Cystic Fibrosis Disease

Patient and Family Education

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Understand the Disease... Take Control of Your Life

What is Cystic Fibrosis?

It is a chronic inherited disease that affects various organs in the body, mainly:

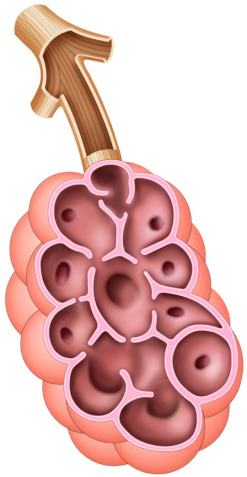
- The lungs
- The digestive system (especially the pancreas)
- The sweat glands

In this disease, there is a defect in a specific type of protein called CFTR, which is responsible for regulating the movement of salt and water in and out of body cells.

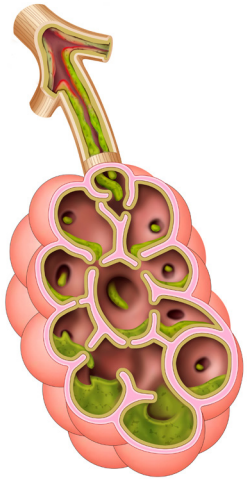
This defect leads to:

- The production of thick, sticky mucus that builds up in the lungs and is difficult to clear
- Blockage of digestive ducts might lead to pancreas insufficiency which prevent proper absorption of food.

Healthy Airway

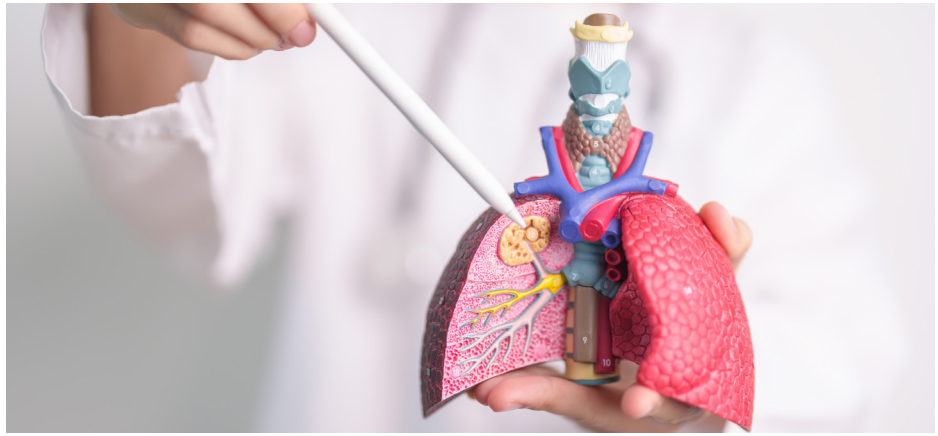


Cystic Fibrosis



How does a person get it?

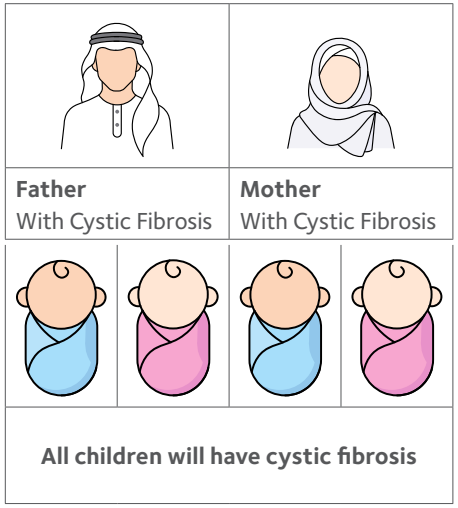
- The condition results from a defect in a specific gene
- A child must inherit the gene from both parents to develop the disease
- If the child inherits the gene from only one parent, they won't develop the disease, but they can pass the gene to their children
- Cystic fibrosis is not contagious
- Cystic Fibrosis is an inherited genetic condition passed from parents to their children. It cannot be spread from one person to another through contact, coughing, or touching.

