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CASE REPORT

Case-Based Approach on Teenager with Cystic Fibrosis: Chronic Pulmonary and Pancreatic Complications

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Abstract

A 16-year-old girl with a history of inability to grow, perpetual breathing difficulties, and symptoms of pancreatic insufficiency presented to the clinic. Though her symptoms had lasted many years, a clear diagnosis was not made until recently. Homologous ΔF508 mutation discovered through genetic testing supported the diagnosis of cystic fibrosis (CF). Particularly in teenagers with mild or concurrent symptoms, this case illustrates the need for early diagnosis of CF. Furthermore, it stresses how important a multidisciplinary approach to therapy is in improving the quality of life and clinical results for CF patients.

INTRODUCTION

Depending on the area and cultural background, estimates of the incidence of cystic fibrosis (CF), a reasonably common autosomal recessive genetic condition, range from 1 in 10,000 to 1 in 40,000 live births in Indian origin [1]. The disease is brought on by mutations in the gene for the cystic fibrosis transmembrane conductance regulator (CFTR), which is essential for controlling the flow of chloride ions across the membranes of epithelial cells [2]. The equilibrium of salt and water on different epithelial surfaces is upset when chloride transport is compromised or non-existent due to CFTR protein

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defects. This leads to excessively thick, sticky, dehydrated secretions that mostly impact the lungs and gastrointestinal tract in people with cystic fibrosis. Prolonged respiratory infections, lung damage, and issues with malabsorption and pancreatic insufficiency are all caused by these recurrent discharges [3].

CASE PRESENTATION

History of Present Illness

The pulmonology clinic was referred to a sixteen-year-old girl who had been having persistent respiratory problems for six months. She has experienced several respiratory tract illnesses, each needing multiple antibiotic treatments. She also reported a persistent, productive cough and increasing dyspnoea during vigorous activity in addition to these recurring illnesses. Despite having a healthy satiety, she reported having trouble gaining weight and producing large, oily, and unpleasant-smelling excrement, which are signs of malabsorption. These characteristics prompted supplementary investigations because they aroused medical suspicion of an underlying systemic disease.

Previous Health History

Within 48 hours of birth, the patient had undergone surgery to treat neonatal meconium ileus. Pneumonia and bronchitis were prevalent in children, and Pseudomonas aeruginosa and Staphylococcus aureus were frequently growing in cultures. At the age of eight, the child was detected with chronic sinusitis, which led to multiple sinus surgeries.

Family History

Both parents had Northern Indian heritage, and there was no prior family history of cystic fibrosis. However, family reports state that a paternal uncle had passed away from "lung problems" when he was a toddler.

Physical Assessment

The patient appeared slender since their body mass index was in the 10th percentile for their age at presentation. Vital signs revealed mild tachypnea with a respiratory rate of 24 breaths per minute and an oxygen saturation of 94% on room air. A physical examination showed bilateral coarse crackles across the lung fields, barrel chest deformities, and clubbing of the fingers and toes. An abdominal examination revealed no splenomegaly but mild hepatomegaly. During the rhinoscopic examination, nasal polyps were discovered.

Laboratory and Diagnostic Studies

Sweat Chloride Test: The diagnosis of cystic fibrosis was confirmed by two different sweat chloride tests, which yielded readings of 89 mEq/L and 92 mEq/L (normal <30 mEq/L).

Genetic Testing: The most prevalent mutation that causes CF, homozygous ΔF508 mutations in the CFTR gene, were found by DNA analysis.

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Tests for Pulmonary Function: Spirometry showed a mild obstructive pattern, with a predicted FEV1 of 65% and an FVC of 78%.

Imaging Studies: Bilateral bronchiectasis, mucus plugging, and tree-in-bud opacities that are indicative of persistent cystic fibrosis lung disease were seen on high-resolution computed tomography of the chest.

Microbiology: Methicillin-sensitive Staphylococcus aureus and a mucoid strain of Pseudomonas aeruginosa were grown in sputum cultures.

Pancreatic Function: Severe pancreatic insufficiency was revealed by a significantly reduced faecal elastase level of 45 μg/g (normal >200 μg/g). All the fat-soluble vitamin levels (A,D,E,K) were below normal.

Treatment and Management

A comprehensive treatment plan was initiated involving multiple disciplines:

Pulmonary Management:

- Using high-frequency chest wall oscillation as a daily airway clearance therapy.
- Inhaled drugs such as dornase alfa, hypertonic saline, and alternate months of inhaled aztreonam and tobramycin.
- Three times a week, take oral azithromycin for its anti-inflammatory effects.
- Using intravenous antibiotics aggressively to treat pulmonary exacerbations based on culture sensitivity.

Gastrointestinal Management:

- Meals and snacks combined with pancreatic enzyme replacement treatment.
- Vitamin supplements soluble in fat.
- Diet that is heavy in calories and fat and includes nutritional coaching.
- To maximise pancreatic enzyme function, use a proton pump inhibitor.

Additional Therapies:

CFTR modulator therapy with elexacaftor/tezacaftor/ivacaftor was initiated given the patient's ΔF508 homozygous genotype

Regular exercise program to maintain pulmonary function

Psychosocial support and genetic counseling

Follow-up and Outcomes

After starting comprehensive CF care, the patient showed notable improvement at the six-month follow-up. Eight kilogrammes more weight was added, and the BMI rose to the 25th percentile. Tests of pulmonary function revealed improvement, with FEV1 rising to 78% of the expected level. Over the course of six months, the frequency of pulmonary exacerbations dropped from every six to eight weeks to twice. Validated CF-specific questionnaires showed a considerable improvement in quality of life scores.

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DISCUSSION

This instance highlights a number of crucial elements of diagnosing and treating cystic fibrosis. The need for greater awareness among healthcare practitioners is highlighted by the delayed diagnosis, even in the face of classic signs including infant meconium ileus and recurrent respiratory infections [6]. Since meconium ileus affects 15–25% of CF patients, its presence in the newborn period should trigger an early CF evaluation.

Pancreatic insufficiency and progressive pulmonary disease are among the severe disease phenotypes linked to the homozygous ΔF508 mutation [5]. An important advancement in the therapy of cystic fibrosis is the advent of CFTR modulator therapy, which targets the underlying protein problem instead of only treating its symptoms. For the best results, a multidisciplinary approach to CF management is necessary.

CONCLUSION

The significance of early detection and all-encompassing treatment of cystic fibrosis is illustrated by this case. Implementing evidence-based CF care, such as CFTR modulator medication, led to a notable clinical improvement despite the delayed diagnosis. Many of the problems this patient has can be avoided, and long-term results can be enhanced, by early diagnosis through newborn screening programs and timely beginning of specialised CF care.

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