

# CF BEHIND THE SCENES WITH EUGENE

## CAUSES AND IMPACTS

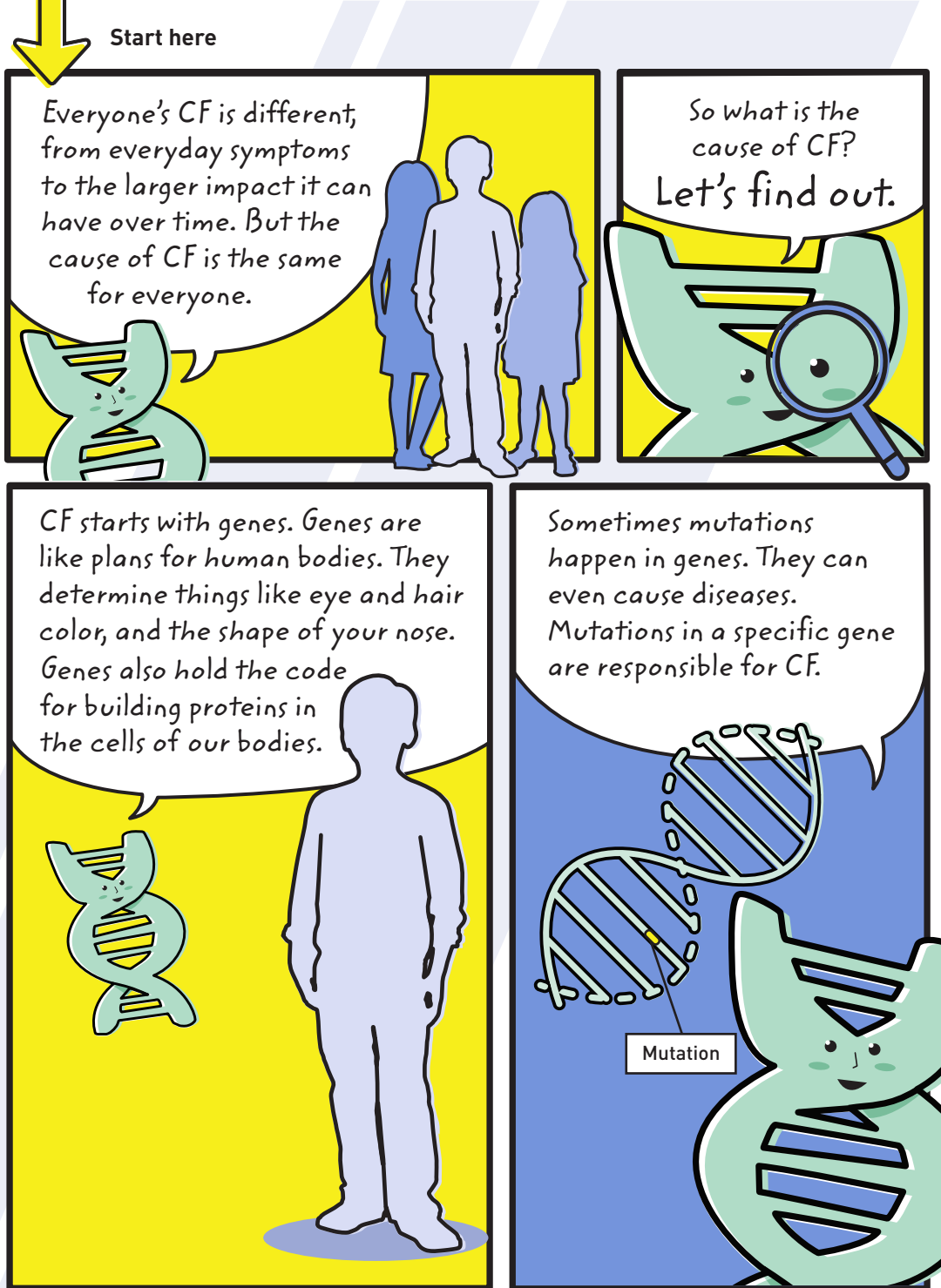
Hi there!

