Case Report

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Situs Inversus Totalis with Chronic Respiratory Ailment in a Fertile Male

Internal Medicine Section

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ABSTRACT

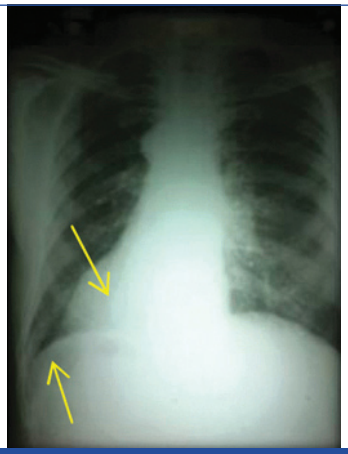
Primary Ciliary Dyskinesia (PCD) is a rare ciliopathic autosomal recessive genetic disorder that causes defect in the action of cilia lining the respiratory tract. In a total of 50% cases of situs inversus totalis fertility is preserved. Here, we report a case of situs inversus totalis with chronic respiratory ailment in a fertile male. A 60-year-old non smoker male, father of four children presented with recurrent episode of infection for past five years. Investigation was done which showed situs inversus totalis (Dextrocardia) with right lower lobe bronchiectasis. He was managed with antibiotics, mucolytics, bronchodilators and chest physiotherapy. Patient status improved well with above mentioned treatment.

Keywords: Bronchiectasis, Dextrocardia, Primary ciliary dyskinesia

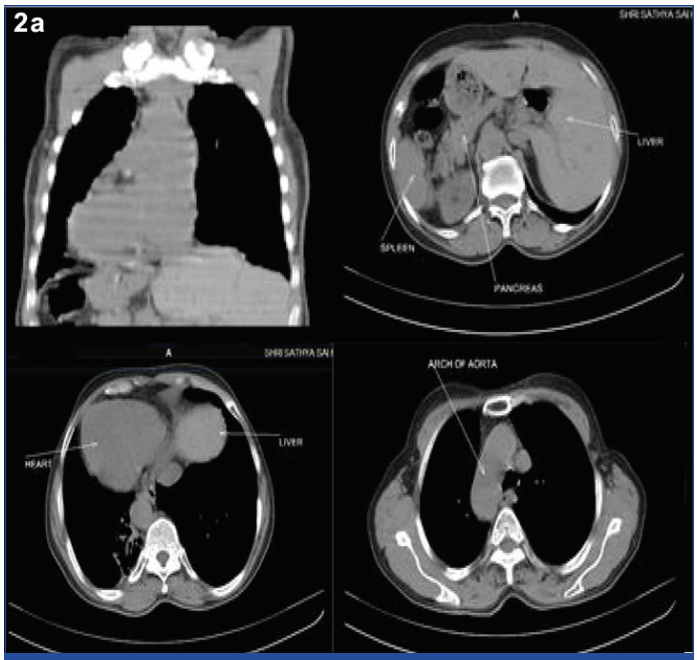
CASE REPORT

A 60-year-old, non smoker male presented with complaints of cough with expectoration and with history of recurrent episode of infections for past five years which were treated symptomatically. Patient had no history of haemoptysis, chronic constitutional symptoms or taking anti-tubercular treatment. He was born out of non-consanguineous marriage, had four children, and is farmer by occupation.

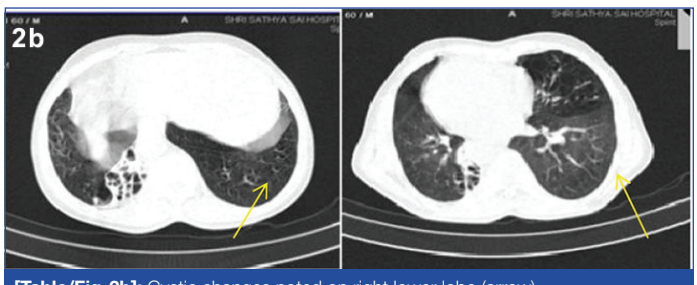
On examination, he had grade II clubbing, vitals were stable, traube's space was obliterated on left side, liver dullness absent on right side. On auscultation coarse crepitations were heard on right lower hemithorax and heart sounds were best heard on right side of the chest. Blood count was normal and sputum for acid fast bacilli was negative. His chest X-ray [Table/Fig-1] showed dextrocardia and fundus gas was noted on right side. Further, CT scan [Table/Fig-2a,b] was taken which showed dextrocardia, arch of aorta on left, liver on left side with cystic changes appreciated on right lower lobe. X-ray PNS revealed hypoplastic frontal sinus bilaterally [Table/Fig-3]. ECG was taken dextrocardiac features were noted [Table/Fig-4]. For further evaluation USG abdomen and bronchoscopy was done which showed situs inversus totalis with simple renal cyst and inversion of bronchial pattern with mucopurulent secretion noted on right lower lobe respectively. For assessing ciliary function saccharin test was done, test prolonged for one hour twenty minutes. Final diagnosis was primary ciliary dyskinesia, hypoplastic frontal sinus, right lower lobe bronchiectasis, situs inversus totalis (no structural defect).