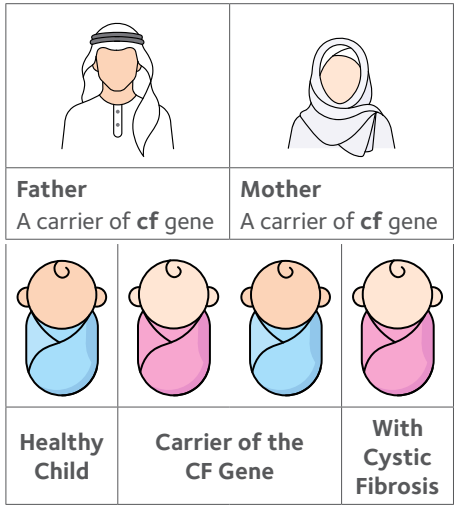


How is Cystic Fibrosis Passed from Parents to Child?

1. When both parents have cystic fibrosis, each parent has two defective copies of the CFTR gene. all childrens will receive 2 faulty genes.
   - **Result:**
     All children will have cystic fibrosis.

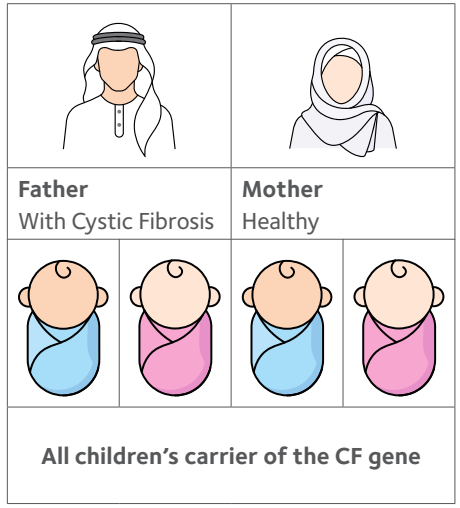
2. If both parents are carriers of the faulty gene (even they are not affected by desease.), there is a 25% chance that the child will be born with the disease.

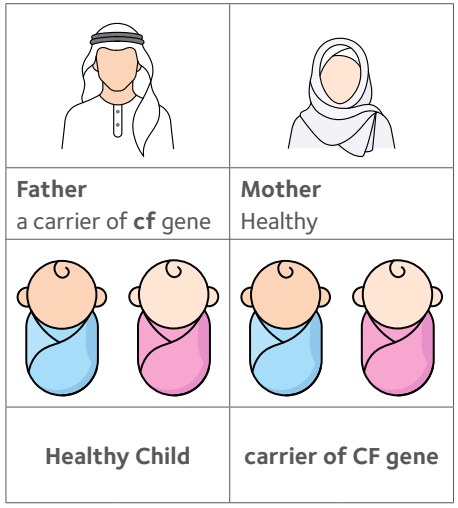




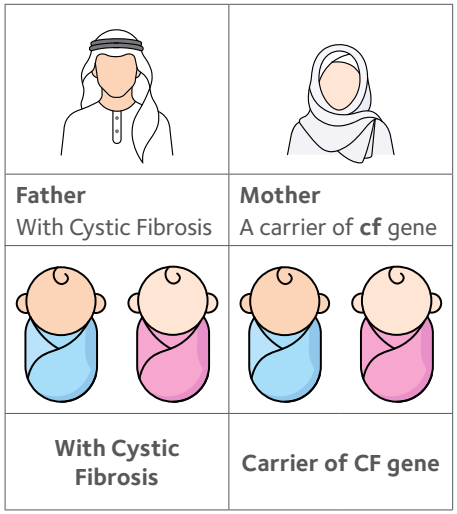
3. When one parent has cystic fibrosis (carries two defective copies of the CFTR gene) and the other parent is completely healthy (carries two normal copies of the gene):
   - All children resulting from this marriage will be carriers of the disease (they will have one faulty and one healthy copy of the gene), but they will not be affected by the disease themselves.

4. If one parent is completely healthy (does not carry the cystic fibrosis gene) and the other parent is a carrier of the CF gene, then:
   - 50% chance the child will be completely healthy.
   - 50% chance the child will be a carrier of the CF gene from both parents.
   - None of the children will have cystic fibrosis.