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.

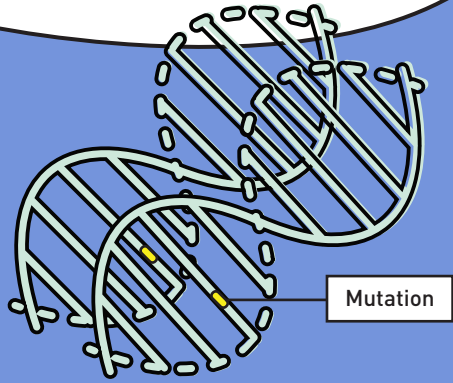
Let's go!



**Eugene  
THE GENE**



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



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



CFTR stands for cystic  
fibrosis transmembrane  
conductance regulator.



Thank goodness  
for abbreviations,  
right?

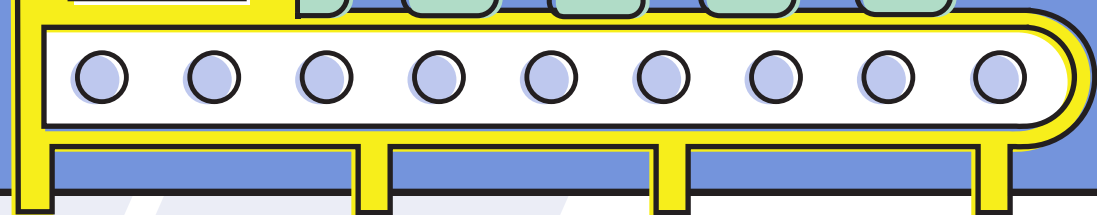
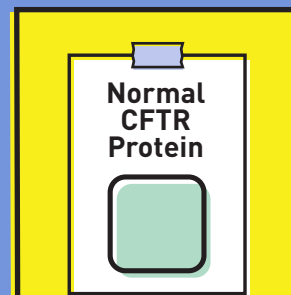
Our bodies use genes to  
build proteins, but what  
do proteins do?

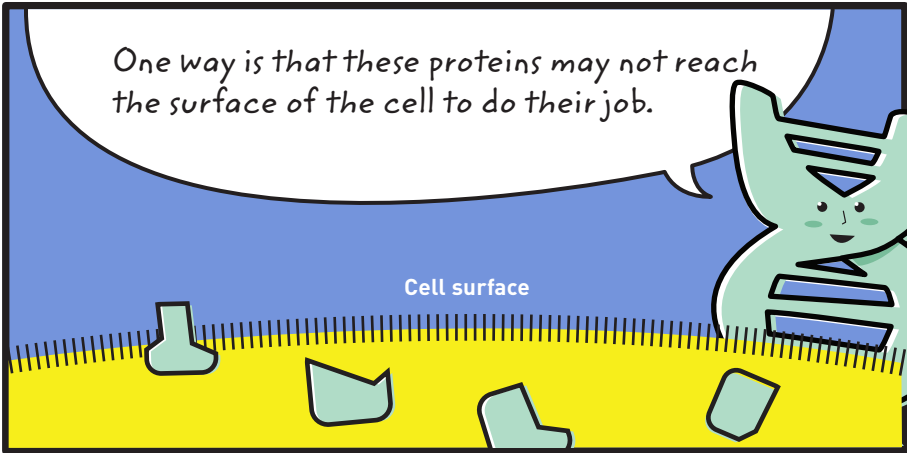
Within the cells of  
our bodies, proteins  
work to do a bunch  
of different jobs.



