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Kartagener Syndrome

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Abstract: Kartagener syndrome with the triad of situs inversus, sinusitis and bronchiectasis develops due to the basic defect of primary ciliary dyskinesia (PCD). The PCD leads to recurrent upper and lower respiratory tract infections and bronchiectasis. Bronchial asthma if coexistant further complicates the condition with dyspnea and viscid secretions. It is very important to recognize this defect in mucociliary clearance and manage the patients with early antibiotics, mucolytics, bronchodilators and physiotherapy to prevent further lung damage.

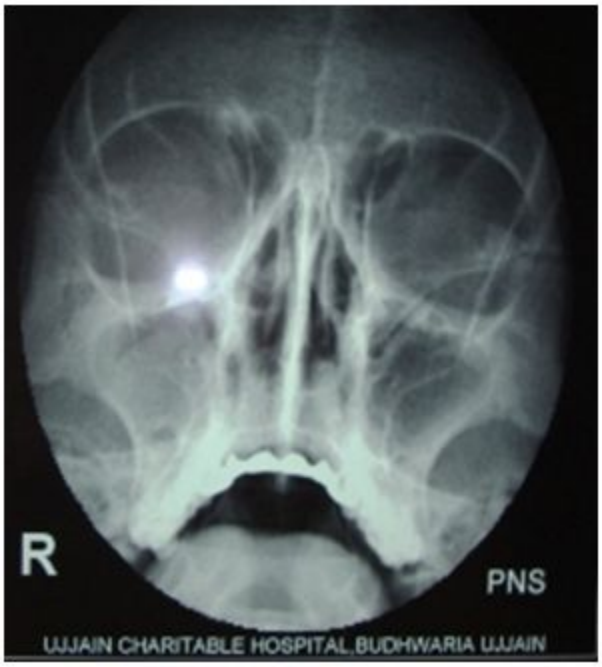
Introduction: A rare case of situs inversus with long standing naso-bronchial allergy on irregular treatment who presented to OPD in severe respiratory distress.

Care Report: A 31 year old married woman presented to the hospital with severe respiratory distress. On perusal of her old records it was found that she had dextrocardia. She had a previous history of recurrent episodes of cough with mucopurulent expectoration, sinusitis and progressive breathlessness since childhood. She gave a history of seasonal variation of sneezing and wheezing during the winter months. She was not on any regular treatment only taking medications on exacerbation. She had failed to conceive and been evaluated for primary sterility.

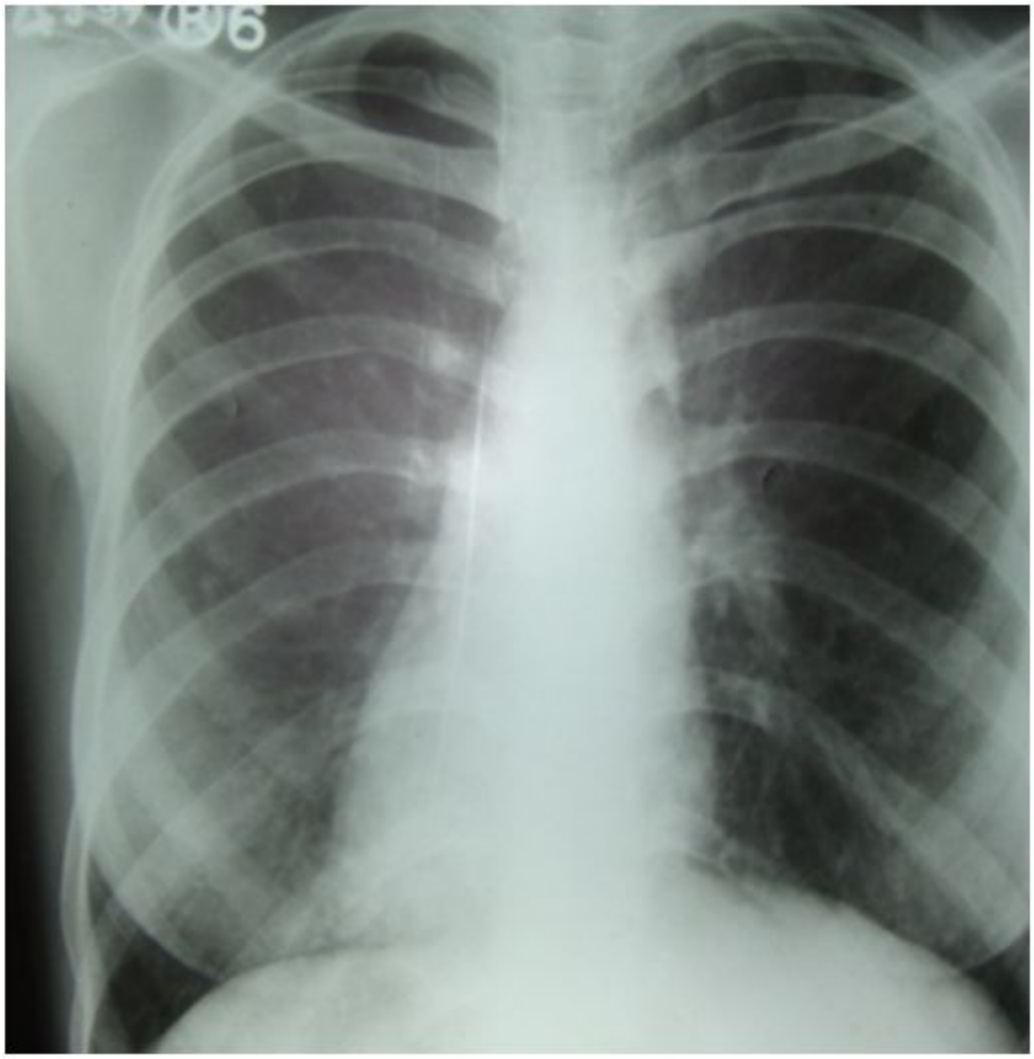
On physical examination the patient was averagely built and nourished. She was tachypenic with a respiratory rate of 34 per min. Her vital signs were: Temperature of 103 degrees Fahrenheit, blood pressure of 130/88 mm of Hg. Jugular venous pressure was raised and pedal edema was present. Nasal examination revealed edematous mucosa and there was sinus tenderness. On examination of the patient's chest, vesicular breath sounds were heard bilaterally with coarse crepitations & rhonchi. Cardiac auscultation revealed a S1 and S2 sound with a loud second heart sound. No rubs or murmurs were heard. Abdominal Examination revealed tender hepatomegaly. CNS examination was normal.

