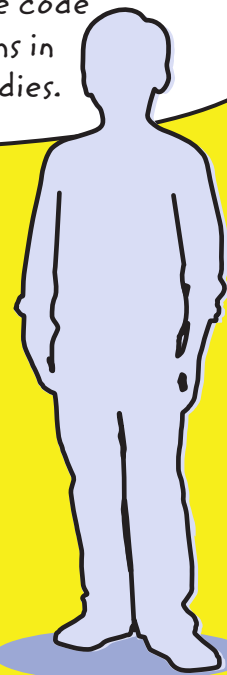
Everyone's CF is different, from everyday symptoms to the larger impact it can have over time. But the cause of CF is the same for everyone.



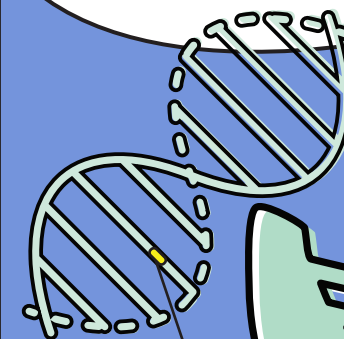
So what is the cause of CF? Let's find out.



CF starts with genes. Genes are like plans for human bodies. They determine things like eye and hair color, and the shape of your nose. Genes also hold the code for building proteins in the cells of our bodies.



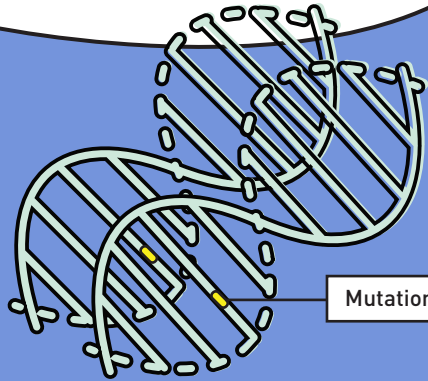
Sometimes mutations happen in genes. They can even cause diseases. Mutations in a specific gene are responsible for CF.



Mutation



Each gene has two copies. CF is caused by having two mutations in the CFTR gene, one in each copy.



Mutation

What is CFTR, you ask?
Let's look it up!



CFTR stands for cystic
fibrosis transmembrane
conductance regulator.



Thank
goodness
for abbreviations,
right?

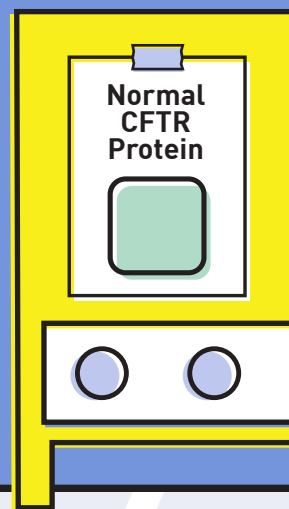
Our bodies use genes to
build proteins, but what
do proteins do?

Within the cells of
our bodies, proteins
work to do a bunch
of different jobs.



One type of protein is called a CFTR protein.
The CFTR genes we talked about earlier code
for CFTR proteins.

In people with CF, CFTR proteins aren't built
correctly. This is because of mutations in
the CFTR gene. These mutations change
how CFTR proteins are built in
2 different ways.



One way is that these proteins may not reach the surface of the cell to do their job.

Cell surface

Another is that the proteins that reach the cell surface may not work the way they should.

To understand what happens when CFTR proteins aren't built correctly, it's important to know that in the body water likes to follow salt.



In organs like the lungs, CFTR proteins help salt move outside the cell, and water follows it. This helps keep mucus thin and watery.

Mucus

When CFTR proteins don't work properly, salt can't get outside the cell and water stays with it.

Without them, mucus gets thick and sticky and builds up throughout the body.

The buildup of mucus can lead to many of the problems related to CF, affecting organs such as the lungs, liver, and pancreas. We can dive into that more in another issue...

I bet your brain is full!

Let's take another look at some of the big ideas we covered!

Eugene's big ideas

- CF is caused by 2 mutations in the CFTR gene. 1 gene mutation in each copy.
- With CFTR proteins that don't work right, salt cannot get out of the cells, and water can't follow
- Without salt and water outside of the cell, thick, sticky mucus builds up throughout the body

If you or your child would like more information on anything we learned today, be sure to ask your care team.

