



Case Report

Unusual Presentation of Cystic Fibrosis

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ABSTRACT

Cystic fibrosis is common disorder in Indian children. The presenting manifestation is lengthy, although pulmonary and gastrointestinal presentations predominate. Only gastrointestinal symptoms without any respiratory symptoms are rarely documented. The diagnosis is often delayed if presentation is atypical. We describe here, a case of a 9 years old male child with only gastrointestinal symptom which can be considered as an atypical presentation of cystic fibrosis.

Key words: Cystic fibrosis, Sweat chloride test, wrinkling.

INTRODUCTION

Cystic Fibrosis (CF) is most common life limiting autosomal recessive multisystem disorder. The clinical features of cystic fibrosis are variable and depend on the age and the organs affected at the time of diagnosis. Patient with atypical disease tend to present with symptoms late in childhood or as an adult. The common presentation include meconium ileus and failure to thrive in neonatal period, recurrent bronchiolitis in infancy and recurrent lower respiratory tract infections, chronic lung disease, bronchiectasis, failure to thrive, steatorrhea and rectal prolapse in early childhood. [1] They usually present as poor growth. Although the respiratory and gastrointestinal symptoms are well known, only

gastrointestinal symptoms without any respiratory symptoms are rarely documented.

CASE REPORT

A 9 years old Indian male child presented to outpatient department (OPD) with chief complaints of wrinkling of fingers immediately within few seconds of commencement of food and increase in frequency of passing of oily and sticky stools since 2 years. The patient had visited many private practitioners before being referred to us for not gaining adequate weight and height. He was born at term, with average birth weight to Indian consanguineous parents. His neonatal period was uneventful. He was exclusively breast

feed for 6 months. According to mother he was thriving well and no past medical history was noted. There was no history of any major lower respiratory tract infections. He has one elder sibling; she is 17 years old medically and physically fit. We admitted him for detailed examination and investigations. Clinical examination revealed anthropometry far below his accepted level, malnourished with proportionate short stature (weight 18 kg and height 122 cm, which was less than 3rd percentile), wrinkling of fingers and rectal prolapse. Respiratory examination was unremarkable. Several investigations were done to rule out causes of short stature. Complete blood count revealed microcytic hypochromic anaemia. Keeping celiac disease as one of close differential diagnosis, duodenal biopsy was carried out which revealed normal mucosa. Tissue transglutaminase-IgA was negative. Three days stool collection revealed fat but not in excess amount. Sweat chloride test on two different occasions was more than 60 mEq/L (84 mEq/L and 82 mEq/L). However genetic analysis performed in a research laboratory failed to identify any mutation.

As diagnostic criteria for cystic fibrosis was fulfilled, he was treated with enzyme replacement therapy. Within few months he has started gaining weight and height.

DISCUSSION

Cystic fibrosis is inherited as an autosomal recessive trait. The disease is responsible for wide varieties of respiratory and exocrine pancreatic symptoms or problems as the underlying defect is in chloride transport protein which lines ductular epithelium. [1] The hallmark of cystic fibrosis is salty tasting skin but our patient presented with wrinkles on fingers. This was supported by few authors suggesting it was due to the excessive salt

concentration which increased the water binding capacity of keratin. [2]

False positive result of sweat chloride may be seen in condition such as malnutrition, Addison's disease, anorexia nervosa, ectodermal dysplasia etc. In our case the chance of false positive result was remote as on two different occasions the sweat chloride was more than 60 mEq/L. [2]

The chromosome 7 has been identified as the gene for cystic fibrosis with DF508 as commonest mutation. Facilities for identification of mutations are not readily available in India. One of the Indian study showed frequency of DF508 mutation is 19%. [4] Therefore inability to demonstrate mutations or negative genetic mutation does not entirely rule out the disease. [5] Hence sweat test remains the first line as diagnostic test. Cystic fibrosis does occur in Indian children as earlier thought; this disease is not rare in India. Several published reports support that cystic fibrosis is common disease in Indian children. [6-8] Delayed diagnosis may be due to lack of awareness of the existence of disease and under diagnosis or missed diagnosis as in majority of cases, which may contribute to poor prognosis. Our case highlights the importance of considering the diagnosis of cystic fibrosis in children presenting with wrinkling of fingers and poor growth.

CONCLUSION

There is need to create awareness about occurrence of cystic fibrosis in Indian children as with atypical presentation diagnosis is often delayed and the disease is advanced in most patients at the time of diagnosis. Screening of cystic fibrosis in newborn should be made mandatory which can be carried on Guthrie card after birth.

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