[Table/Fig-1]: Dextrocardia, fundus gas was noted on right side (arrow).

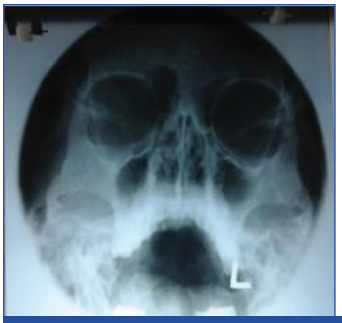
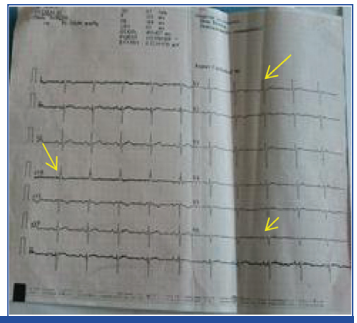


[Table/Fig-2a]: CT dextrocardia, arch of aorta on left, liver on left.



[Table/Fig-2b]: Cystic changes noted on right lower lobe (arrow).

Initially he was managed with antibiotics; Inj.Augmentin 1.2 gm IV BD with T.Levofloxacin 500 mg 1OD, Inj.Deriphylline 2 cc IV BD with Nebulisation, chest physiotherapy and postural drainage. He was clinically and symptomatically better after the above treatment and was discharged with oral antibiotics Tab.Ciprofloxacin 500 mg BD, mucolytics, metered dose inhaler combination of LABA and ICS. Patient was advised for follow up after 1 week. He was symptomatically better and had an improved quality of life. Subsequently family screening was done for one of his daughters using saccharin test which turned out to be normal.

[Table/Fig-3]: X-ray PNS hypoplastic frontal sinus bilateral.
[Table/Fig-4]: ECG with unreversed leads showing positive wave in aVR and tall R wave in V1 and absent R wave in V6. (Images from left to right)

DISCUSSION

Bronchiectasis is a pathological description of lung characterized by inflamed and dilated thick walled bronchi from distal to terminal bronchi. Poor ciliary structure or function result in impaired clearance of mucus leading to bronchiectasis, PCD is characterised by chronic rhino sinusitis, chronic bronchial sepsis [1]. Structural or functional changes of cilia end up in poor clearance of mucus which may contribute to nasal polyposis, otitis media and chronic sinusitis, further leading to bronchiectasis [2]. Immotile cilia syndrome is an autosomal recessive disorder characterised by poor motility of cilia in airway and sperms. PCD is a disease characterised by change in ciliary beat pattern, ciliary beat frequency or both and has a heterogenous genetic basis. The prevalence of PCD ranges from 1:2,200 to 1:40,000 [3,4]. Investigations show the congenital defect in the cilia and sperm tail which cause chronic respiratory tract infection and male sterility [5]. In a study conducted on 12 men with PCD, six with dextrocardia, who presented with upper and lower respiratory tract infections; four had normal spermatozoa. The possible mechanism for this paradox could be associated with genetically mediated heterogeneity influencing the phenotypic presentation [1]. Normal respiratory cilia and spermatozoa with Kartagener's syndrome explain the genetic heterogeneity and unreliable presentation of phenotype [6].

Subgroup of immotile cilia syndrome (Kartagener's syndrome) is associated with situs inversus, chronic sinusitis, and bronchiectasis. Young's syndrome, however is characterised by congenital epididymis obstruction with bronchiectasis and differs from immotile cilia syndrome by the absence of ultra structural cilial disorder [7,8]. The lower respiratory tract disease which is seen in young's syndrome does not worsen without any attributable evidence of microbial culture; they mostly present with testicular dysfunction and normal sweat chloride test value [9]. Male patients with Kartagener's syndrome invariably present infertility while women present reduced fertility. Most infertile patient with Kartagener's syndrome have normal spermatozoic count, but with a structural defect and complete lack of motility [10].

PCD is a disease of serious threat for lung function, early diagnosis did not protect against decline in lung function. About 50% of males are sterile [11,12]. PCD/kartagener's syndrome is usually associated with many complications which affect patient's quality of life. Medical approach to the disease aims to prevent further progression of infections with an appropriate treatment and timely diagnosis which may help to delay the progression of lesion [13,14]. Mainstay of treatment for PCD involves airway clearance, infection control and different techniques that guarantee airway clearance including chest physiotherapy and postural drainage. More rapid, accurate, effective and economic means of diagnosis will be helpful in low resource settings [15,16].

CONCLUSION

Kartagener's syndrome (PCD) is usually reported with infertility, but preserved fertility has been rarely reported. The cause of bronchiectasis must be searched meticulously especially in the setting of recurrent infection. The aim of reporting the case is a crude association between bronchiectasis with preserved fertility which is rarely seen in Kartagener's syndrome.

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