CASE REPORT

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Case report of a middle-aged man with bronchiectasis, situs inversus and cirrhosis

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Abstract

Ciliopathies are anatomical and functional disorders of the cilia in various organ systems such as the urinary and respiratory tracts. Ciliopathies involving the respiratory system include primary ciliary dyskinesia and situs inversus totalis. Patients present with recurrent infections of the middle ear/sinuses, and of the upper and lower respiratory tracts. Chronic infections result in atelectasis and bronchiectasis. When bronchiectasis occurs with impaired fertility and sinusitis, Young's syndrome, Kartagener's syndrome and cystic fibrosis may be considered. When bronchiectasis coexists with cirrhosis, alpha1antitrypsin deficiency is generally thought of. Our patient, a middle-aged teetotaller, presented with abdominal distension and pedal edema. His past history was significant for recurrent upper and lower respiratory tract infections and treatment with antituberculous therapy. Investigations suggested Kartagener's syndrome. Cirrhosis due to hepatitis B contributed to his ascites. Bronchiectasis related corpulmonale and amyloidosis were ruled out. Chronic illnesses with new symptoms may unexpectedly yield other coexisting diseases.

Key words

Alpha1 antitrypsin deficiency, bronchiectasis, ciliopathy, cirrhosis, Kartagener's syndrome.

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Background

Bronchiectasis is a chronic inflammatory and fibrotic lung disease caused by infections, immune deficiency, immune mediated disorders and inherited diseases [1]. The prevalence is not known. Congenital disorders causing bronchiectasis include alpha1 antitrypsin deficiency and cystic fibrosis (cellular or biochemical defect), primary ciliary dyskinesia and Kartagener's syndrome (ciliary defect), William–Campell syndrome (structural defect) and hypogammaglobulinemia (immune deficiency syndromes) [1, 2]. Concomitant bronchiectasis and cirrhosis, when congenital can be associated with alpha1 antitrypsin deficiency, wherein the emphysema leads to bronchiectasis; occasionally bronchiectasis precedes the emphysematous changes, but develops during adulthood [2]. Bronchiectasis, sinusitis and impaired male fertility are seen in disorders such as primary ciliary dyskinesia (PCD), Young's syndrome and cystic fibrosis. In PCD, a triad of situs inversus, bronchiectasis and sinusitis, constitutes Kartagener's syndrome which was first reported in 1904 [1]. Inherited bronchiectasis and acquired cirrhosis is an unusual combination in a middle-aged adult and such a patient is described in the following report, when he presented with a symptoms suggestive of corpulmonale. There are no similar such reports from India or Asia.

Case Report

This 52 year old man presented to the out-patient Department of General Medicine, Indira Gandhi Medical College & RI, Kadirkamam, Pondicherry, with history of pedal edema, abdominal distension and breathlessness of two months' duration. Past history was significant for recurrent upper and lower respiratory tract infections since childhood. He had received 6 months' antituberculous therapy 30 years ago. He was a teetotaller and had never smoked. His only child was born 18 years after his marriage. On examination: pulse 94/minute, blood pressure 96/70mm Hg, respiratory rate 24/minute, pallor, pan-digital clubbing, pedal edema, non-palpable cardiac apex, bilateral coarse crepitations, scattered wheeze and ascites were seen. Investigations revealed hemoglobin 8.5 g/dL, total leukocyte count 93x109 /L with 67% neutrophils, platelets 130x109/L, total bilirubin 1.6mg/dL, protein 5.7g/dL, albumin 2.8g/dL, SGOT 76U/L, SGPT 45U/L, alkaline phosphatase 116U/L, urea 14.2mmol/L, creatinine 106µmol/L, albuminuria(1+), ascitic fluid protein/albumin 2.4/1.1g, cell count 36/µL, with 90% lymphocytes. Situs inversus, bronchiectasis, maxillary sinusitis and cirrhosis were observed on imaging (Figure 1). HbsAg was positive, while HBeAg was negative. Echocardiography was normal. He was treated with inhaled bronchodilators, spironolactone, furosemide, pantoprazole, vitamins, and chest physiotherapy. He was unwilling for upper gastrointestinal endoscopy.

