Cystic Fibrosis

Cystic fibrosis (CF) is an inherited disease that mainly causes problems with the lungs and gastrointestinal system. The genetic defect causes the mucus to become thick, leading to blockage of the airways and respiratory infections. Children with CF also have problems with digesting and absorbing enough nutrients to grow properly. There is no cure, but medical advances have greatly improved the length and quality of life for patients with CF.

What is cystic fibrosis (CF)?

CF is the most common cause of severe, chronic (ongoing) lung disease in children. Children with CF have an abnormal gene that affects the cells producing mucus and other fluids in various parts of the body, especially the lungs and pancreas. The mucus of children with CF is very thick, sticky, and difficult to clear; this causes problems with plugging and blockage of the airway and lungs. Lung infections are a frequent and long-term problem for most children with CF and are the most common cause of death.

CF also causes abnormalities of the digestive organs, especially the pancreas—an organ in the abdomen that makes enzymes that help with digestion. Children with CF don't make enough of these enzymes to digest and absorb certain foods and minerals (called "malabsorption"). CF can also affect the liver and other organs.

In the past, patients with CF usually died in childhood. Today, with good medical care, children with CF can live well into adulthood. The results are best when families work in close partnership with doctors, nurses, and other health professionals.

What does it look like?

The symptoms of CF vary, depending on the gene causing the disease and other factors. Symptoms can begin at different ages—sometimes in the first few months of life, sometimes later. Some children have severe lung disease and digestive problems, others have fewer symptoms.

- *Respiratory symptoms* (breathing-related) include:
  - Cough. Coughing occurs as the body tries to eliminate thick, sticky mucus. Cough may be worse in the morning or after your child has been active.
  - Repeated respiratory infections, including pneumonia (lung infection), bronchitis (infection of the airways), and sinusitis (infection of the sinuses). Children with CF may need many hospital stays to treat these infections.
  - Wheezing (noisy breathing) and asthma-like symptoms may develop.
  - As the lung disease of CF gets worse, your child may have low energy and shortness of breath.
  - With time, the chest can become "barrel-shaped." The fingertips may appear swollen—this is called "clubbing."
- *Digestive symptoms*:
  - Some CF babies are born with intestinal obstruction (blockage), so stools can't pass out of the body. The blockage occurs when the baby's first stool, called meconium, doesn't pass. Other symptoms include bloating of the abdomen and nausea.
  - Abnormal function of the pancreas leads to poor digestion, with poor growth and weight gain. Your child may have frequent, foul-smelling BMs. The stools may float because they contain large amounts of fat.
- *Other symptoms*:
  - Children with CF lose too much salt in their sweat; their skin may even "taste" salty. This can lead to problems with low sodium levels (hyponatremia), especially during times of warm weather or illness.
  - Other problems may develop later in childhood, including diabetes and delayed sexual development.
  - Many other medical problems are possible, including cirrhosis (scarring and damage) of the liver.

How is cystic fibrosis diagnosed?

The doctor may first suspect CF because of typical symptoms or a family history of CF. In some states, all newborns are tested for CF as part of newborn screening for many diseases.

Several tests may be done to confirm that CF is present:

- *Sweat testing.* This is the best test to make the diagnosis. A machine is used to produce and collect a sample of sweat (usually from the arm) and measures the level of chloride. The sweat of children with CF contains increased levels of chloride.
- *Genetic tests* can be done to look for the defective gene causing CF. These tests detect 90% of patients with the disease. They are usually done if the child is too little (such as newborns) to obtain enough sweat for sweat testing, or to identify carriers, that is, people who have the abnormal CF gene but don't have the disease.

What causes cystic fibrosis?

One of many different gene abnormalities (mutations) occurs. Which specific mutation your child inherits has a major impact on the severity of CF.

Your child has CF because he or she inherited a copy of the mutated gene from *both parents.* People who have just one copy of a mutated gene do not develop CF; they are *carriers* of the CF gene.

Genetic testing is done to identify the gene mutation causing your child's CF. Parents and other family members need to be tested as well. Genetic counseling can help you to understand the risk of passing CF on to future children.

What are some possible complications of cystic fibrosis?

