

# What is cystic fibrosis?



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Your child is under weight, has greasy, smelly diarrhea, a chronic cough and seems to always be getting pneumonia. He wheezes when he breathes and has impaired exercise ability. He seems to never being growing to the average size of his peers and his skin is very salty (5). The doctors first instinct should be to test your child for a disease called cystic fibrosis. Cystic fibrosis is a disease, which causes the mucus in your body to be thicker in some areas. The most affected areas are the lungs and digestive system. As a result, the person may get chest infections and have difficulty digesting food. When cystic fibrosis is a consideration for a persons disease the physician will do what is called a sweat test. This is a simple and cheap way to determine if your child has the disease. Because people who have cystic fibrosis (CF) have saltier sweat than others, the sweat test detects the amount of salt in the sweat of the individual (4). This is the best way to diagnose a person with the disease (5).

CF is a genetic disease, meaning that a person does not catch it, it is inherited in the genes passed down to the person. Cystic fibrosis lasts with the patient for their whole life. This makes it a chronic disease, not communicable (9). All symptoms of CF are caused by a mutation to the single chromosome 7. This particular gene is responsible for the building of the protein called the cystic fibrosis trans membrane conductance regulator (CFTR). Normally, the gene regulates the passage of chloride ions in and out of the cell, but when mutated as in the case of CF patients the chloride ions cannot move throughout the cell membrane because the CFTR does not open. In about three- quarter of CF cases the CFTR is not just broken, but is completely missing from the cell. The missing CFTR has many effects on the

human body. One is saltier sweat because as the sweat rises to the skin, cells reabsorb sodium and chloride molecules. The loss of the CFTR does not allow the body to reabsorb the sodium ions making a CF patient's sweat five times saltier than that of a normal person. This can lead to irregular heart rhythms. The loss of chloride ions affects the pancreas also. The loss of the CFTR ultimately makes it impossible for digestive enzymes to be sent to the intestines. A normal CFTR allows water to follow chloride through the duct and out of the cells, but when the CFTR is missing then the chloride and water remain inside the cells. The enzymes are unable to be carried away, causing the enzymes to digest the pancreas itself. This leads to inflammation and thick mucus plugging the duct. It is not exactly known why, however the organ that suffers the most damage from the lack of CFTR is the lung. For some reason, the mucus becomes very thick in the lungs and leads to a lot of bacterial growth. The mucus is the perfect living environment for bacterial growth causing all kinds of infections. Being hard to rid, the mucus builds up in the bronchial tubes and reduces the surface available for oxygen exchange. When the body tries to kill the infection, the enzymes in turn causes more lung damage. More lung cells are killed, and chronic infections seriously damage the lung (whole paragraph from 6).

There is no way to prevent someone from contracting CF. The only thing that could be done is for everyone to be know whether or not they are a carrier, and whether or not their partner is a carrier. In order for a child to be born with CF, both parents must be carriers for the disease. A person may be a carrier and not even know it, carriers of the disease have absolutely no characteristics of the disease, but they simply are able to pass on the gene

to their offspring. When one carrier has a child with another there is a twenty-five percent chance that the child will have CF. There a fifty percent chance that the child will be a carrier for the disease and there is a twenty-five percent chance that the child will not be a carrier or have the disease (7).

There is no cure for cystic fibrosis, but treating the effects of the disease is the only thing that can be done. Doctors generally will put the patient on anti-biotics to control infections. Chest physical therapy can be done to help break the mucus in the lungs and to help clear it out of the lungs. In order to deal with the digestive portion of the disease, digestive enzyme supplements have been deigned to help the CF patient digest what they would normally not be able to (1). This includes all fats, proteins, and carbohydrates. These pancreatic supplements should be taken with each meal, snack and even drinks high in fat like milk. The pancreatic enzymes include the important enzyme lipase, which digests fat. Enteric-coated granules are contained within a gelatin capsule. When swallowed whole, the outer capsule dissolves in the stomach releasing the granules, which then mix with food (8). The enzymes in the granules are protected from the acid in the stomach by a special 'enteric' coating, which enables them to be released later down in the small bowel where they are needed (the small bowel is where most digestion takes place). In order to make sure that the patient remains healthy, it is recommended that the patient have their health monitored by a clinic or a hospital (3). Frequent hospital stays are not uncommon and can be beneficial to the over all health of the individual. The CF patient must have a diet high in vitamins, salt, and minerals. In 1993 the Food and Drug

Administration approved the first drug specifically for CF patients in over 30 years. The mucus-thinning drug, Pulmozyme, has shown to reduce respiratory infections and improve over-all lung function (3). The drug ibuprofen has shown to reduce the inflammation of lungs of children, in high doses (2). In 1997, TOBI (tobramycin solution for inhalation) was approved by the FDA, this drug is a concentrated anti-biotic inhaled into the lungs. This drug has shown to reduce hospitalizations. This drug is so effective because it is put directly into the site of lung infections making it harder for infections to occur (3). The only effect of the disease that can not be treated is the short life expectancy, however the advanced treatments that have been developed have assisted in prolonging the life expectancy of CF patients. In 1940 the majority of children with CF did not live until their first birthday. By 1969 most patients with CF lived until their 14th birthday, and in 1996 the average life expectancy of a person with CF is 39 years old, this figure is constantly improving (2).

Perhaps the hardest thing to deal with is the fact that there is nothing that can be done to prevent it from happening, it is simply in the DNA makeup of the individual. Cystic fibrosis is a challenging disease to live with but the treatments that have been developed are forever improving the quality of life of those diagnosed with the fatal disease. Maybe with the advancements in medicine, one day there will be a cure, but all that can be done is hope.

Words

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