Cystic Fibrosis

What is cystic fibrosis?

Cystic fibrosis (CF) is an inherited disease characterized by an abnormality in the glands that produce sweat and mucus. It is chronic, progressive, and is usually fatal. In general, children with CF live into their 30s.

Cystic fibrosis affects various systems in children and young adults, including the following:

- respiratory system
- digestive system
- reproductive system

There are about 30,000 people in the US who are affected with the disease, and about 2,500 babies are born with it each year. It occurs mainly in Caucasians, who have a northern European heredity, although it also occurs in African-Americans, Asian Americans, and Native Americans.

Approximately one in 20 people in the US are carriers of the cystic fibrosis gene. These people are not affected by the disease and usually do not know that they are carriers.

How does CF affect the respiratory system?

The basis for the problem with CF lies in an abnormal gene. The result of this gene defect is an atypical electrolyte transport system within the cells of the body. The abnormal transport system causes the cells in the respiratory system, especially the lungs, to absorb too much sodium and water. This causes the normal thin secretions in our lungs to become very thick and hard to remove. These thick secretions put the child with CF at risk for constant infection.

The high risk of infection in the respiratory system leads to damage in the lungs, lungs that do not work properly, and eventually death of the cells in the lungs. The most common causes for infection in the lungs of the CF patient are the following bacteria:

- Staphylococcus aureus
- Haemophilus influenzae
- Pseudomonas aeruginosa (PA)

Over a period of time, PA becomes the most common bacteria that causes infection and becomes difficult to fight. About 60 percent of respiratory infections in the CF patient are due to PA.

As a result of the high rate of infection in the lower respiratory tract, people with CF may develop a chronic cough, blood in the sputum, and sometimes can even have a collapsed lung. The cough is usually worse in the morning or after activity.
People with CF also have involvement of the upper respiratory tract. About 10 to 20 percent of patients have nasal polyps that need surgical removal. Nasal polyps are small protrusions of tissue from the lining of the nose that go into the nasal cavity. Children also have a high rate of sinus infections.

**How does CF affect the gastrointestinal (GI) system?**
The organ primarily affected is the pancreas, which secretes substances that aid digestion and help control blood-glucose levels.

As a result of the abnormal electrolyte transport system in the cells, the secretions from the pancreas become thick and lead to an obstruction of the ducts of the pancreas. This obstruction then causes a decrease in the secretion of enzymes from the pancreas that normally help to digest food. A person with CF has difficulty absorbing proteins, fats, and vitamins A, D, E, and K.

The problems with the pancreas can become so severe that some of the cells in the pancreas can become destroyed. This may lead to glucose intolerance and insulin-dependent diabetes. About 15 percent of CF patients develop this type of diabetes.

The symptoms that may be present due to the involvement with the GI tract include the following:

- bulky, greasy stools
- rectal prolapse - a condition in which the end part of the bowels comes out of the anus.
- delayed puberty
- fat in the stools
- stomach pain
- bloody diarrhea

The liver may also be affected. A small number of patients may actually develop liver disease. Symptoms of liver disease may include:

- enlarged liver
- swollen abdomen
- yellow color to the skin
- vomiting of blood

**How does CF affect the reproductive system?**
About 98 percent of males with CF have obstruction of the sperm canal and are sterile. This results from the abnormal electrolyte transport system in the cells, causing the secretions to become thick and lead to an obstruction. Women also have an increase in thick cervical mucus that may lead to a decrease in fertility, although many women with CF have children.

**What are the symptoms of cystic fibrosis?**
The following are the most common symptoms for cystic fibrosis. However, individuals may experience symptoms differently. Symptoms may include:

- abnormalities in the glands that produce sweat and mucus

This may cause a loss of salt. A loss of salt may cause an upset in the balance of minerals in the blood, abnormal heart rhythms, and, possibly, shock.

- thick mucus that accumulates in the lungs and intestines

This may cause malnutrition, poor growth, frequent respiratory infections, breathing difficulties, and/or lung disease.
other medical problems, such as:
  - sinusitis
  - nasal polyps
  - clubbing of fingers and toes - a condition marked by the ends of the fingers and toes become enlarged; more prevalent in the fingers.
  - pneumothorax - the presence of air or gas in the pleural cavity causing the lung to collapse.
  - hemoptysis - coughing blood.
  - cor pulmonale - enlargement of right side of heart.
  - abdominal pain
  - gas in the intestines
  - rectal prolapse
  - liver disease
  - diabetes
  - pancreatitis
  - gallstones

As stated above, the symptoms of CF differ for each person. Infants born with CF usually show symptoms within the first year. Some children, though, may not show symptoms until later in life. The following signs are suspicious of CF, and infants having these signs may be tested for CF:

- diarrhea that does not go away
- foul-smelling stools
- greasy stools
- frequent episodes of wheezing
- frequent episodes of pneumonia
- persistent cough
- skin tastes like salt
- poor growth

The symptoms of cystic fibrosis may resemble other conditions or medical problems. Consult a physician for a diagnosis.