Discussion:

Ciliopathies

Cilia are seen in the respiratory tract, cerebral ventricles, and female oviducts and in spermatoazoa. Ciliopathies are either motor or sensory, and arise due to anatomical or physiological dysfunction of cilia (ultrastructural defects) in organs-systems such as the respiratory and genito-urinary tracts [3]. Examples of such disorders include autosomal recessive primary ciliary dyskinesia (bronchiectasis, sinusitis, and otitis media) and autosomal dominant polycystic kidney disease. Absence or truncation of the dynein arms of the cilia is the most common abnormality seen in patients suffering from PCD. Impaired mucosal clearance leads to frequent sinopulmonary infections and subsequently, bronchiectasis due to inflammation and lung damage. Kartagener's syndrome is the triad of sinusitis, bronchiectasis and situs inversus.

Bronchiectasis-sinusitis-impaired male fertility

A combination of these manifestations yields a differential diagnosis of cystic fibrosis (CF), Young's syndrome and PCD/ Kartagener's syndrome [2]. All these three conditions may have both bronchiectasis and sinusitis; azoospermia is seen only in the first two conditions, while immotile spermatozoa is observed in PCD. Defective sperm flagella movement leads to male infertility, but not always [3, 4]. Additionally, the vas deferens is absent in cystic fibrosis and congenitally obstructed in Young's syndrome. Our patient had a child born 18 years after his marriage. There are very few reports of male fertility in Kartagener's syndrome. Situs inversus is seen only in PCD, and hence this diagnosis was favoured in our patient. The bronchiectatic pattern in cystic fibrosis involves the upper lobes predominantly which was also not seen in our case.

Bronchiectasis-cirrhosis

There have been many case reports describing the association between these two conditions in regard to alpha1 antitrypsin deficiency (AT1) [5, 6]. AT1 deficiency has been reported to cause liver disease in both children (neonatal hepatitis) and adults. Our patient did not have features of prior chronic liver disease. Except for HBsAg, his other workup returned negative. We did not have facility for genetic testing to diagnose AT1 deficiency. With chronic respiratory symptoms and new-onset ascites, corpulmonale and renal amyloidosis was considered, but the man was finally diagnosed to have an inherited and acquired disease each.

