Eugene THE GENE

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!

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.

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. But the cause of CF is the same for everyone.

So what is the cause of CF? Let’s find out.
Each gene has two copies. CF is caused by having two mutations in the CFTR gene, one in each copy.

Thank goodness for abbreviations, right?

CFTR stands for cystic fibrosis transmembrane conductance regulator.

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

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

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

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.

What is CFTR, you ask?

Let's look it up!
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. 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.

Don’t forget to read more CF Behind the Scenes and visit CFSource.com for videos on the underlying impact of CF.