**How is cystic fibrosis diagnosed?**
In addition to a complete medical history and physical examination, diagnostic procedures for cystic fibrosis include a sweat test to measure the amount of sodium chloride (salt) present. Higher than normal amounts of sodium and chloride suggest cystic fibrosis. Other diagnostic procedures include:

- chemical tests
- chest x-rays
- lung function tests
- sputum cultures
- stool evaluations

For babies, who do not produce enough sweat, blood tests may be used.

**Treatment for cystic fibrosis:**
Specific treatment for cystic fibrosis will be determined by your physician based on:

- your age, overall health, and medical history
- extent of the disease
- expectations for the course of the disease
- your tolerance for specific medications, procedures, or therapies
- your opinion or preference
Currently, there is no cure for CF. A cure would call for gene therapy at an early age and this has not been developed yet, although research is being done in this direction. The gene that causes CF has been identified and there are hopes that this will lead to an increased understanding of the disease. Also being researched are different drug regimens to help stop CF. Goals of treatment are to ease severity of symptoms and slow the progress of the disease. Treatment may include:

- management of problems that cause lung obstruction, which may involve:
  - physical therapy
  - exercise to loosen mucus, stimulate coughing, and improve overall physical condition
  - medications to reduce mucus and help breathing
- management of digestive problems, which may involve:
  - appropriate diet
  - pancreatic enzymes to aid digestion
  - vitamin supplements
  - treatments for intestinal obstructions

Newer therapies include lung transplantation for patients with end-stage lung disease. The type of transplant done is usually a heart-lung transplant, or a double lung transplant. Not everyone is a candidate for a lung transplant. Discuss this with your physician.

The genetics of cystic fibrosis:
Cystic fibrosis (CF) is a genetic disease. This means that CF is inherited. A person will be born with CF only if two CF genes are inherited - one from the mother and one from the father. A person who has only one CF gene is healthy and said to be a "carrier" of the disease. A carrier has an increased chance of having a child with CF. This type of inheritance is called "autosomal recessive." "Autosomal" means that the gene is on one of the first 22 pairs of chromosomes which do not determine gender, so that the disease equally affects males and females. "Recessive" means that two copies of the gene, one inherited from each parent, are necessary to have the condition. Once parents have had a child with CF, there is a one in four, or 25 percent chance with each subsequent pregnancy, for another child to be born with CF. This means that there is a three out of four, or 75 percent chance, for another child to not have CF.

The birth of a child with CF is often a total surprise to a family, since most of the time (in eight out of 10 families) there is no previous family history of CF. Many autosomal recessive conditions occur this way. Since both parents are healthy, they had no prior knowledge that they carried the gene, nor that they passed the gene to the pregnancy at the same time.

Genes are founds on structures in the cells of our body called chromosomes. There are normally 46 total, or 23 pairs of chromosomes in each cell of our body. The seventh pair of chromosomes contains a gene called the CFTR (cystic fibrosis transmembrane regulator) gene. Mutations or errors in this gene are what cause CF. This gene is quite large and complex. Over 1,000 different mutations in this gene have been found which cause CF.

The risk for having a mutation in the gene for CF depends on your ethnic background (for persons without a family history of CF):

<table>
<thead>
<tr>
<th>Ethnic Background With CF</th>
<th>Carrier Risk</th>
<th>Risk to Have a Child</th>
</tr>
</thead>
<tbody>
<tr>
<td>Caucasian</td>
<td>1 in 25</td>
<td>1 in 2500</td>
</tr>
<tr>
<td>Ashkenazi Jewish</td>
<td>1 in 29</td>
<td>1 in 3364</td>
</tr>
<tr>
<td>Hispanic</td>
<td>1 in 49</td>
<td>1 in 9600</td>
</tr>
<tr>
<td>African-American</td>
<td>1 in 65</td>
<td>1 in 17,000</td>
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</tbody>
</table>

Revised on 8/18/2008
Asian  
1 in 150  
1 in 90,000

Testing for the CF gene can be done from a small blood sample or from a cheek swab, which is a brush rubbed against the inside of your cheek to obtain cells for testing. Laboratories generally test for the most common mutations, and most labs test for anywhere from 30 to 100 total mutations. The detection rate depends on the person's ethnic background. In general, the detection rate for the Caucasian population is around 90 percent, 97+ percent for the Ashkenazi population, 57 percent for Hispanics, 75 percent for African-Americans, and 30 percent for Asians. The detection rate differs because CF is more common in certain geographical areas and certain populations of the world. There are many people with CF whose mutations have not been identified. In other words, all of the genetic errors that cause the disease have not been discovered. Because not all mutations are detectable, a person can still be a CF carrier even if no mutations were found by carrier testing. Testing for the CF gene is recommended for anyone who has a family member with the disease, or whose partner is a known carrier of CF or affected with CF.