UNDERSTANDING CYSTIC FIBROSIS

INTRODUCTION

Cystic fibrosis is a chronic disease that affects the respiratory and gastrointestinal systems. People with cystic fibrosis have a genetic defect of the lungs and the gut and they suffer from serious respiratory and gastrointestinal signs, symptoms, and complications. There is no cure for cystic fibrosis, but medications and lifestyle adjustments can help prevent complications and make this disease easier to live with. However, cystic fibrosis is a very serious disease, and patients often require a lot of care and support.

Learning Break: The name cystic fibrosis was given to this disease many years ago because people with cystic fibrosis have cysts (a small closed sac) in the pancreas and fibrosis in the lungs.

OBJECTIVES

After finishing this module, the learner will be able to:

1. Identify the cause of cystic fibrosis.
2. Identify people at risk for developing cystic fibrosis.
3. Identify the two organ systems affected by cystic fibrosis.
4. Identify common signs, symptoms, and complications of cystic fibrosis.
5. Identify the most common cause of death from cystic fibrosis.
6. Identify how cystic fibrosis is diagnosed.
7. Identify the goals for treatment of cystic fibrosis.
8. Identify surgical options that can be used to treat cystic fibrosis.
9. Identify non-pharmacological options used to treat cystic fibrosis.
10. Identify life style options used to treat cystic fibrosis.

WHAT CAUSES CYSTIC FIBROSIS?

Cystic fibrosis is a genetic disease; the tendency to develop cystic fibrosis is inherited. Some people who have the gene that can cause cystic fibrosis only have one copy of this gene, but if someone has two - if a child inherits a copy of the gene from each parent - then that person will develop cystic fibrosis.

The gene involved in cystic fibrosis is responsible for the production of mucous in the lungs and the gastrointestinal tract. Mucous is a naturally occurring compound that helps keep the lungs warm and moist and acts as protective barrier in the stomach and the intestines. For most people, mucous is relatively thin and watery. But in cystic fibrosis, the gene that regulates mucous production is abnormal and people who have this disease produce mucous that is unusually thick, viscous, and sticky. The abnormal mucous can block the bronchi and other air passages in the lungs, and it can also block the gastrointestinal tract in various places, e.g., the gall bladder, the bowels, and the pancreas. This thick, sticky mucous also is an excellent place for bacteria to grow, and because of
that people who suffer from cystic fibrosis often have chronic infections and inflammation of the lungs and the gastrointestinal tract.

**Learning Break:** Genes are the part of a cell that contain the information that determines how cells reproduce and what their structure and functions will be. In cystic fibrosis, the genes involved control the cells that produce mucous, but the genetic information is faulty and the mucous that is produced, as mentioned above, is thick and viscous. Cystic fibrosis is an autosomal recessive disorder. That means that in order for these abnormal genes to express themselves and to cause the mucous-producing cells to make abnormal mucous, there must be two of these genes present. Genes are found in pairs and for a disease such as cystic fibrosis to occur, both of the genes that control the production of mucous must be abnormal.

**RISK FACTORS: WHO GETS CYSTIC FIBROSIS**

Cystic fibrosis is the most common lethal inherited disease in the white population. In the United States, cystic fibrosis affects approximately 1 of 3500 Caucasians, 1 of 9500 Hispanic Americans, 1 of 17,000 African Americans, and 1 of 31,000 Asian Americans. Males and females are equally affected, but females who have cystic fibrosis have more severe lung problems and die at a younger age. *The signs and symptoms of cystic fibrosis usually begin in childhood*, but in about 5% of people who have cystic fibrosis, the disease developed when they were adults.

**SIGNS, SYMPTOMS AND COMPLICATIONS OF CYSTIC FIBROSIS**

Patients who have cystic fibrosis have respiratory and gastrointestinal signs, symptoms, and complications. In most cases, these develop during infancy or childhood.

- **Respiratory:** The typical respiratory signs and symptoms of cystic fibrosis are *cough*, *difficulty breathing* at rest or during physical activity, and *wheezing*. The excessive mucous production prevents oxygen from moving through the lungs, so people who have cystic fibrosis often feel short of breath. The excessive - and excessively thick - mucous is very irritating to the respiratory passages and in response, people with cystic fibrosis develop a persistent cough to try and clear the secretions. And the thick, sticky mucous also is an excellent place for bacteria to grow, and *people who suffer from cystic fibrosis often have chronic infections and inflammation of the lungs*. The great majority of people who die from cystic fibrosis die from a lung infection.

**Learning Break:** In the normal lungs, mucous gets moved out by the cilia. The cilia are tiny fibers that move in a wave-like fashion and push mucous in the respiratory passages up and out towards the mouth. However, because the mucous in the lungs of someone with cystic fibrosis is so thick, it cannot be easily moved by the cilia and much of it stays trapped in the lungs.
• Gastrointestinal: People who have cystic fibrosis typically suffer from abdominal cramps, constipation, obstructions, nutritional deficiencies and weight loss. These signs and symptoms happen for two reasons: a) the excess, thick mucous in the digestive tract can block the bowels, it can block the bile ducts of the gallbladder, and it can block the drainage ducts of the pancreas, and b) the mucous provides an environment that allows bacteria to grow and the gastrointestinal tract and organs can become inflamed. Because cystic fibrosis prevents the gallbladder and the pancreas from secreting bile and digestive enzymes into the gut, people with cystic fibrosis often have nutritional deficiencies: in particular they are at risk for nutritional deficiencies associated with the fat-soluble vitamins A, D, E, and K. They also are at a greater risk for developing diabetes because blockage of the drainage ducts of the pancreas can cause permanent damage to this organ.

