ORIGINAL RESEARCH PAPER

Paediatrics

CASE REPORT OF CYSTIC FIBROSIS

KEY WORDS:

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INTRODUCTION

Cystic fibrosis is an inherited multisystem disorder of children and adults. The presuming incidence in India is 1 in 2500 live birth. Dysfunction of CFTR protein leads to variable presenting manifestation and complication including severe lung disease and exocrine pancreatic insufficiency. Low level of suspicion and poor availability of facilities have delayed the diagnosis in India. Delay diagnosis of cystic fibrosis results in severe malnutrition, which is one of the bad prognostic indicators for survival.

Pathogenesis

CFTR is a member of the adenosine triphosphate binding cassette function as chloride channel and other regulatory functions affected by different mutations. The most prevalent mutation of CFTR is the deletion of a single phenylalanine residue at amino acid 508(F508del) CF gene code for CFTR protein expressed largely in epithelial cell in airway, gastro intestinal, sweatgland and genitourinary system. Mutation leads to inability to secrete chloride or bicarbonate, which impairs clearance of mucous secretion, elevated salt content in sweat, and paucity of water in mucous secretion.

Clinical Features

Diagnosis

Presence of typical features (respiratory, gastro intestinal or genitourinary) OR a history of CF in a sibling OR a positive newborn screening test PLUS laboratory evidence for CFTR dysfunction: two elevated sweat chloride concentrations obtained on separate days or identification of two CF mutations or an abnormal nasal potential difference measurement.

Clinical

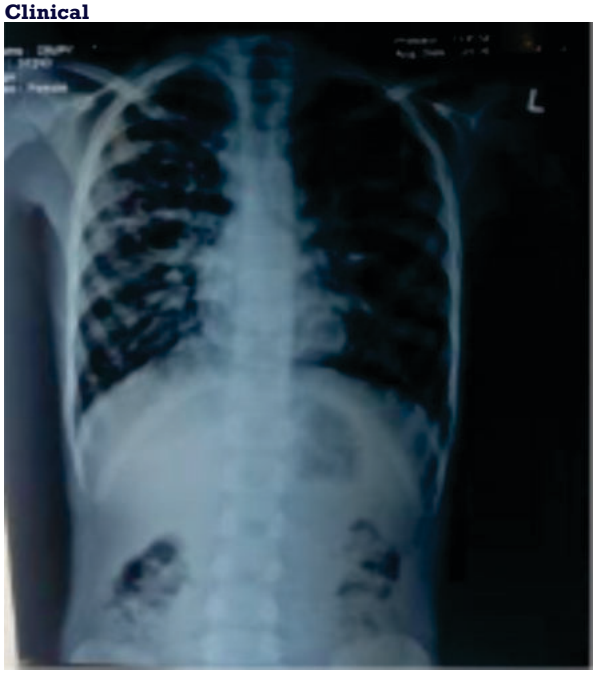

Figure 1: B/L lung field show patchy haziness with few cavitatory lesion s/o infective etiology.

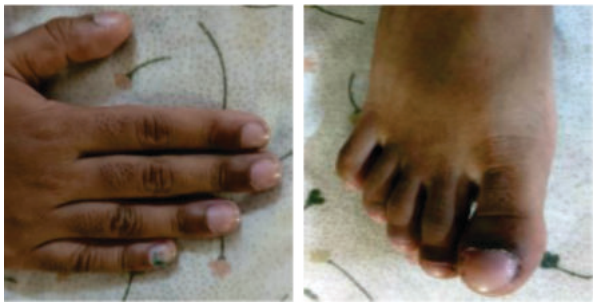

Figure 2 : Changes of clubbing

Figure 3: Genetic testing

Case Presentation

8 year old female patient is the 2nd child of non consagious marriage. The first child was undiagnosed, had similar complain since 4 months of age, died at 1.5 years of age. Patient had uneventful course till 5 months of age, presented with complain of cough, cold, fever and breathing difficulty diagnosed as pneumonia. After 1 month again presented with pneumonia with gall bladder stone. Subsequently repeated admission for similar complain added on that h/o hyponatremic convulsion at 1.5 year of age. Over the time complain of progressively increasing sputum production with chronic cough became a clinical problem with exercise intolerance and recurrent episodes of loose stool and poor weight gain. CT thorax s/o multiple intrapulmonary nodule with changes of bronchiectasis in B/L lower zone. Due to multiple episodes of similar complain genetic testing was done, which is positive for CFTR mutation for 2 genes : 2044delA(14) and 3700A>G(22) , heterozygous. Culture of sputum and blood suggestive of higher colonization with pseudomonas, methicillin resistance staphylococcus areus, once positive for allergic bronchopulmonary aspergillosis.

Treatment & Prognosis

Therapy is directed to prevent pulmonary complication and nutritional maintenance and education of the patient and parents. Intravenous therapy for dehydration should be initiated early. Inhalational therapy with hypertonic saline, airway clearance therapy like chest PT and antibiotics ,antifungal, Anti-inflammatory agents, pancreatic enzyme replacements and vitamins and mineral supplements given. Newer modalities including CFTR modulator therapies with IVACAFTOR, LUMACAFTOR, TEZACAFTOR.

REFERENCES

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