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# Case Report on Cystic Fibrosis

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**ABSTRACT:** Cystic fibrosis is a genetic disorder that primarily affects the lungs and digestive system. It is caused by mutations in the CF trans-membrane conductance regulator gene, which encodes for a protein involved in the regulation of salt and water movement across cell membranes. This mutation leads to the production of thick, sticky mucus that can clog the airways and obstruct the pancreas. Research efforts have significantly intensified since the identification of the CF trans-membrane conductance regulator gene, which harbors over 2000 distinct mutations. CF trans-membrane conductance regulator modulators, particularly transformative for patients with prevalent mutations like F508del, have the potential to prevent significant complications if initiated early in childhood. However, for individuals with rare CF trans-membrane conductance regulator mutations, the development of a treatment strategy remains to be established.
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**KEYWORDS:** cystic fibrosis; diagnosis; early screening; genetic mutations; survival rates.

## INTRODUCTION

Cystic fibrosis (CF) is a hereditary condition stemming from a faulty gene inherited from both parents. This gene, known as the cystic fibrosis trans-membrane conductance regulator (CFTR) gene, plays a crucial role in regulating salt movement across tissues. When this gene is flawed, it leads to either the absence or malfunction of the protein it produces. Consequently, there's a decrease in salt transport and water movement, leading to the accumulation of thick mucus in different areas of the body.(1) The CFTR gene is situated on chromosome 7's long arm. It generates a 6.5-kilobase messenger RNA (mRNA) from the normal allele, encoding CFTR, a 1490-amino acid protein integral to the cell membrane. This protein functions as a controlled chloride ion channel within the epithelial cells of various organs.(2) Since its cloning in 1989, more than 2000 CFTR variants have been documented in the CF Mutation Database(3). This database has been pivotal in establishing a mechanism-based classification system (classes I–VI) for CF, facilitating the identification of therapeutic targets. However, a newer initiative, the Clinical and Functional Translation of CFTR project (CFTR2), has emerged to evaluate the severity of CF in patients. CFTR2 researchers have collected global data from CF patient registries, functional studies, and clinical information to standardize the annotation terminology of CFTR variants.(4)

A comprehensive review and update on the diagnosis and management of lung disease in cystic fibrosis (CF) patients is provided. The sweat chloride test (SCT) remains the primary diagnostic tool for CF and should be conducted accurately. However, in some cases, SCT results may not definitively confirm a CF diagnosis. CF patients should receive ongoing care at specialized units staffed by multidisciplinary teams following standardized clinical protocols. This care should include regular lung function tests, microbiological analyses, and imaging studies. Recommendations for treating early-onset and chronic infections caused by common pathogens like Pseudomonas aeruginosa and Staphylococcus aureus, as well as less common pathogens, are outlined. Additionally, management strategies for other aspects of CF lung disease and associated complications are provided, along with guidelines for lung transplantation. This updated document, prepared by the CF working group of the Spanish Paediatrics Pulmonary Society, aims to revise and supplement previous publications in this journal from 1999.(5)

The National Institute for Health and Care Excellence (NICE) released guidance on "Cystic Fibrosis: Diagnosis and management" (NG 78) in October 2017. Our goal is to outline the main recommendations from this guidance for general pediatricians to incorporate into their daily practice. Additionally, we'll highlight other recent guidelines, including the European Cystic Fibrosis Society's "Standards of care: best practice guidelines" from 2018, the Cystic Fibrosis Foundation's recommendations on managing pulmonary exacerbations from 2009, and consensus documents from the UK Cystic Fibrosis Trust.(6)

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**CASE REPORT**

A 42-year-old male patient presented with complaints of coughing with thick mucus, wheezing, salty-tasting skin, and constipation over three days. He denied any history of fever. On examination, the patient appeared stable, but his oxygen saturation levels were below 90%, indicating inadequate oxygenation mentioned in the Table No. 1. The physician suspected cystic fibrosis and advised chest radiography and CT scan, which revealed right upper lobe vesicles, bilateral bronchiectasis with obvious exudative lesions, pulmonary fibrosis, and bilateral para sinusitis mentioned in the Fig. No. 1.

The patient was started on treatment as outlined in Table No. 2, which was continued for 8 days. On the 10$^{th}$ day, the patient's condition had improved, with oxygen saturation levels increased to 90%.

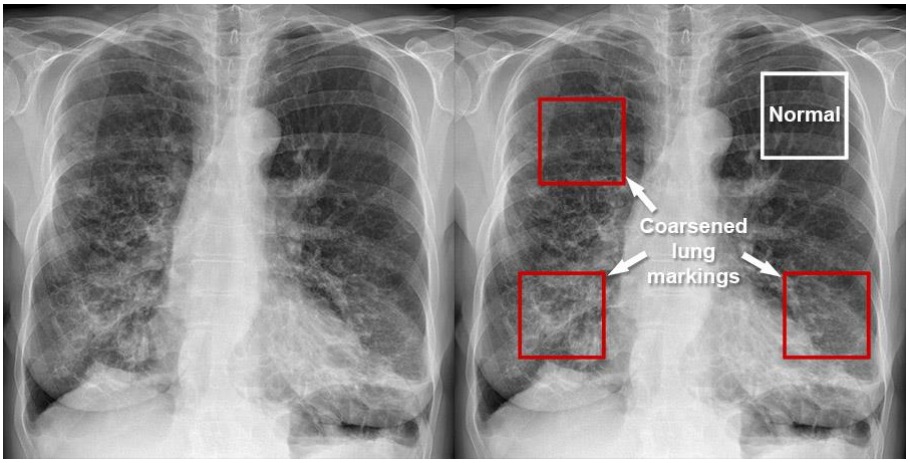


**Fig. No. 1: Chest X-ray- Pulmonary Bronchiectasis and Cystic Fibrosis**

**Table No. 1: Vitals on examination.**

| | |
|---|---|
| Temperature | Afebrile |
| Blood pressure | 110/90 mmhg |
| $Spo_2$ | 80% RA |
| Pulse rate | 120bpm |
| CVS | $S_1S_2$ |

