

Indian Academy of Pediatrics (IAP)



GUIDELINES FOR PARENTS

Care of a Child with Cystic Fibrosis

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10 FAQs on CARE OF A CHILD WITH CYSTIC FIBROSIS

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Care of a Child with Cystic Fibrosis

Q1

What is cystic fibrosis disease? Why does it happen?

Cystic fibrosis is a family-run genetic/hereditary disease. It is commonly known as CF. It is not a contagious disease. In Western countries, the disease is found in 1 in 2,500 newborns. The one gene of CF is found in one person per 25 people and these individuals are called carriers. If both parents of a child are carriers of CF, they may transmit one CF gene each to the child and he/she will manifest CF disease due to presence of two CF gene in his/her body.

It has now become clear that this disease also occurs in India. However, actual prevalence of it in India is not known. Its diagnosis is not possible everywhere due to lack of testing facilities. This is the reason, why many doctors have not seen CF children and they do not know much about this disease.

Q2

Nobody in our family has cystic fibrosis (CF) disease, then how did our child get it?

Every person has 1,000s of genes and these are necessary for normal body work. But, if there is any defect in any of these genes, then they will not be able to do their work properly. Every person has a problem with 7–9 genes. Genes in a child come from his/her parent. If the father's bad gene gets into the child but the mother's gene is normal, then the child is called a carrier and does not get the disease.

Incidentally, if the gene that causes disease of both mother and father gets into the child, then it becomes disease (**Fig. 1**). It may be that the mother and father's family has a bad gene running for the last several generations, but disease does not happen until both parents are carrying a bad gene each.

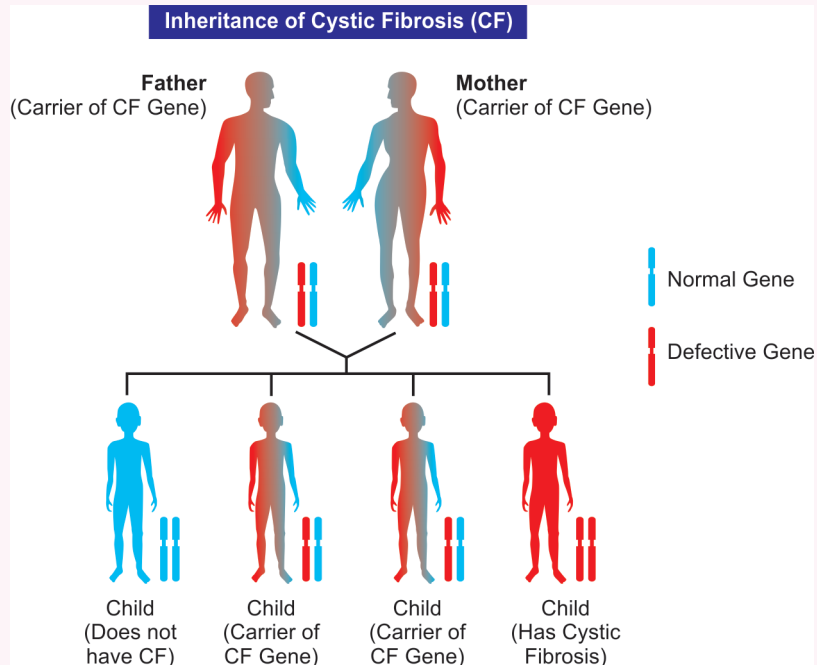


Fig. 1: Inheritance of cystic fibrosis (CF).

Q3

What are the symptoms of CF? Why do they occur?

The body is made up of innumerable cells. For the regular work by the cells, it is necessary to control the amount of salt in them. The cells have a chloride channel, whose job is to control the movement of salt in the cells. This channel does not function properly due to the inherent disadvantages/defects in CF and that causes increased thickness/viscosity of all secretions of the body. Most symptoms of CF are due to thick mucus in airways of lung, lack of digestive juice (pancreatic enzymes) in intestine, and excess salt in sweat.

Q4

What happens due to thickening of mucus and lack of pancreatic enzymes?