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

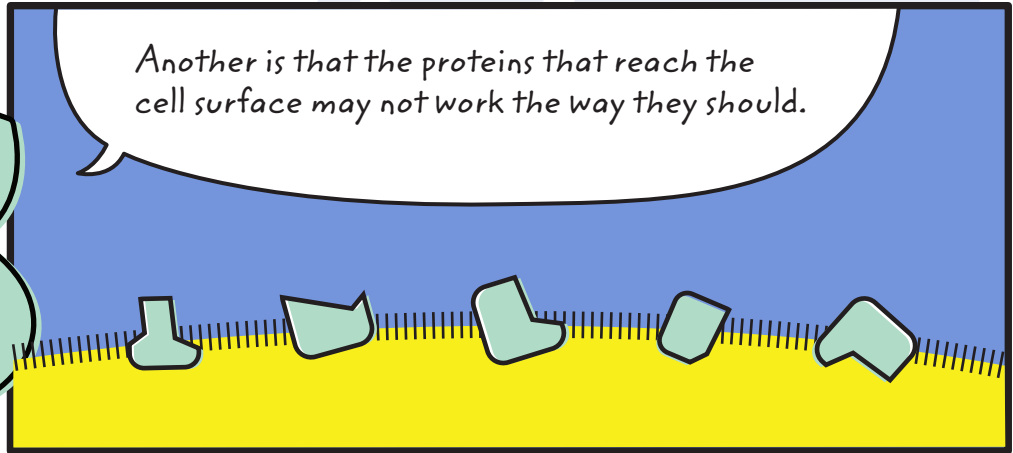
In people with CF, CFTR proteins aren't built  
correctly. This is because of mutations in  
the CFTR gene. These mutations change  
how CFTR proteins are built in  
2 different ways.



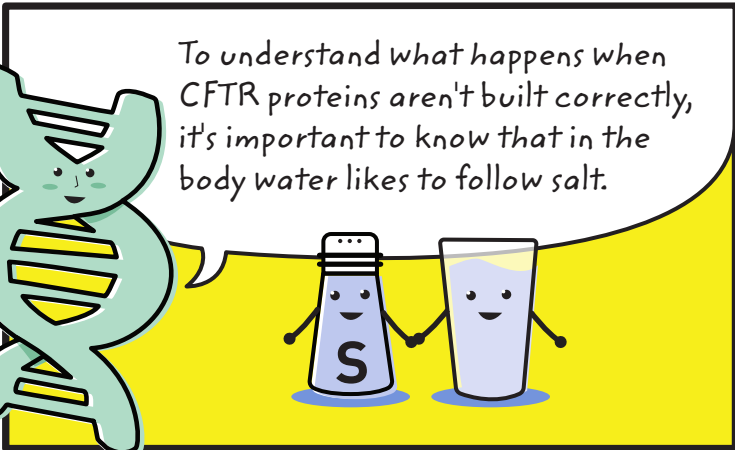


One way is that these proteins may not reach the surface of the cell to do their job.