She was suspected to have underlying defective ciliary functions and evaluated accordingly.

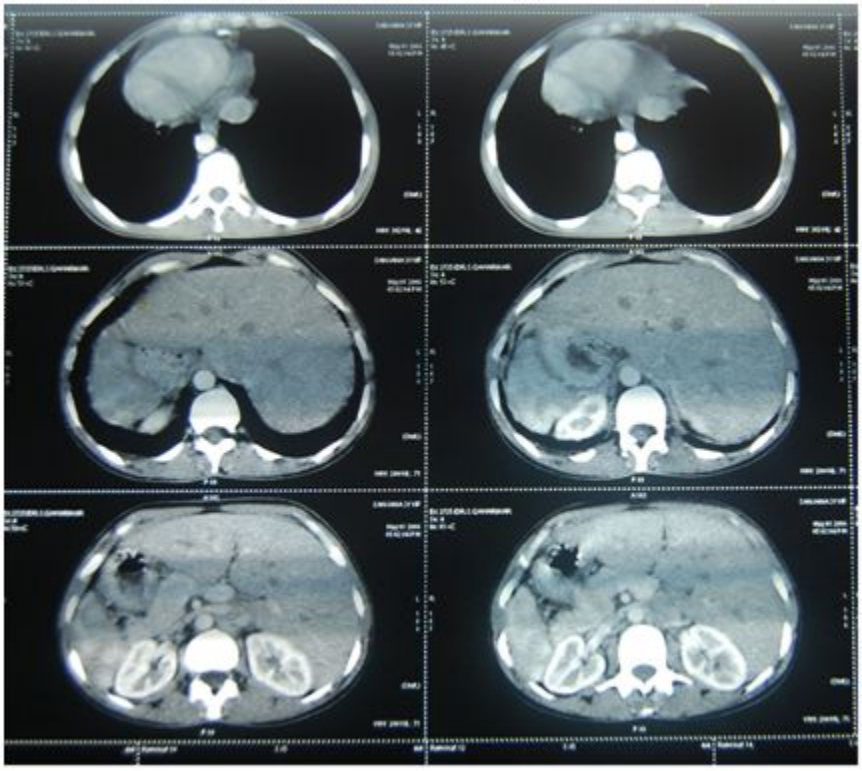
Patient Investigation: Blood count showed leucocytosis. Radiograph of chest showed dextrocardia with bronchiectasis. X-Rays of the paranasal sinuses showed right maxillary polyp with frontal sinus hypoplasia. Sputum Culture grew Pseudomonas. Abdominal ultrasound showed a spleen and stomach on the right and the liver was on the left. CT scan of the thorax confirmed the situs inversus and also showed severe bronchiectasis. ECG showed QS and T wave inversion in Lead 1 due to the dextrocardia. Pulse Oximetry recorded the O_2 saturation at 90%. However, arterial blood gas showed severe hypoxemia with a pH of 7.34, $PaCO_2$ of 36 mm of Hg, and a Pa O_2 of 60 mm of Hg. There was also severe pulmonary artery hypertension of 75 mm of Hg. The left ventricular ejection fraction was 50%. Pulmonary Hypertension had caused changes in the right side of the heart and ECHO cardiogram showed dilatation of the right ventricle and right atrium. Spirometry showed severe obstructive and restrictive defect. A saccharine pellet was laced under the inferior turbinate and it took more than one hour to produce a sweet taste showing marked delay due to defective ciliary functions.