5. If one parent is a carrier of the cystic fibrosis (CF) gene (meaning they have one normal gene and one CF gene) and the other parent has cystic fibrosis (meaning they have two CF genes), the potential genetic outcomes for their children are as follows:
    - 50% chance the child will receive one CF gene from the CF parent and a normal gene from the carrier parent, making them a carrier of CF (just like the carrier parent).
    - 50% chance the child will receive two CF genes (one from each parent) and will have cystic fibrosis.



Common Symptoms:

Symptoms in adults

They may vary from person to person, but typically include:

Respiratory System:

- Chronic cough with thick mucus
- Recurrent chest infections
- Shortness of breath, especially during physical activity
- Wheezing or crackling sounds while breathing

Digestive System:

- Weight loss despite eating well
- Fatty or foul-smelling stools
- Abdominal pain or bloating
- Delayed growth or poor weight gain
- Poor absorption of vitamins and fats

Other Symptoms

- Male infertility
- Liver problems
- Frequent sinus infections
- Diabetes

How to diagnose?

- Newborn screening
- Sweat test (to measure salt levels in sweat, which are elevated in CF)
- Genetic testing to detect CFTR gene mutation

Is there a cure?

- There is no definitive cure for cystic fibrosis yet, but symptoms can be managed and complications delayed. Treatment depends on the symptoms and condition of the patient and includes:

Medications:

- Bronchodilators
- Antibiotics to treat infections
- Mucus-thinning drugs
- Pancreatic Enzyme Replacement
- New Gene Therapy

Respiratory Therapy:

- Mucus-clearing exercises
- Mucus-clearance devices

Nutrition:

- People with cystic fibrosis need a specialized nutrition plan that is high in calories, protein, fats, and salts. This can be achieved by following the guidelines below:

1. A high-calorie diet (120–150% of the normal daily requirement)
   It is recommended to increase caloric intake by:
   - Consume 5–6 small meals/day.
   - Add oil or butter to food.
   - Consume full-fat dairy products.
   - Add nuts to meals or eat them as snacks throughout the day.
2. Protein
   - You need to increase the amount of protein to support growth and immunity.
   - Good sources include eggs, meat, chicken, fish, dairy products, and legumes.
3. Healthy fats
   - A key source of energy. Healthy fats include olive oil, avocados, and nuts.
   - They can be added to meals or eaten as snacks throughout the day.

4. Salt

You need extra salt because you lose significant amounts through sweating.

And you need more salt in the following situations:

- Hot weather
- Physical activity
- Fever or illness

5. Fluids

You need an adequate amount of fluids (2-3 L/day) to prevent dehydration and improve digestion and metabolism.

To avoid dehydration, notice the following signs:

- Dry mouth.
- Reduced urination.
- Feeling fatigued.

6. Nutritional supplements

- Due to the increased caloric and nutrient needs, you may require high-calorie or high-protein supplements to maintain adequate energy and protein intake, promote weight gain, and meet nutritional needs.
- Nutritional supplements are prescribed by physicians and clinical dietitians.

How can patients live with the disease?

- Stick to daily treatment
- Avoid smoking (active and passive)
- Avoid contact with people with infectious diseases
- Avoid certain environments containing dust, excessive dirt and ponds dust
- Exercise regularly to improve breathing
- Maintain a healthy weight
- Attend regular medical check-ups
- Wash hands frequently to prevent infections
- Get necessary vaccinations (like flu and pneumonia)
- Social support

Can the patient get married?

Yes, you can get married, with your physician's guidance

Can People with Cystic Fibrosis Have Children?

Men with cystic fibrosis are usually infertile, but modern techniques will help.

Women are often able to conceive with continuous medical care.

When should you see a doctor?

You should attend regular medical appointments

Visit your healthcare provider if you notice:

- If coughing or phlegm increases in severity.
- In case of fever or blood presence in the phlegm.
- Weightloss or decreased appetite
- Any sudden change in health condition

Key Message:

- With proper care, cystic fibrosis patients can breathe easier and live a healthier life.