**Learning Break:** The gallbladder produces bile: bile is a compound that is secreted into the bowels and is necessary for the absorption and digestion of fats and fat soluble vitamins. The pancreas is a gland that produces insulin and also several digestive enzymes that are necessary for the breakdown and absorption of nutrients.

Other complications of cystic fibrosis are infertility (men and women), osteoporosis (this is associated with poor absorption of nutrients and minerals), and fluid and electrolyte imbalances.

Not all cases of cystic fibrosis are severe. There are variations in the presentation and some people who have cystic fibrosis have a mild form of the disease. But cystic fibrosis is, for most people, a very serious disease. Advances in treatments have improved the outlook - more than half the people with cystic fibrosis are 18 years or older - but 20% of the people who have cystic fibrosis do not live to adulthood and the median age of survival is only 37 years.

Cystic fibrosis is diagnosed using the following three methods:

• Genetic testing: Newborns undergo genetic testing for several inherited diseases, including cystic fibrosis. A blood sample is obtained soon after birth and examined for the genetic abnormality that causes cystic fibrosis.

• Sweat test: People with cystic fibrosis produce sweat that has a very high sodium (salt) content. If the initial test shows high sodium content, the test is repeated several weeks later for confirmation.

• Examining the patient for the presence of respiratory and gastrointestinal signs, symptoms, and complications that are typical of cystic fibrosis.
TREATMENT

There is no cure for cystic fibrosis, and living with this disease is very difficult. The three main goals of treating cystic fibrosis are a) maintain healthy lung function, b) maintain good nutrition, and c) manage complications. All of these are important but maintaining healthy lung function is the most important. In order to meet these goals, medications, diet and exercise, physical therapy (PT), and surgical treatments can all be used.

- Medications: Medications are not used to cure cystic fibrosis; they are used to manage and to prevent specific complications. For example, patients who have cystic fibrosis often have lung infections, and these are treated (or prevented) with antibiotics. The antibiotics can be given orally, intravenously, or by using an inhaler. Bronchodilators such as albuterol that are delivered by an inhaler can open the respiratory passages and help patients breathe, and inhaled mucous thinning drugs such as dornase alfa (Pulmozyme®) can help break up thick secretions. Many patients also use an inhaler with a sterile saline solution; this helps to liquefy the thick, sticky mucous in the lungs. Patients with nutritional deficiencies will be given fat-soluble vitamins A, D, E, and K, and pancreatic digestive enzymes are given as well. Both of these will help with digestion and absorption of nutrients.

- Diet and exercise: Smoking cessation is obviously very important if someone has cystic fibrosis. Exercise can help boost the function of the immune system, and patients should be encouraged to exercise within their tolerance levels. Patients with cystic fibrosis do not need a highly specialized or restricted diet. However, because some nutrients - especially the fat-soluble vitamins A, D, E, and K - are not well absorbed, patients who have cystic fibrosis should have a diet that with extra fat and extra fat-soluble vitamins. Good hydration is very important; a patient with cystic fibrosis should be encouraged to drink lots of fluids.

- Physical therapy: Physical therapy can help clear secretions from the lungs. Physical therapy techniques that are used include postural drainage, specialized breathing exercises, and vibration and percussion.

Learning Break: When performing percussion and vibration, the physical therapist will use cupped hands and gently, rhythmically clap her/his hands against the patient’s chest. After percussion, the physical therapist will place his/her hands flat against the chest wall and vibrate them. These techniques can be helpful in breaking up and moving excess mucous in the lungs.

- Surgical treatments: For some people who have cystic fibrosis, a lung transplant is necessary. If they are producing large amounts of mucous that cannot be managed with medications, diet, or exercise, and/or if they are having persistent lung infections that do not respond to antibiotics, a lung transplant may be the only
treatment that can help. If the patient’s gastrointestinal tract is compromised, a feeding tube may need to be surgically placed.

CARING FOR A PATIENT WHO HAS CYSTIC FIBROSIS

Caring for a patient who has cystic fibrosis can be very challenging. It can also be very rewarding, as well, because your intervention can make a big difference. The patient who has cystic fibrosis needs skilled care and psychological support to successfully cope with this illness, and as a health care professional you can monitor the patient’s condition and provide encouragement.

When you are caring for a patient who has cystic fibrosis, focus on these four areas of care.

- Infection control: The patient who has cystic fibrosis is very susceptible to developing respiratory tract infections, so hygiene is very important. Good hand washing is one of the most important and effective ways to prevent the spread of infections, and it is especially critical when you are caring for a patient who has cystic fibrosis. If you have an infectious disease - and especially if you have a respiratory infection - you should not be caring for a patient who has cystic fibrosis.

- Monitor respiratory status: Remember, patients who have cystic fibrosis are very susceptible to developing respiratory tract infections, and respiratory tract infections are the major cause of death for these patients. You will need to perform a basic assessment of the patient’s respiratory status to make sure they are breathing normally: check the respiratory rate, check skin color and most importantly, ask the patient if she/he is short of breath. The respiratory rate and the skin color may be normal, but if the patient is having difficulty breathing that information is more important than the physical assessment.

- Hydration and nutrition: Good hydration and good nutrition are essential for patients who have cystic fibrosis. Dehydration will cause the mucous in the lungs and the gastrointestinal tract to become thicker and more viscous and this can lead to complications, and nutritional deficiencies can easily occur. The patient’s physician will establish guidelines for fluid and nutrient intake. Familiarize yourself with these guidelines, monitor the patient’s fluid and nutrient intake, and report any problems in these areas to a nurse or your supervisor.

- Monitor for complications: Patients who have cystic fibrosis are at risk for respiratory and/or gastrointestinal complications. These complications will usually present as infections, difficulty breathing, or gastrointestinal complaints. If the patient has a fever, difficulty breathing, or a problem with his/her stomach of bowels, notify a nurse or your supervisor.