Cystic Fibrosis

What is Cystic fibrosis?

Cystic fibrosis (CF), a rare, progressive, life-threatening disease, results in the formation of thick mucus that builds up in the lungs, digestive tract, and other parts of the body. It leads to severe respiratory and digestive problems as well as other complications such as infections and diabetes.

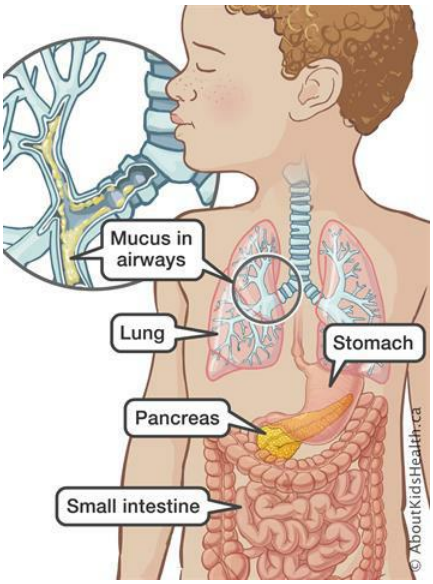
How does it occur?

Cystic fibrosis is a genetic disease caused by defects in the in the cystic fibrosis transmembrane conductance regulator (CFTR) gene that encodes for CFTR protein. The severity of the disease depends upon the type of defect in the gene and resultant deficiency and dysfunction of CFTR protein. Delta F 508 is the commonest gene defect identified .

Cystic fibrosis is autosomal recessive disease. People with CF inherit two copies of the defective CF gene one copy from each parent. People with only one copy of the defective CF gene are called carriers, but they do not have the disease. Each time two CF carriers have a child, the risk of CF will be 25 percent (1 in 4).

What are the problems with this disease?

People who have cystic fibrosis make thick, sticky mucus that can build up and lead to blockages, damage, or infections in the affected organs. Inflammation also causes damage to organs such as the lungs and pancreas. The presentation may be very early in neonatal period or in early infancy with bowel obstruction for which emergency surgery may be needed. The common presenting clinical features are shown in box and figure below



i) Salty-tasting skin on kissing

ii) Persistent coughing, at times with phlegm

iii) Frequent lung infections including pneumonia or bronchitis

iv) Wheezing or shortness of breath

v) Poor growth or weight gain in spite of a good appetite

Internet source :support@upopolis.com

The long term complications of cystic fibrosis may affect the function of lungs, liver, pancreas, intestine, kidney, urinary bladder, bones and reproductive organs. It may cause male infertility.

What are the treatment options?

Supportive therapy: As of now, diverse medications are used that have the purpose of loosening mucus, expanding airways, decreasing inflammation, and fighting lung infections, improving digestion respectively to relieve the symptoms of CF for healthier and meaningful life,

i) Pancreatic enzymes replacement
ii) Vitamin and salt supplementation
iii) Airway clearance using bronchodialators, mucolytic agents and various techniques including devices
iv) Steroid inhalers
v) Antibiotics
vi) Recombinant human DNase I (rhDNase I) inhalation

vii) Specific therapy: Breakthrough research in the last decade has led to discovery of drugs called CFTR modulators . These drugs target the specific gene defect . The results on long term benefits of these very expensive drugs are awaited. The clinical trials have shown some improvement in the markers of disease progression in lungs. Though the effect is small but functionally it can cause changes in quality of life of patients having cystic fibrosis.

Is this disease completely curable?

Though there is no definite cure, with over 6-7 decades of research survival has improved from a median of 6 months to 4-5 decades, with enzymes, airway clearance, antibiotics and other supportive care. The prognosis for CF has improved due to earlier diagnosis through screening and better treatment and access to health care. The ongoing clinical trials with newer molecules may bring new rays of hope in the life of children suffering from this fatal and debilitating disease.

What is the cost of therapy?

For a 20 kg child per month cost
1. Enzyme+ vitamin and electrolyte supplementation: Rs 10000 (Ten thousand)
2. Add inhaled antibiotics: Rs 25 000 (Twenty five thousand)
3. Add DNAse (Not being marketed by Indian Pharmaceutical agencies: INR 100000 per month
4. Potentiators and correctors (Only for Specific gene defects): Not marketed by Indian pharmaceutical company.

- Trikafta (elexocaftor+ivacaftor+ Tazacaftor): For children with at least one gene variation of Delta F 508: Rs 3000000 (Thirty lacs per month)
- Orkambi (ivacaftor+ Lumacaftor) for children with Homozygous for Delta F 508: Rs 1500000 (fifteen lacs per month)

- Ivacaftor alone (only for specific class 3 mutations) not commonly seen in India: Rs 1500000 (Fifteen lacs per month)

5. Additional course of oral or IV antibiotics and annual investigations: Rs 500000 (Fifty thousand) per annum, presuming that child may need hospitalization twice and ambulatory treatment twice

What happens if the CF patients are left untreated?

If the patients are left untreated they develop progressive deterioration of lung function and respiratory failure also

Prevention: Families having affected children are encouraged to consult a clinical geneticist for counseling and prevention of recurrence in future pregnancies.