Due to the formation of thick and viscid mucus, it tends to get stuck on the wall of a windpipe (the tracheobronchial tree) and is difficult to remove with normal clearance function of body. This thick mucus leads retention of secretion inside the windpipe of lung. On progression of disease, retention of thick mucus secretion blocks the airway/windpipe and leads to recurrent cough with breathing difficulty. This retention of thick secretion also causes growth of harmful bacteria inside the airway that presents as frequent pneumonia. Ultimately, lung gets damaged due to recurrent pneumonia.

During pregnancy (inside the mother’s abdomen), sometimes intestines of CF children can get obstructed due to increased thickening of the digestive juice; and may burst, which is called meconium ileus. This condition requires surgery immediately after birth.

After birth, as thick pancreatic juice (enzymes) cannot enter into the intestine (**Fig. 2**), deficiency of pancreatic enzymes causes following problems:

- Frothy, oily, and foul-smelling stool which is difficult to clean.
- Stool floats in water while flushing in toilet.
- Poor weight gain even after having adequate food intake, child asks for meals frequently due to hunger.
- Recurrent pain in abdomen and sometimes lower part of the intestine (rectum) comes out of body which has to be pushed back inside manually (known as “prolapse”).

Due to excess salt in the sweat, white marks are formed over skin when the sweat gets dry, and kissing the baby gives salty taste. Child tends to drink more water and eats more salts due to dehydration in summer season.

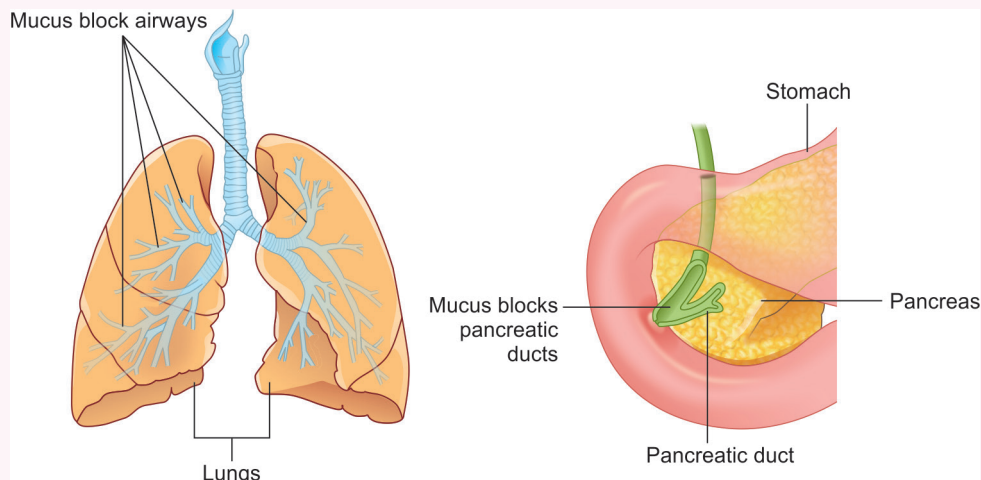


Fig. 2: Mucus plug in wind pipe and pancreatic duct.

Q5

How to diagnose CF disease in a child?

It is identified/diagnosed by sweat test. The amount of chloride in CF baby sweat is >60 mEq/L.

Genes can also be tested in the blood sample, but $>1,500$ genetic variants responsible for CF disease have been identified so far and it is difficult to detect all of them in blood. Therefore, usually 7–10 common genes responsible for around 90% cases CF are investigated. It is not necessary that blood test for genetic study of all CF children to be defective, sometimes the child's blood genetics report can be normal even if child has CF due to above reason.

Q6

What is the treatment for CF? How can the symptoms of the disease be reduced?

Cystic fibrosis is not a curable disease. The purpose of treatment is to reduce the child's suffering, to prevent the child from being hospitalized repeatedly, and to control the pace of disease progression. The speed of progression of the disease can be regularized/reduced by following measures:

- Regular follow-up in CF clinic/treating physician
- Keeping the chest clear by removing thick mucus from airway
- Taking pancreatic enzymes supplements to digest food
- Taking vitamin supplements

When you take the child to the hospital, staff/physician takes measurements of his/her weight, height along with check-up of his health. This gives an idea of the pace of progression of disease. Sputum/cough swab is also taken during hospital visit for culture sensitivity test. According to the report of sputum culture/cough swab, antibiotic medicine is given when needed. The amount of medicine should be increased according to the weight gain of the child.

