



Cystic Fibrosis Information for Parents

➤ Overview

Cystic fibrosis (CF) is an inherited condition. It affects organs in the body, particularly the lungs and digestive system, which become clogged with sticky mucus, making it difficult to breathe and digest food.

➤ What is cystic fibrosis?

Cystic fibrosis is an inherited disease of the mucus glands that affects many body systems. The disorder's most common signs and symptoms include progressive damage to the respiratory system and chronic digestive system problems.

Mucus is a slippery substance that lubricates and protects the linings of the airways, digestive system, reproductive system, and other organs and tissues. In people with cystic fibrosis, the body produces mucus that is abnormally thick and sticky. This can obstruct the airways, leading to severe problems with breathing and bacterial infections in the lungs. These infections cause chronic coughing, wheezing, and inflammation. Over time, mucus buildup and infections result in permanent lung damage, including the formation of scar tissue (fibrosis) and cysts in the lungs.

Most people with CF also have digestive problems because the abnormal mucus interferes with the function of the pancreas.

➤ Why is newborn screening done for cystic fibrosis?

Newborn screening is done for CF so that babies with this condition can be diagnosed quickly. Research studies conducted over the past two decades have shown that early intervention with nutritional therapies provides distinct benefits including improved height, weight and cognitive function for people with CF. These therapies may impact respiratory function and life expectancy, and reduce hospitalizations.

➤ Does a positive result from the Kansas Newborn Screening Lab mean that my baby has cystic fibrosis?

No, not necessarily. Kansas has a two-tiered testing system. The initial test is for the baby's level of immunoreactive trypsinogen (IRT). Kansas has adopted rigorous screening cut-offs for the IRT in order to identify children with CF as early as possible. If the IRT is above the cutoff, the lab will perform a second test on the same blood spot card. This test is a DNA test, and looks for mutations or changes within the cystic fibrosis transmembrane conductance regulator (CFTR) gene. If the laboratory finds any mutations, the next step is to perform a sweat chloride test, which is a diagnostic test for cystic fibrosis. If the IRT result is extremely high, with no DNA mutations, a sweat chloride test is also recommended. **It is important that the sweat chloride test be performed at an accredited cystic fibrosis center. The two centers in Kansas are listed on the back of this information sheet.** With early diagnosis and treatment, long-term health outcomes are improved.

➤ How common is cystic fibrosis?

In the United States, CF occurs in 1:3200 Caucasians, 1:15,000 African Americans, and 1:30,000 Asians.

➤ **How is cystic fibrosis inherited?**

Cystic fibrosis is inherited in an autosomal recessive pattern. Parents of a child diagnosed with CF are unaffected. These individuals are carriers of the condition and have one normal CFTR gene and one abnormal CFTR gene. Each pregnancy between carrier parents has a 25% chance of producing a child affected with CF, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

➤ **What are the signs and symptoms of cystic fibrosis?**

Signs and symptoms of CF usually appear in babies under the age of 12 months. However, some children may not develop signs and symptoms of cystic fibrosis until they reach preschool age. In some newborns, the first sign may be a blockage of their intestines (meconium ileus). Other signs in newborns may include:

- Failure to grow
- Bulky and greasy stools
- Frequent respiratory infections

Cystic fibrosis symptoms in children and young adults may include:

- Salty taste to the skin. People with CF tend to have higher than normal amounts of salt (sodium chloride) in their sweat. This may be one of the first signs parents notice because they can taste the salt when they kiss their child.
- Blockage in the bowels.
- Foul-smelling, greasy stools.
- Coughing or wheezing.
- Delayed growth.
- Thick sputum. It's easy for parents to overlook this sign because young children tend to swallow their sputum rather than cough it up.

➤ **How is cystic fibrosis diagnosed?**

Any newborn screening result that has an elevated IRT and one or two DNA mutations should have a sweat test arranged by your doctor.

➤ **Is there a cure for cystic fibrosis?**

No, there is no cure for cystic fibrosis. The pace of CF science suggests that there is good reason to feel optimistic about the future.

➤ **How is cystic fibrosis treated?**

Common treatments include lung therapies and breathing treatments, as well as antibiotics, to minimize the risk of lung infections. Complications within the pancreas can be managed by controlling the patient's diet, and providing supplements of pancreatic enzymes to add to food.

➤ **Certified cystic fibrosis centers in Kansas**

KU Pediatric Cystic Fibrosis Center
Kansas City, KS
KU sweat test scheduling: 913-588-6224

Wichita Cystic Fibrosis Clinic
Wichita, KS
Wichita sweat test scheduling: 316-689-6212

➤ **Where can I get additional information?**

www.cff.org

www.cysticfibrosis.com

<http://www.genetests.org/>