- CF progresses gradually over time. Most people with CF eventually die of complications related to the lungs and heart.
- Respiratory complications include pneumothorax (air leaking out of the lungs into the chest) and bleeding into the lungs and airways.
- Digestive problems and malabsorption can lead to vitamin deficiencies.
- Diabetes mellitus may occur if the pancreas does not make enough insulin. Some patients with CF develop diabetes as they get older.
- Fertility problems may occur, especially in males.
- Other complications are possible as well. Prevention, early identification, and treatment of complications are major goals of treatment for CF.

What increases your child's risk of cystic fibrosis?

Inheriting a gene mutation from both parents is the only cause of CF. These genes are most common in families of central or northern European origin. In the United States, CF is much more common in whites (1 in 3500 infants) than in African Americans (1 in 17,000).

If your family has a history of CF, genetic testing and counseling may help you understand your risk of passing the abnormal gene on to your children.

How is cystic fibrosis treated?

Your child will most likely be referred to a pulmonologist, an expert in lung diseases. Your child may see other specialists as well, including a gastroenterologist, an expert in diseases of the digestive system, and a nutritionist or dietitian.

A comprehensive plan will be developed to manage your child's CF. He or she will receive regular medical follow-up. The goal is to keep your child in stable condition for as long a time as possible.

Treatment of respiratory/lung problems includes:

- *Percussion and postural drainage.* Your child's body needs help to clear thick, sticky mucus more effectively. Postural drainage means positioning your child's body to help secretions drain. Chest percussion means tapping on the chest to help loosen secretions. These steps help your child get the secretions out of the body by coughing. They are among the most important things you can do to keep your child's lungs working as well as possible.
- *Aerosolized saline solution.* Using a device called a nebulizer, your child breathes in a fine mist of salt water. This helps to thin the mucus so that it can be better removed by coughing.
- *Antibiotics* are used to control infections. Sometimes they are given through a vein (intravenously, or IV), sometimes orally, sometimes inhaled.
- *Human recombinant DNAse (Pulmozyme).* This is an enzyme that helps thin and clear mucus from the airway. It is inhaled using an inhaler or nebulizer.
- *Bronchodilators.* Commonly used for asthma, drugs called "bronchodilators" can help to reduce airway blockage for some children with CF. They are especially useful for patients with asthma-like symptoms such as wheezing, which may be called "reactive airway disease." These drugs, such as albuterol, are given through an inhaler or nebulizer.
- *Steroids.* Oral steroids may be used for some severe lung problems. Inhaled steroids are often used for patients with asthma-like symptoms.

Other treatments may include:

- Macrolide antibiotics: a type of antibiotic that also helps reduce inflammation.
- Ibuprofen is commonly used, also because of its effect on inflammation.
- Oxygen may be needed, especially during sleep.
- Lung transplantation is an option for CF patients with very severe lung disease.
- *Nutritional therapy.* Without treatment, most CF patients have severe problems digesting fats, proteins, vitamins, and minerals. Treatment may include:
  - Pancreatic enzymes to replace those not produced normally.
  - Special diets to increase calories. A nutritionist or dietitian can help in planning a diet.
  - Other methods may be tried to get more food and calories into the child. These may include placing a feeding tube into the stomach or through the nose (nasogastric tube) or through the skin of the abdomen. Your child may receive hyperalimentation, that is, food in the form of proteins, fats, and sugars, along with vitamins and minerals, given through a vein (IV).
  - Treatments may be needed for other digestive problems, including obstruction of the intestines and liver disease.
- *Mental health care* and support are also important. Having a child with such a serious disease can be overwhelming for parents. Several national and local organizations are

available to provide information and support for children with CF and their families.

When should I call your office?

Your child with CF will receive specialized team care, with frequent health visits and monitoring. Regular medical follow-up is essential to keep your child in the best possible health and avoid complications. Between appointments, contact your treatment team or doctor immediately if any problems occur.

Where can I get more information about cystic fibrosis?

The Cystic Fibrosis Foundation is on the Internet at *www.cff.org* or call 1-800-FIGHT CF (1-800-344-4823).

Please type your custom instructions and/or office contact information here.