Cystic Fibrosis

Cystic fibrosis (CF)is an inherited disease caused by a defective gene. This defective gene leads to a change in a protein that affects cells that make mucus and sweat. Mucus is usually thin and watery. It is needed to keep the body working properly. But in CF, mucus becomes thick and very sticky. It clogs up the glands, airways and other passageways in the body. This makes it difficult to breathe freely. It also makes lung infections more likely. And, it can make it hard for the body to get the fat and protein it needs from food.

CF is often thought of as a lung disease. Breathing (*respiratory*) failure is its most serious consequence. But, in CF, other parts of the body can also be affected including the skin, pancreas, liver, intestines, sinuses and sex organs.

The signs and symptoms of CF vary from person to person. They also differ by age. CF is a lifelong disease. However, people can live long, productive lives with care and treatment.

CAUSES

CF is a genetic disease. CF is inherited as a recessive trait. That means a child can have CF even if neither of the child's parents have the disease. Both parents simply carry a gene that can cause CF.

You are at greater risk for CF if:

- One of your brothers or sisters has the disease.
- Your parents carry the gene that causes it.
- One of your parents has the disease.

SYMPTOMS

- Difficulty gaining weight.
- Not growing as big as other children the same age.
- Chronic gas or bloating.
- Bulky or greasy bowel movements.
- Tissue from the rectum protrudes from the anal opening (*rectal prolapse*).
- Inflammation of the pancreas (*recurrent pancreatitis*).
- Prolonged yellowing of the skin (*jaundice*).
- Unusually salty sweat or skin.
- Heat exhaustion with low salt in the blood.
- Chronic coughing and/or wheezing.
- Shortness of breath.
- Frequent lung infections (*bronchiolitis, bronchitis, pneumonia*).
- Frequent sinusitis and/or *nasal polyps* (small growths in the nose).
- Club-shaped fingers or toes.

DIAGNOSIS

A caregiver will probably ask many questions and order some tests. These will help determine if CF is present. They also will provide valuable information about the nature and extent of the disease. Questions and tests may include:

- Family history. You will be asked whether any relatives have CF.
- Symptoms. You will be asked whether any of the symptoms listed above are present.
- Sweat test. This is the standard test used to make a diagnosis of CF. It is also called a sweat chloride test. A chemical will be applied to make a patch of skin sweat. Then the sweat will be analyzed to see if it contains the chemicals present in CF.
- DNA testing. This can be used to find what genetic mutations in CFTR are present.

- Newborn screening. A blood test measures a chemical in the blood that is often elevated in newborns with CF. Extra testing is done if a blood test is abnormal or if a new baby shows signs of CF.
- Nasal potential difference measurement. This test shows how CF is affecting the lining of the body's upper breathing passages.

Other tests that will be performed after a diagnosis of CF is made:
- Blood tests to evaluate salt balance, liver function, vitamin levels and blood counts.
- X-rays. Chest and/or sinus x-rays can show how the lungs and breathing system are affected by the disease.
- Lung (*pulmonary*) function tests. They tell how well the lungs are working.
- Sputum culture. This test analyzes mucus from the lungs to find out if there is an infection.
- Stool testing. This is done to see if the pancreas is working normally.
- Fertility testing. In males, an ultrasound scan of the testes might be done.

COMPLICATIONS

CF can lead to other problems. They might include:
- Frequent lung infections. These include pneumonia and bronchitis.
- Frequent sinus infections (*sinusitis*).
- Coughing up blood (*hemoptysis*).
- Collapsed lung (*pneumothorax*). A small hole develops in the lung, and air leaks out.
- Malnutrition. This can occur because the disease prevents the body from digesting food properly and absorbing needed nutrients.
- Delayed growth. This is caused by malnutrition.
- Nasal polyps.
- Thick mucus can block the ducts into and out of the pancreas (*pancreatitis*). This causes the pancreas to become inflamed.
- Diabetes. This develops if thick mucus keeps the pancreas from working properly. The pancreas controls the level of sugar in the blood.
- Gallstones.
- Liver damage. Thick mucus can block the duct that carries *bile* (a digestive fluid) out of the liver to be expelled from the body.
- *Rectal prolapse* (the inner lining of the rectum pushes outside the body). Excessive coughing can cause this.
- Fertility problems. In men, the tube that connects sexual organs can become clogged by mucus. Also, the *vas deferens* (a tube that moves sperm through the testes) may be missing in men with CF. Women with CF are sometimes less fertile than those without the disease.
- Development of weak, brittle bones (*osteoporosis*). This occurs when the body does not absorb the vitamins needed to keep bones strong.

TREATMENT

Patients with CF should be managed through an accredited CF Center (to find a CF Center near you go to the Cystic Fibrosis Foundation website at www.CFF.org).

Treatment may include:
- A high calorie diet. This can help maintain a healthy weight. That, in turn, helps reduce coughing and breathing problems.
- A high salt diet or salt supplements. The body needs a balance of salt and other chemicals.
- Drinking lots of fluids.
- Exercise.
- Enzyme capsules to aid digestion.
- Nutritional supplements.
- Medications to help with breathing.
- Medications to thin the mucus.

- Vaccines to prevent infections.
- Insulin medication, if diabetes is present.
- Antibiotics to treat infections.
- Chest percussion ("thumping") treatments to help move mucus.
- Oxygen.
- Surgery:
  - To relieve a digestive system blockage.
  - To remove nasal polyps.
- Lung or liver transplants.

At times, people with CF need to be given some treatments in a hospital.

HOME CARE INSTRUCTIONS

To get started with home care:

- Learn how to give all medicines that have been prescribed.
- Learn which symptoms require medical attention.
- Learn how to give treatments to clear mucus.
- Learn techniques to improve breathing.
- Learn how to prepare the right kind of foods.
- Join a support group for people with CF. Groups also exist for family members.
- Do not smoke and keep yourself and your child away from smoke.
- Be as physically active as possible.
- Keep shots (*immunizations/vaccinations*) up-to-date.
- Wash hands often. This will reduce the chance of infection.

SEEK MEDICAL CARE IF:

- Mucus contains blood.
- There is a new cough or change in your cough.
- The amount or consistency of mucus changes.
- It becomes more difficult to breathe.
- You or your child does not feel like eating or is losing weight.
- You or your child has no energy.
- You or your child has severe constipation.
- You or your child has severe diarrhea.
- You or your child is throwing up dark green fluid.
- You or your child has an oral temperature above 102° F (38.9° C).
- Your baby is older than 3 months with a rectal temperature of 100.5º F (38.1° C) or higher for more than 1 day.

SEEK IMMEDIATE MEDICAL CARE IF:

- You or your child has an oral temperature above 102° F (38.9° C), not controlled by medicine.
- Your baby is older than 3 months with a rectal temperature of 102º F (38.9º C) or higher.
- **Your baby is 3 months old or younger with a rectal temperature of 100.4º F (38º C) or higher.**

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