

MAIL TODAY

Cystic fibrosis is a genetic condition that results in the formation of too much mucus

By MEGHAN IEN DUTTA LINGAM

IF YOU notice that your child has a perpetual cough and wheezing don't brush it off as a bout of chest infection. If he is also not growing at the rate he should be, there is good reason to head to the doctor. He may very well be affected with cystic fibrosis (CF). This disorder causes severe lung damage and nutritional deficiencies.

Cystic fibrosis is an inherited condition, which affects the cells that produce mucus, sweat, saliva and digestive juices. "Normally, these secretions are thin and slippery, but in cystic fibrosis, a defective gene causes them to become thick and sticky. Instead of acting as a lubricant, the secretions plug up tubes, ducts and passageways, especially in the pancreas and lungs," says Dr Sabita Jain, HOD, paediatrics, Max Healthcare.

THE GENES

IN CYSTIC fibrosis, a defective gene alters a protein that regulates the normal movement of salt in and out of cells. This results in thick and sticky mucous-like secretions in the respiratory and digestive tracts, as well as in the reproductive system. It also increases the salt level in sweat. This condition causes lung obstruction, infection, poor digestion and food absorption. Cystic fibrosis is genetic and its greatest risk factor is a family history of the disease. If both parents come from families with CF, there is a 25 percent chance that the child will inherit it, a 50 percent chance the child will be a carrier of the cystic fibrosis gene, and a 25 percent chance the child will neither have the disease nor be a carrier. Hence, if one has a family history of cystic fibrosis, it is advisable to seek genetic counselling before starting a family. People who carry the CF gene may be healthy and manifest no symptoms of the disease — they may be carriers and not know it. Although parents often blame themselves when a child is born with cystic fibrosis, it's important to remember that it is not caused by anything a parent consciously does.

Lifestyle factors — environment, diet, exercise, etc do not cause CF. It is not contagious and cannot be passed from one person to another except through inheritance.

LUNGS ARE STRUCK HARD

FREQUENT chest and sinus infection with recurring pneumonia and bronchitis is the most common symptom. Coughing and wheezing often accompany this. Also, thick sputum cannot be missed in such patients, though it can be easily missed in children, as they tend to swallow their sputum rather than cough it up. Another symptom is a salty taste to the skin which parents notice when they kiss their child, caused by higher than normal amounts of salt in their sweat. Constipation, due to blockage of bowels, accompanied by foul-smelling and greasy stools is also common in such patients. Diarrhoea also occurs in older children. Since digestion is not optimum delayed or undeveloped growth is also seen.

"In some newborns the very first sign may be a delay in the passage of stools, which indicates blockage of the intestines. It should ideally be passed within a day of birth. The stools become too thick to pass through the intestines. Moreover when the baby does pass stools it is greenish black in colour," says Dr Neelam Mohan, paediatric gastroenterologist, Sir Gangaram hospital.

Parents need to worry about cystic fibrosis only if the child displays both respiratory and gastrointestinal symptoms.

DIAGNOSIS GETS DELAYED

PARENTS usually tend to overlook symptoms until it persists for a long time and mostly children are detected with this condition at the age of around two. Once the symptoms indicate CF it is confirmed through a sweat test. This is a standard diagnostic test which measures the amount of sodium or chloride in a person's sweat. During the procedure, a small

amount of a sweat-producing chemical is applied to a small area on the arm or leg. An electrode attached to the area stimulates a weak electric current, causing a tingling or warm feeling. After several minutes, sweat is collected from the stimulated area and sent for analysis. A high level of salt indicates cystic fibrosis.

"Once diagnosed with this condition it is advised regular check ups — monthly or tri-monthly — with the doctor and a cystic fibrosis clinic where the patient is screened for any infections and have any lung or digestive problems monitored," advises Dr Jain.

COMPLICATIONS ABOUND

FREQUENT complications of cystic fibrosis are chronic respiratory infections, because thick mucus blocks the airways and provides an ideal breeding ground for bacteria. Sinusitis and bronchiectasis — an abnormal

dilation of the walls of the bronchial tubes that makes it difficult to clear the airways — are also common. Asthma too can result from chronic inflammation of the bronchial lining. People may also develop bleeding from the lungs thereby coughing up blood and respiratory failure. Ultimately, complications from lung problems are the commonest and prove fatal for many with cystic fibrosis.

This condition also makes people prone to chronic diarrhoea and severe nutritional deficiencies. Thick secretions obstruct the ducts in the pancreas, preventing enzymes that digest fats and proteins from reaching intestines. These secretions also prevent the body from absorbing the fat-soluble vitamins A, D, E and K. "Since it affects the pancreas which in turn controls the level of sugar in blood, one may develop cystic fibrosis related diabetes. In addition, the bile duct (that carries bile from liver and gallblad-

der to the small intestine), may get blocked and inflamed, leading to liver problems," says Dr Mohan. Other complications include rectal prolapse (protrusion of part of the rectum through the anus caused by stools that are difficult to pass or by frequent coughing), growths (like polyps) in the nasal passages.

Cystic fibrosis affects the reproductive system too. The thick secretions often block the tubes in sexual organs. It may even result in infertility in men and women. But certain surgical procedures may provide a solution. Those with CF are prescribed pulmonary therapy and inhalation therapy to tackle any respiratory infections. Pancreatic enzyme supplements are given to regulate the digestive juices and nutritional supplementation like vitamin and mineral supplements are a must. A higher than normal caloric diet is also advised.

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BLAME IT ON YOUR DNA



LIFESTYLE AND HOME REMEDIES

Certain lifestyle measures can be adopted by those with cystic fibrosis that can help alleviate their symptoms

UP TO DATE IMMUNIZATIONS

Keep your child's immunizations up to date. In addition to other usual childhood vaccines, this includes the pneumococcal and influenza vaccines. Cystic fibrosis doesn't affect the immune system, but patients are more likely to develop complications when they become sick.

LEAD AN ACTIVE LIFE

A patient should be encouraged to live an active and normal life. Exercise is extremely important for people of all ages who have cystic fibrosis. Regular exercise helps loosen mucus in the airways and strengthens the heart and lungs.

EAT HEALTHY

As nutrients don't get absorbed too easily, it is important for one to eat high calorie food.

ADD NUTRITIONAL SUPPLEMENTS

Augment your nutritional needs through mineral and fat-soluble vitamin supplements and pancreatic enzymes to stay as healthy as possible.

GULP DOWN MORE

Encourage your child to drink plenty of liquids to help loosen the mucus. This is especially important in the summer when children are active and tend to lose a lot of fluids.

HANDWASHING IS IMPORTANT

Teach everyone in the family to wash their hands thoroughly before eating, after using the bathroom, after returning from work or school, and after being with a sick person. This is the best way to protect against infection.