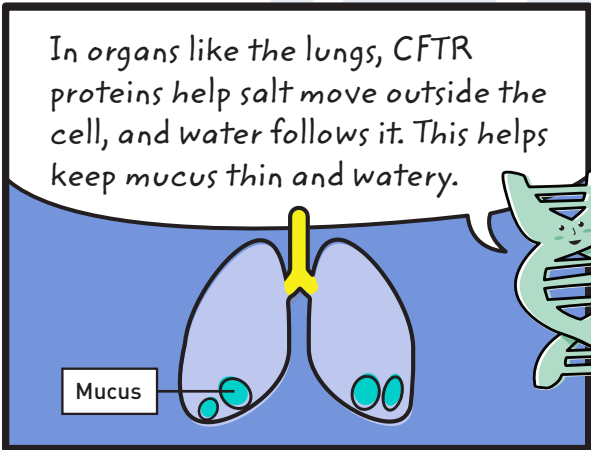
Cell surface



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

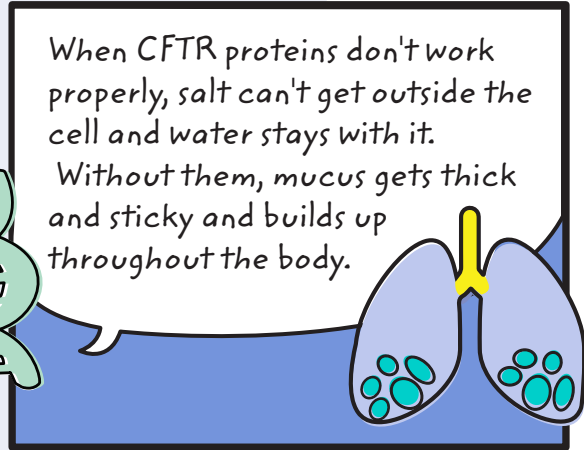


To understand what happens when CFTR proteins aren't built correctly, it's important to know that in the body water likes to follow salt.

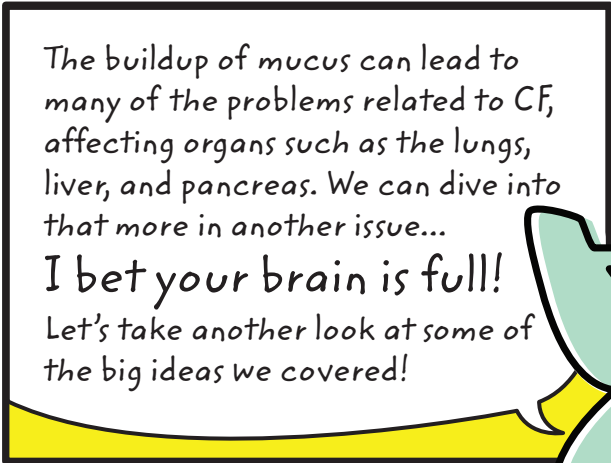


In organs like the lungs, CFTR proteins help salt move outside the cell, and water follows it. This helps keep mucus thin and watery.

Mucus



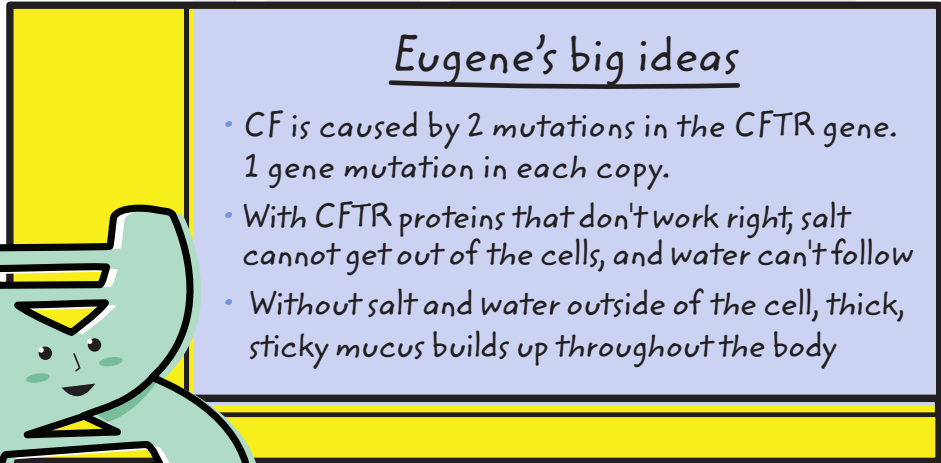
When CFTR proteins don't work properly, salt can't get outside the cell and water stays with it. Without them, mucus gets thick and sticky and builds up throughout the body.



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...

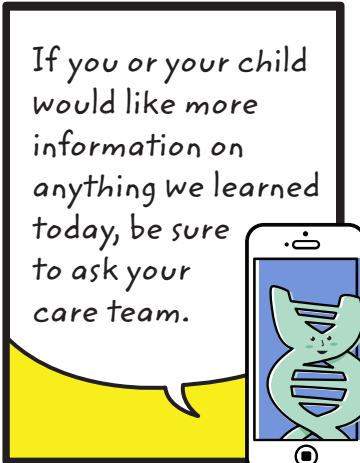
**I bet your brain is full!**

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.