**Table No. 2: Treatment provided during therapy for 1-8 days.**

| Drug prescribed | Generic name | Dose | Frequency | Route of administration |
|---|---|---|---|---|
| Inj.Pan | Pantoprazole | 40 mg | OD | IV |
| Inj. ondan | Ondansetron | 4 mg | OD | IV |
| Tab. Tezar | Tezacaftor | 100 mg | BD | PO |
| Inhaler | Salmeterol | 50 mcg | BD | PO |
| Inj. Azithrol | Azithromycin | 500 mg | BD | IV |
| Inj. Ibuprofin | Ibuprofen | 250 mg | BD | IV |

**DISCUSSION**

- **Genetics**: CF is inherited in an auto-somal recessive pattern, meaning a child must inherit two copies of the defective CFTR gene (one from each parent) to develop the disease. Individuals who inherit only one copy are carriers and typically do not show symptoms but can pass the gene to their children.

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- **Clinical Presentation:** Symptoms of CF vary widely and can manifest in different organ systems. Respiratory symptoms include chronic cough, wheezing, recurrent lung infections, and clubbing of the fingers. Digestive symptoms may include poor growth, malnutrition, greasy stools, and pancreatic insufficiency.
- **Diagnosis**: Diagnosis of CF typically involves a combination of clinical evaluation, sweat chloride testing (which measures the concentration of salt in sweat), genetic testing to identify CFTR mutations, and imaging studies such as chest X-rays or CT scans.
- **Treatment:** Management of CF aims to alleviate symptoms, prevent complications, and improve quality of life. This often involves a multidisciplinary approach including medications to thin mucus, antibiotics to treat infections, airway clearance techniques, nutritional support, pancreatic enzyme replacement therapy, and, in some cases, lung transplantation.
- **Prognosis:** While CF remains a life-limiting condition, advancements in treatment and care have significantly improved life expectancy and quality of life for individuals with CF. Many people with CF are now living into adulthood and pursuing careers and families.
- **Research and Future Directions:** Ongoing research continues to focus on developing new therapies aimed at targeting the underlying genetic defect in CF, improving lung function, and addressing complications such as chronic infections and inflammation.

Overall, cystic fibrosis is a complex condition that requires comprehensive management and support from healthcare professionals, caregivers, and support networks to optimize outcomes and quality of life for individuals affected by the disease.

## CONCLUSION

As cystic fibrosis is a genetic disorder affecting the lungs and digestive system, improvement in the patient condition was seen after prescribing a Tezacaftor (cystic fibrosis trans-membrane regulator) and supporting tests such as chest x-ray and CT scan. The outlook for individuals with cystic fibrosis has undergone significant improvement since its initial description in 1938. Nowadays, being diagnosed with cystic fibrosis is not necessarily a prediction of death within a year or two, and patients now have a fair chance of living into adolescence and adulthood. However, as patients with cystic fibrosis grow older, they encounter unique challenges that are not typically seen in pediatric patients. Therefore, it is crucial that specialized units are available to provide appropriate care for these older patients.[7]Epidemiological changes create new challenges for the management of cystic fibrosis. Approximately 10 % of patients still lack a therapeutic option. The community of researchers, pharmaceutical industries, patient associations, and health authorities remains committed to monitor the long-term effects of these still poorly characterised treatments, and to explore new pharmacological approaches, such as gene therapies[8].

## REFERENCES

1. Tori M.Endres; What is cystic fibrosis; JAMA network; 2022(January), 327(2), 191.
2. Christine E.Bear; Purification and function of the cystic fibrosis trans-membrane conductance regulator; Acell press journal; 1992(February),68(4), 809-818.
3. Stanley F LO; Adavance in thr diagnosis and management of cystic fibrosis in the genomic era; Clinical chemistry; 2018(June), 64(6), 898-908.
4. US cystic fibrosis foundation; John's Hopkins university hospital for sick children; Clinical and functional translation of $CFTR_2$; https://www.cftr.org\files\$CFTR_2$13 2015(August)..
5. Carroll E.cross; cystic fibrosis in adults; Clinical reviews in allergy and immunology; 2003(December),25,275-287.
6. Sahana rao; NICE guidance in diagnosis and management of cystic fibrosis; British medical journal; 2021(February), 106(1),31-34.
7. Brock DJ(1979); Methylum belli ferylguanidino benoate reaction protease and premature diagnosis of cystic fibrosis, Lancet, 1245.
8. Iwona pranke; Therapeutics in cystic fibrosis; Clinical revolution and new challenges; Med.science; 2024(March); 40,258-267.

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