



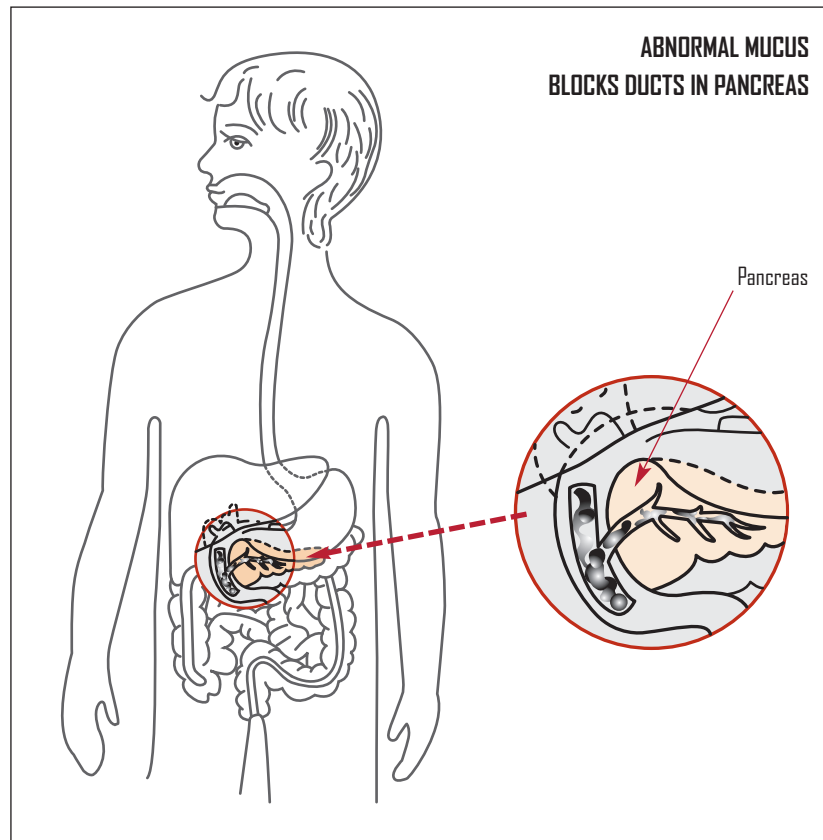
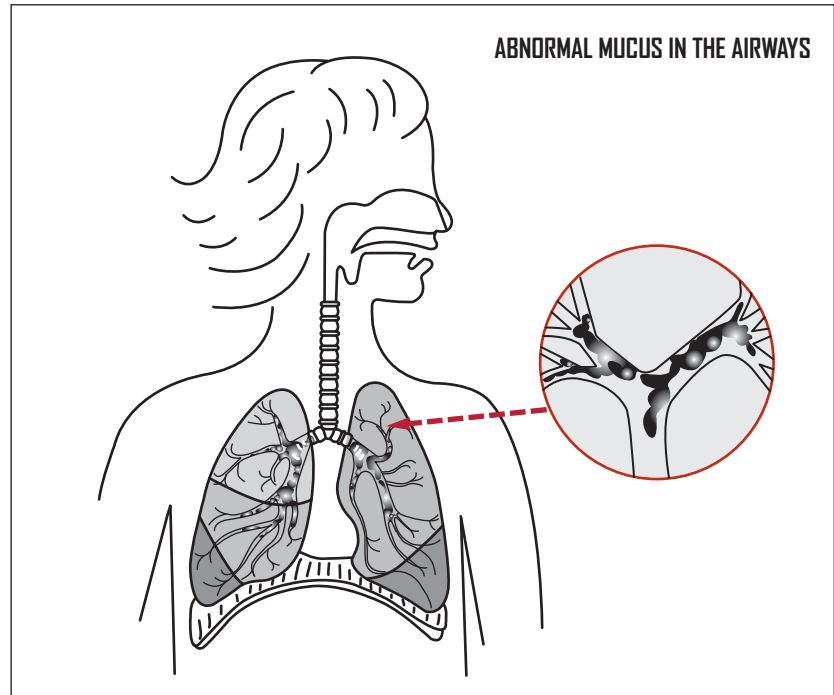
## WHAT CAUSES PROBLEMS IN CYSTIC FIBROSIS?

