

CYSTIC FIBROSIS IN ONE FAMILY

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SUMMARY

Two patients with cystic fibrosis from the same family are reported. Both patients had features of chronic respiratory disease with evidence of pancreatic and other systemic manifestations. To our knowledge this is the first report from our country of cystic fibrosis multiplex cases occurring in the same family.

Key Word: Diabetes mellitus

INTRODUCTION

Cystic fibrosis (CF) is an autosomal recessive disorder characterised by multisystem exocrine dysfunction.¹ The single most important test for diagnosis is the sweat chloride test. The manifestations of CF are protean but obstructive respiratory disease, malabsorption due to pancreatic insufficiency and failure to thrive are more common.² CF is no longer solely a paediatric disease because because early diagnosis, good paediatric care, availability of newer antibiotics and good physiotherapy have resulted in a marked improvement in the survival period of patients, many of whom reach adulthood.

Case Report

Case 1: A 13 year old boy born of consanguineous parents was admitted for cough and wheezing, since the age of three years. Cough was productive at times with mucoid sputum and there was an episode of haemoptysis three months prior to his visit to hospital. He was diagnosed as an 'asthmatic' and was treated with bronchodilators. There were no other relevant respiratory or cardiac symptoms. He had recurrent abdominal pain with bulky frothy stools of one year duration. He was not a known diabetic.

On examination, he was emaciated, with stunted growth. The basal metabolic index (BMI) was 12.2 kg/m². He was not anaemic or cyanosed but had severe carpedal clubbing. The pulse and BP were normal and the respiratory rate was 40/min. Respiratory examination revealed barrel shaped chest with bilateral extensive wheeze. The liver was palpable 3 cm below the right costal margin and all other systems were normal.

Investigation: The sweat chloride test was elevated on two occasions 147 mEq/l and 89 mEq/l (normal 70), confirming the diagnosis of CF.

Sputum for AFB and culture, and Mantoux test were negative. X-ray sinuses showed bilateral maxillary sinusitis. X-ray chest heart

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normal; (R) hilar gland was enlarged; multiple small cysts with opacities in the (R) apex were seen. There was a small cavity in the (L) apex with opacities in the (L) midzone. Lung function tests: FVC 0.54 l; FEV₁ 0.35 l; FEV₁/FVC% 65%; PEFR - 0.7 l/sec. Impression: Obstructive lung disease. Arterial blood gas analysis; PaO₂ 52.2 mmHg; PaCO₂ 45.8 mmHg; pH 7.26. Impression: hypoxaemia. Barium meal showed malabsorption pattern. Serum amylase was mildly elevated - 214 Somogyi units (Normal - 60-180). Faecal chymotrypsin was normal - 27.1 u/g (Normal >6). Ultrasonography of the pancreas showed evidence of chronic pancreatitis.

The glucose tolerance test was normal and the glycosylated haemoglobin was 8.0% (Normal - 5-8%). The fasting and post-prandial C-peptide levels were 0.4 pmol/ml (Normal - 0.6-1) and 1.0 pmol/ml (normal - 1.5) respectively. The fasting IRI was 12 uU/ml (normal - 6-24) and the post-prandial 88 uU/ml (normal - 50-90).

Case 2: A 19 year old man, the elder brother of the first patient, was screened for CF, since he also had chronic respiratory symptoms and was a known diabetic on insulin for 5 years. He had two episodes of haemoptysis in the past for which he was given antituberculous drugs. He had symptoms of sinusitis since 2 years but denied any history of bulky frothy stools.

On examination, his BMI (kg/m²) was 14.2. There was clubbing of the fingers. The pulse and BP were normal. Respiratory examination showed a barrel shaped chest with a chest expansion of 3 cm and scattered rhonchi bilaterally. Other systems were normal except for a palpable liver 3 cm below the right costal margin.

Investigation: The sweat chloride test was high on two occasions - 140 mEq/l and 68 mEq/l confirming the diagnosis of CF.

Sputum for AFB and culture and Mantoux test were negative. X-ray sinuses showed pansinusitis. X-ray chest showed evidence of scoliosis; the heart was normal. Cystic lesions were seen in the (L) midzone. Lung function tests: FVC - 1.27 L; FEV₁ - 1.03 L; FEV₁/FVC 82%; PEFR - 1.5 L/sec. Impression: restrictive lung disease. Arterial blood gas analysis PaO₂ 105.4 mmHg PaCO₂ - 43.4 mmHg pH - 7.31. Impression - normal.

Barium meal test was normal. Serum amylase was high 310 Somogyi units. Faecal chymotrypsin was normal - 26.1 u/g. Ultrasonography of the pancreas showed evidence of chronic pancreatitis.

The fasting and post-prandial blood sugar levels were 160 mg/dl and 400 mg/dl respectively and the glycosylated haemoglobin was 13.6%, indicating poor glycaemic control. The fasting and post-prandial C-peptide levels were 0.06 pmol/ml and 0.19 pmol/ml respectively.

Seminal analysis showed evidence of azoospermia.

Discussion

CF is the commonest genetically inherited condition in caucasians, with an incidence of 1:2000 live births. The gene for CF is located in the long arm of chromosome 7 and is closely linked with other genetic markers, e.g. with the PON gene for the enzyme paraoxonase.³ It was earlier believed that CF is rare among Indians.¹ Studies by Reddy *et al*⁴ suggested that this disorders is not as rare as was believed to be in this country. There have been reports from Madras on a case of cystic fibrosis with multisystem involvement⁵ as well as on with beta cell in CF.⁶

In this paper, we report two cases of CF multiplex from the same family. The first patient had predominant respiratory disease with pancreatic exocrine abnormality, but normal endocrine and exocrine dysfunction. C-peptide assay confirmed the poor pancreatic beta cell function and ultrasonography showed evidence of chronic pancreatitis. Two our knowledge, this is the first report of ,ultiplex cases of CF reported from this country.

There are several other points of interest in our cases. One of the patients is 19 years old and has overt diabetes mellitus. Diabetes mellitus is rare and occurs in only 1 in 130 cases of cystic fibrosis.⁷ The clinical history of passing oily stools plus the ultrasound evidence of chronic pancreatitis are suggestive of involvement of the exocrine pancreas in these patients. The faecal chymotrypsin levels were normal but this is not surprising as the test becomes abnormal only in advanced pancreatic insufficiency.

The other interesting features of our patients are the occurrence of CF arthropathy in one patient and azoospermia in the other. CF arthropathy is a newly recognised entity and occurs in 7-8% of CF patients. Absent vas deferens and absent epididymis has been described as part of the constellation of pathological changes in CF, which explains the occurrence of azoospermia.

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BOOK REVIEW

Advances in Dermo-venereo-Leprology -1st Ed. 1992. Editors PS Shankar, BM Biradar, published by PM Biradar, Organising Secretary, Third South Zone Conference of Dermatologists, Venerologists and Leprologists, MR Medical College, Gulbarga 585 105 Total pages 115.

A collection of review articles on Dermatology, Venereology and Leprology is presented. The ground covered includes an approach to a patient with photosensitivity with photopatch testing, interesting articles on ageing and the skin, psychodermatology, skin problems in chronic renal failure and newer advances in the treatment of psoriasis. In the venerology section the causes of nonvenereal dermal lesions on the genitals is discussed as well as cutaneous manifestations and an Indian experience of the HIV virus. The need for early diagnosis of leprosy and health education in leprosy patients is emphasised. These articles may be briefly gone through by the physician to give him a wider knowledge in his practise of medicine.

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