Keeping the chest clear: Following things help in keeping the chest clear:

- Drinking more water
- Taking appropriate amount of salt
- Performing regular chest physiotherapy
- Using medicines as per doctor's advice (salbutamol, 3% saline, budesonide/foracort, tobamist, and DNase)
- Giving antibiotic medicine as per doctor's advice

Q7

What are the medicines to help in digestion of food in CF child? How to give these medicines?

Most CF children require pancreatic enzymes supplements. This is available in capsule or tablet form. In capsule, this drug comes in the form of small particles (spherule). There is a thin membrane on these spherule which goes away after reaching in the intestine and starts digesting food. If their membrane gets removed in the stomach or before reaching into intestine, then their food digesting power is destroyed. Therefore, it is necessary to swallow spherule or tablet as whole to digest food. This medicine should be given before meals. If the child is unable to swallow the capsule, then, after opening of capsule shell, spherule can be put on vegetable/jam/butter to feed the child. The quantity of capsules is to be given at the time of eating as per doctor's instruction. Pancreatic enzyme supplement with fruit, vegetable or juice is not required. If the food is greasier or having more fat content, then the amount of capsules/tablet must be increased.

You have to be watchful about the baby's stool consistency and habit. If child passes stool 1–2 times/day, does not smell, is not oily, and does not float in water, there is no pain in the stomach and body weight is increasing, then the dose of pancreatic enzyme is fine. Otherwise, it has to be increased. Keep the pancreatic enzyme safe in the cool/dry part of the house. If possible, keep it in the refrigerator (4–8°C).

Q8

Describe vitamin supplements and other medicines.

- *Vitamin:* Vitamin (A, D, and E) is deficient due to lack of power to digest food (which are fat soluble). Therefore, these vitamins supplements should be given every time when you give pancreatic enzymes. Because without the pancreatic enzymes, these vitamins will not be absorbed.
- *Salt, potassium, and water:* As it is known, the amount of salt in sweat is high in CF, so due to excessive sweating in summer, there is deficiency of salt in the body. Therefore, salt and water need to be given more in summer. Syrup Potklor also should be given along with it in summer. Syrup Potklor has a slightly bitter taste, you can mix it with water and/or sugar.
- *Antibiotics:* In CF, germs/bacteria grow in airway/windpipe which is difficult to clear. If we do not give antibiotics, the speed of lung worsening will increase. After receiving antibiotics injection or pills repeatedly and for prolonged duration, one may have some adverse effects. Therefore, antibiotics are given as inhalation when required. As the dose used is less, the adverse effects may be reduced. Inhalation antibiotic should be given continuously for few months.
- *Salbutamol:* Helps in clearing the airways and can be given with inhalation (by nebulizer or inhaler). This drug is usually given before giving 3% saline.
- *3% saline:* After reaching the airway, it draws water from the body. This causes thickened mucus to become thin which then can be expelled easily by physiotherapy.

Q9

To pay attention for lung infection/pulmonary exacerbation. Explain.

Most lung infections begin with a mild cold. The symptoms of which are mild runny nose, mild fever, and cough. But, if any of the following symptoms occur in your child, then you should take them to the doctor:

- Increased frequency and severity of cough
- Shortness of breath or increased breathlessness
- Cough with vomiting
- Weight loss
- Loss of appetite
- Decreased ability to play or exercise
- Persistent fever
- Increased or fresh onset of mucus/sputum production, or yellow/green mucus

If you think that your child has a lung infection, you should do chest physiotherapy three to four times a day and contact the CF center where antibiotics could be given to control lung infection.

Q10

Will there be a definitive cure for cystic fibrosis in future?

In the last 50–60 years, the development and progress of medicine have greatly improved the treatment of CF. We hope this will improve more in the future. You are not alone in the battle of CF. People involved in CF care of your child are:

- Clinic coordinator
- Family service coordinator
- Cystic fibrosis doctor
- Cystic fibrosis nurse/CF research student/nurse
- Dietician
- Child psychologist/Gastroenterologist
- Genetic counselor
- Respiratory physician (Physiotherapist)
- Social worker
- Cystic fibrosis parents self-help group
- Your family and your child