

University of Chicago CF Care Center
**CYSTIC FIBROSIS (CF)
INFORMATION SHEET**

WHAT IS CYSTIC FIBROSIS (CF)?

Cystic Fibrosis (CF) is an inherited disease that causes certain glands in the body to not work properly. The glands that are involved are called exocrine glands. The exocrine glands usually make very thin “liquidy” secretions like sweat, tears, saliva, digestive juices and mucus. The exocrine glands squirt these secretions into or onto many organs such as the skin, intestines and the airways. In CF there are problems with the salt and water balance in these secretions which makes them very sticky. Since the secretions are so sticky, they are not able to flow easily. This causes plugging up of the glands, airways and intestines. This makes it very difficult for the lungs and intestines to work like they should. In the lungs this can lead to inflammation (swelling) and infection. In the gastrointestinal system this can lead to poor absorption of nutrients, poor digestion and intestinal blockage.

WHAT BODY ORGANS ARE AFFECTED IN CF?

Many organs can be involved in CF. They include: the respiratory system including the lungs, nose, and sinuses, the digestive system including the intestines, pancreas, liver, the reproductive system and the sweat glands.

WHAT CAUSES CF?

CF is caused by mutations or errors in the gene that makes the Cystic Fibrosis Transmembrane Regulator (CFTR) protein. Genes are the blueprints our body uses to make proteins. These proteins are the building blocks for all of our cells and organs. If there are mistakes or errors in our genes, there can be problems in the proteins they make.

Our genes are made up of building blocks called base pairs. Sometimes there are changes in the base pairs or a loss of some base pairs from a gene. Such mistakes are called mutations. A mutation in a gene will lead to defects in the protein it makes. In CF there are over 1,000 mistakes or mutations in the CFTR gene that have been identified and can lead to CF. The most common mutation in the CFTR gene is called delta F508.

HOW IS CF INHERITED?

In order to have CF, a person must inherit two CFTR genes that have mutations (one from each parent). If you only have one CFTR gene with a mutation and your other CFTR gene is normal, you will not have any symptoms or disease. However, you will be a carrier of CF.

WHAT ARE THE HEALTH PROBLEMS SEEN IN CF?

People who have CF usually develop sinus problems, chronic lung disease, increased salt loss from sweating, and digestive problems (pancreatic insufficiency). In addition a number of people will develop liver disease, diabetes, intestinal blockages and problems with fertility.

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