Cystic Fibrosis

Cystic fibrosis causes a build-up of thick mucus that blocks ducts and tubes within the lungs, bowel, and pancreas. This build-up can cause severe and sometimes fatal infections and malabsorption of nutrients. It can also affect the sweat glands and male reproductive system. Cystic fibrosis is mostly a pediatric condition.

To diagnose cystic fibrosis, doctors usually perform blood tests. They may test sweat for high salt content, which can indicate cystic fibrosis. If the diagnosis is confirmed, doctors may evaluate the condition with a chest x-ray, chest or abdominal CT or MRI, abdominal ultrasound, or sinus CT. There is no cure for cystic fibrosis. But, recent advances in patient care have significantly increased patients’ expected life span. Your doctor may recommend lifestyle changes, medications, chest therapies, or surgery to help ease symptoms.

What is cystic fibrosis?

Cystic fibrosis is a life-threatening genetic disease that causes the body to create thick mucus. This thick mucus can build up and obstruct ducts and tubes within the lungs, digestive tract and pancreas. The build-up may cause severe and sometimes fatal infections and digestive issues. It may also affect the sweat glands and male reproductive system.

While cystic fibrosis may occur in people of any race, it is most common among Caucasians of Northern or Central European descent.

Symptoms of cystic fibrosis depend on the severity of the disease, but may include:

- Shortness of breath
- Frequent lung infections
- Persistent coughing
- Wheezing
- Fatigue
- Nasal congestion
- Stomach pain
- Weight loss
- Abnormal and foul-smelling stools
- Constipation
- Intestinal blockage
- Male infertility

There is now required screening for cystic fibrosis and other conditions at birth; therefore newborns are often diagnosed soon after birth, before symptoms occur. However, it is important to recognize early symptoms among infants, which include:

- Salty-tasting skin
- Lack of weight gain
• Delay in growth
• Rectal prolapse in which the rectum protrudes outside the body

How is cystic fibrosis diagnosed and evaluated?

To diagnosis cystic fibrosis, doctors take a blood sample for genetic testing or conduct a sweat test. A sweat test measures the amount of salt in a person's sweat. High salt in the sweat can indicate cystic fibrosis.

The United States now requires screening of newborns for cystic fibrosis through testing blood samples. In some cases, pregnant women can have their babies tested before birth through amniocentesis or chorionic villus sampling (CVS). Amniocentesis removes a small amount of fluid from the amniotic sac (the fluid around a developing embryo/fetus). The doctor inserts a needle through the abdomen and sends the sample for lab analysis. During CVS, the doctor uses a needle to remove a small amount of placenta. Lab analysis evaluates the sample for cystic fibrosis and other genetic diseases.

Doctors may use a stool test, spirometry, or sputum culture to evaluate cystic fibrosis. They may also order the following imaging tests:

• Chest (https://www.radiologyinfo.org/en/info/chestct) or abdominal computed tomography (CT) scan (https://www.radiologyinfo.org/en/info/abdomincl) : These exams use special x-ray equipment and computers to produce detailed images of the inside of the lungs or intestines. These images can help determine the severity of cystic fibrosis by looking for abnormal mucus and for dilated airways in the lungs. The test can also look for infection. This test uses ionizing radiation. See the Radiation Dose in X-Ray and CT Exams (https://www.radiologyinfo.org/en/info/safety-xray) page for more information about CT.

• Chest x-ray: (https://www.radiologyinfo.org/en/info/chesterad) This exam uses a small dose of ionizing radiation to produce images to evaluate for dilated airways filled with mucus and to evaluate lung infections that need to be treated with antibiotics. Doctors use chest x-rays periodically to observe changes in cystic fibrosis patients and rule out other respiratory conditions such as pneumonia (https://www.radiologyinfo.org/en/info/pneumonia) or a collapsed lung. See the Radiation Dose in X-Ray and CT Exams (https://www.radiologyinfo.org/en/info/safety-xray) page for more information about x-rays.

• Chest (https://www.radiologyinfo.org/en/info/chestmri) or abdominal magnetic resonance imaging (MRI): (https://www.radiologyinfo.org/en/info/mri-abdomen-pelvis) These exams use a powerful magnet, radio waves and a computer to produce detailed pictures of the lungs and digestive tract. Although chest x-ray or CT is used more commonly for this disease, MRI can help assess damage caused by cystic fibrosis.

• Abdominal ultrasound (https://www.radiologyinfo.org/en/info/abdominus) : This imaging test uses a small probe, gel, and sound waves to produce pictures of the upper abdomen. It evaluates the pancreas, liver, and gallbladder, all of which are affected by cystic fibrosis.

• CT of the sinuses (https://www.radiologyinfo.org/en/info/sinusct) : This exam combines special x-ray equipment and a computer to produce multiple images of the paranasal sinus cavities. It can help identify nasal polyps on just one side, which are common in patients with cystic fibrosis.

How is cystic fibrosis treated?

There is no cure for cystic fibrosis. However, the doctor may recommend the following to help ease symptoms and improve quality of life:

• Lifestyle changes such as better nutrition, use of vitamins, increased physical activity, avoiding tobacco, and avoiding secondhand smoke.

• Medications such as antibiotics and inhaled medicines may help open the airways and clear them of mucous and infection.

• Chest therapies or airway clearance techniques (ACT) may help to loosen mucus, which makes it easier to cough up sputum and eliminate it from the lungs.
In severe cases, surgery or other procedures may be needed to improve quality of life. These surgeries can include a lung transplant, procedures to stop bleeding from the lung, nasal polyp removal, or the introduction of a feeding tube to help deliver proper nutrients.

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