Adult CF: Understanding Cystic Fibrosis

Cystic fibrosis (CF) is a chronic, genetic disorder. You were born with it, even if you have only recently started to have symptoms. There is no cure for CF. It gets worse over time.

CF affects the secretory glands in your body. These are the glands that make mucus and sweat. CF causes secretions like mucus, tears, sweat, saliva, and digestive juices to become thicker. They then don't flow as freely.

In the lungs, thickened mucus can trap bacteria. This can lead to serious infections. In the digestive system, the enzymes that break down food may be blocked. Without them, the intestines can't fully absorb fats and proteins. CF can lead to vitamin deficiency and malnutrition.

CF may also affect the level of chloride in your body. Chloride is found in your sweat. The sweat chloride level is high in people with CF. Your body needs a certain amount of salt. Salt contains chloride, sodium, and other minerals. With CF, you may lose a lot of salt when you sweat. This can cause an imbalance. It can lead to health problems, such as dehydration, heat stroke, and an irregular heartbeat.

What causes cystic fibrosis?

CF stems from a change (mutation) in the CFTR gene. CFTR stands for cystic fibrosis transmembrane conductance regulator. This genetic mutation causes a problem with the CFTR protein. This protein controls the flow of water and certain salts in and out of the body's cells. In a person with CF, this protein doesn't work right. So, mucus and other secretions become thickened.

The CFTR gene can have many problems that lead to CF. The type of problem may play a role in how mild or severe your symptoms are.

Symptoms of cystic fibrosis

CF may affect many systems in the body. These include those for breathing, digestion, and reproduction. So the disease can cause a lot of different symptoms. The symptoms can range from mild to severe. They often get worse over time.

These are some of the most common respiratory symptoms:

- Thick mucus in lungs
- Frequent coughing, wheezing, or shortness of breath
- Coughing of thick, sometimes bloody mucus
- Chronic lung or sinus infections
- Fleshy growths in the nose (nasal polyps)

Some common digestive symptoms are:

- Intestinal blockages. These are found in babies soon after birth.
- Chronic diarrhea with greasy, foul-smelling, large stools
- Belly (abdominal) cramping or pain
- Being underweight
- Swelling (inflammation) in the pancreas (pancreatitis)

You may also have these other symptoms:

- Salty skin
- Enlarged ends of fingers and toes (clubbing)
- Problems getting pregnant (infertility)

People with CF also tend to develop health problems, such as diabetes, liver disease, and osteoporosis.

How is cystic fibrosis diagnosed?

CF is often diagnosed in children by age 2. Newborns are now routinely screened for the disease. But such testing was not needed in all U.S. states until 2010. As a result, CF may not be found until adulthood. Sometimes its symptoms may be mistaken for other health problems, such as asthma.

Your healthcare provider will diagnose CF based on your symptoms and certain tests. You'll first need a blood test to screen for CF. It may find problems in the CFTR gene. If this test is positive, you'll then have a sweat chloride test. The sweat chloride test measures the amount of chloride. Chloride is found in salt, in your sweat. People with CF tend to have high levels of chloride in their sweat.

These tests may not always be accurate. Plus, some people may have a rare genetic defect not found through regular testing. So, you may need other tests, such as:

- Blood tests to measure levels of vitamins and minerals or to check liver function
- Chest X-rays to view the lungs
- Lung (pulmonary) function tests to check breathing
- Sputum cultures to check for lung infection
- Stool assessments to see how your body absorbs fat
- DNA tests to confirm diagnosis, if needed

How is cystic fibrosis managed?

There is no cure for CF. The goal of treatment is to control symptoms and slow the progress of the disease. To manage CF, you will likely need:

- Medicine to prevent or treat infections, reduce swelling, open airways, or thin mucus
- Pancreatic enzymes to help you absorb nutrients
- Certain vitamins
- Changes in diet, such as adding extra calories, salts, and fluids
- Exercise
- Airway clearance techniques. These include chest physical therapy, special ways of breathing and coughing, and using mouth devices and therapy vests. They help loosen and clear mucus. This lowers your risk of infection and helps you breathe better.

Your healthcare team will include social workers, respiratory therapists, psychologists, physical therapists, pharmacists, nurses, genetic counselors, cystic fibrosis specialist, nutritionists and dietitians, and healthcare providers who specialize in diabetes, the digestive system, and the lungs.

Some people are given CFTR modulator therapies. These are only for people with very specific gene changes. For severe or advanced conditions, you may need surgery. One kind of surgery for cystic fibrosis is a lung transplant. This addresses the lung disease but not the effects on other parts of the body.

Living with CF

Improvements in treatment have extended the lives of people with CF. Many people with CF can now expect to live into their 30s to 40s, and beyond. You will need regular healthcare for life. You will need to stay in touch with all your healthcare providers. You may be able to use the services of a cystic fibrosis center, where there are healthcare providers trained in treating this disease.

As with many chronic diseases, CF can seem to take over your life and that of your family. Make sure to get support to help you cope. Ask your healthcare team or check the Internet for CF support groups. And especially at the beginning, focus on getting through one day at a time. Talk with a professional counselor about ways to help you and your family manage the financial, emotional, and medical issues that can go along with this disease.