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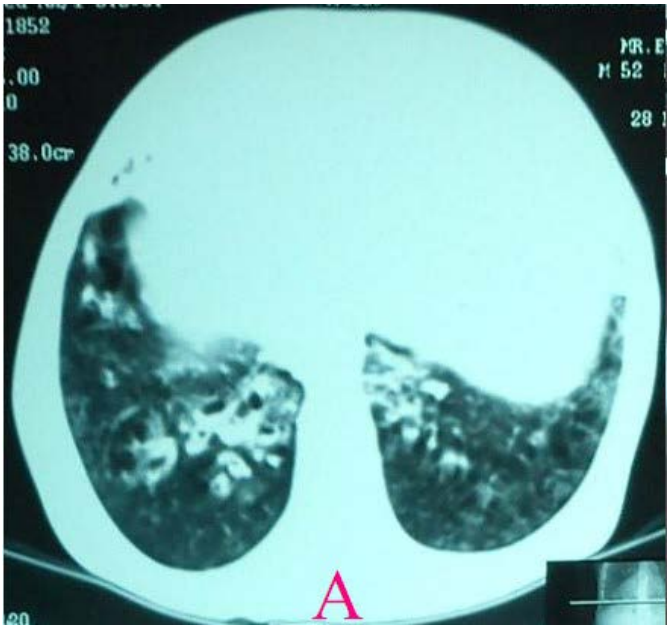
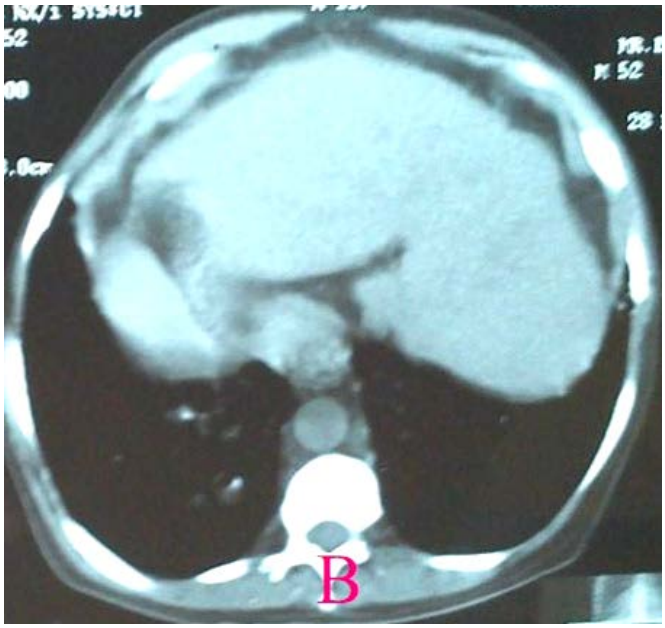
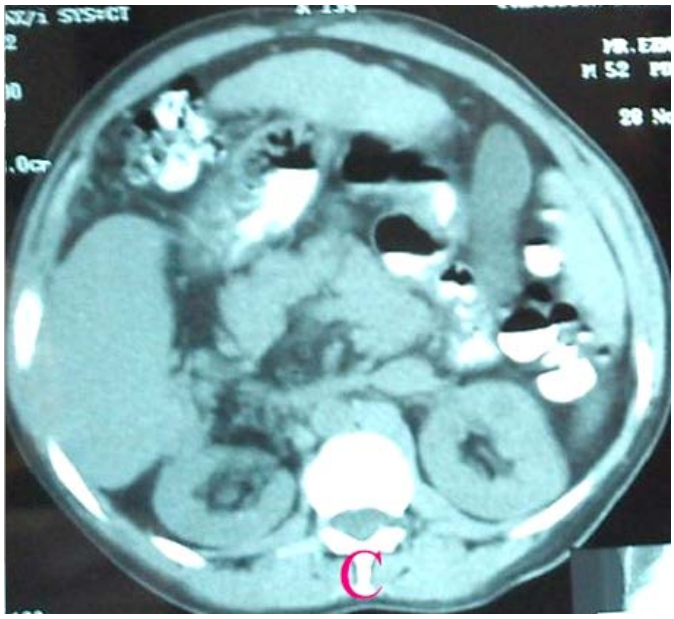
Figure - 1 - Computed tomography of chest and abdomen done one month after discharge, revealed



1 - A - bilateral bronchiectasis



1 - B - Left-sided liver with small right lobe of liver with hypertrophied caudate and lateral segments of left lobe, and surface nodularity



1 - C - Right-sided stomach and spleen with mild enlargement.

Conclusion

Inherited disorders can present much later in life than one might expect to. Incomplete evaluation in this man led to one course of antituberculous therapy. History of chronic sinopulmonary infections from childhood should make onesuspect immune deficiency, cystic fibrosis, AT1 deficiency, PCD and congenital heart disease. With a finding of bronchiectasis, impaired fertility and sinusitis, CF, PCD and Young's syndrome needs to be considered. Patients with chronic illness presenting beyond 50 years of age may still have an inherited disorder. New symptoms in such patients may herald a complication, further progression of disease or discovery of another acquired disorder.

Abbreviations

Alpha1 antitrypsin (AT1), Cystic fibrosis (CF), Primary ciliary dyskinesia (PCD).

Competing interests

None declared.

Authors' contribution

Data acquisition, literature search, manuscript drafting and editing, final approval – performed by the sole author.

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