Doctors and researchers have known for a long time about the symptoms of **cystic fibrosis\*** (CF\*). Through research, they have begun to understand what causes these symptoms.

### ABNORMAL MUCUS

Many of the symptoms in CF are caused by abnormal (thick, sticky) **mucus\*** in the body. It is hard for the body to move this mucus.

- Abnormal mucus forms in the **airways\*** and is hard to cough up. When mucus stays in the **lungs\***, it makes a good place for **bacteria\*** to grow. The bacteria cause **lung infections\*** that over time can damage the lungs. Some CF treatments help clear mucus from the lungs.



- Mucus blocks the passages (**ducts\***) in the **pancreas\*** that connect to the **small intestine\***. The pancreas makes **enzymes\*** the body uses to **digest\*** food in the small intestine. When these passages are blocked, the enzymes cannot get to the small intestine to do their job. Without these enzymes, the body does not digest food as well. **Calories\*** from **fat\*** and **protein\*** in food are not **absorbed\*** and are lost in the stool. The person with CF cannot grow and gain weight normally. This is why most people with CF have to take **replacement enzymes\*** when they eat.

The abnormal mucus that people with CF have is the result of a faulty set of instructions being used by the cells.

\*See CF Words to Know Glossary.



## CELLS AND GENES

**Cells\*** are the very small, basic working units of all living things. The human body is made up of billions of cells. They are so small that they cannot be seen with the naked eye. Some cells make up the skin. Other cells form the heart, **liver\***, muscles, and bones. Cells are what actually do the work in the body. If the body makes something (a tear, an enzyme, a **hormone\***), it is made by certain cells. The cells even do the work to help turn food into energy the body can use.

Each cell needs instructions on what it is supposed to do and how it should do it. These instructions come from the **genes\***. Genes are simply coded messages that tell the cells how to do their jobs. Human beings have thousands of genes. Some genes decide what color eyes or hair will be. Other genes decide blood type. Every cell contains many genes. The genes are arranged on **chromosomes\***. You can learn more about genes and chromosomes in “THE GENETICS OF CYSTIC FIBROSIS” in Appendix 1.

## THE CFTR GENE

One particular gene carries the instructions for making the **CFTR\*** protein. CFTR is short for *cystic fibrosis transmembrane conductance regulator*. We all have this gene and we are all supposed to have the CFTR protein in our cells. But in people with CF, the gene is abnormal. This is called a **gene mutation\***. The instructions are different. There are several ways that the instructions are changed with an abnormal CF gene. Some people with CF have cells that do not make any CFTR protein. In some people, the CFTR protein is not made correctly or it does not end up in the right place in the cell to do its job. These differences cause the problems in CF.

## THE CFTR PROTEIN AND CF

The CFTR protein normally functions as a gate in the cell wall. The CFTR gate allows **chloride\*** to go through the cell wall. Chloride is a part of salt and is important to the body's cell chemistry. The movement of chloride in and out of the cell also affects how water and sodium go in and out of the cell. This is important in cells that produce mucus. Salt and water flow plays a role in how much mucus is made and how thick it is.

Scientists have found that abnormal CFTR protein makes a gate that does not work or works poorly. People with CF have this problem in a number of different cells in their bodies, including the cells in the **sweat glands\***, **lungs\***, pancreas, and intestine. The abnormal CFTR protein explains why people with CF have high chloride levels in their sweat (salty sweat). The high level of chloride in sweat has been known for a long time and is why the **sweat test\*** is used to help diagnose CF.

Scientists are doing research on how the CFTR protein works and how to develop ways to improve chloride movement across the cell wall. Cells can have other problems that cause symptoms in CF, but the abnormal CFTR protein is considered the primary problem in CF. Researchers continue to try to understand more about how CF affects the body and the functions of its cells. In the future, as researchers find out more about what causes CF, their discoveries can help lead to new and better treatments.