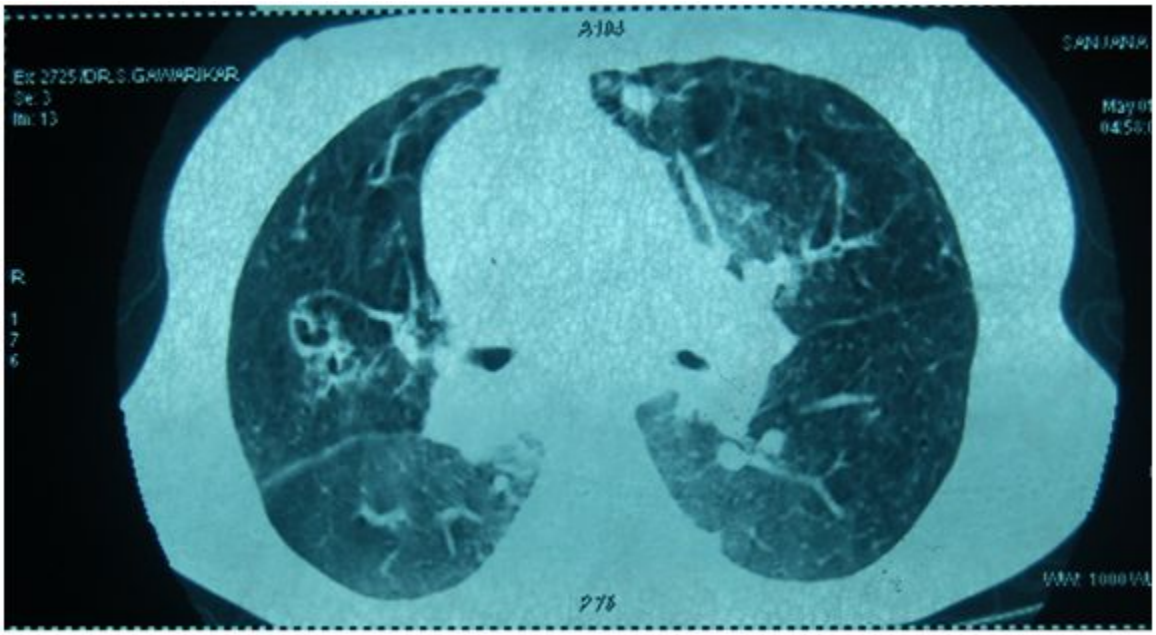
Water's View of the paranasal sinuses showing right maxillary polyp and frontal sinus hypoplasia



Chest X-Ray showing dextrocardia



CT Scan showing dextrocardia and situs inversus of the liver, spleen and stomach.



CT Scan thorax cut showing dextrocarida and bronchiectasis

Treatment: The patient was acutely dyspneic due to bronchospasm and had great difficulty in expelling

out secretions. She was managed by intranasal oxygen, antibiotics, bronchodilators, corticosteroids, mucolytics, diuretics and physiotherapy. She improved on treatment but she was unable to maintain saturation and was put on continuous 02 therapy. She was prescribed regular inhaled bronchodilators and chest physiotherapy to remove the thick viscid secretions aggravating the condition.

Discussion: Kartagener Syndrome is a unique syndrome described in 1933 by Manes Kartagener characterized by a triad of situs inversus, chronic sinusitis and Bronchiectasis.[1] It is an autosomal recessive disease with primary ciliary dyskinesia leading to abnormal ciliary motion and impaired mucociliary clearance. This leads to recurrent or persistent infections, sinusitis, otitis, Chronic bronchitis, recurrent pneumonia and bronchiectasis are commonly associated. Obstructive lung disease may be due to elevated levels of inflammatory mediators in chronically irritated airways. There is diminished female and male fertility.[2]

Approximately one half of patients with primary ciliary dyskinesia have situs inversus and are classified as having Kartagener Syndrome. This occurs in one in 32000 live births. Afzelius proposed that normal ciliary beating is necessary for visceral rotation during embryonal development.

Ciliated epithelium covers most of the upper and lower respiratory tract. Each ciliated cell has 200 cilia . Each ciliary axoneme consists of two central microtubule surrounded by nine microtubular doublets. Patients exhibit a wide range of ciliary ultrastucture and motility but the most common defect is reduction of a dynien arm.[3]

Evaluation of these patients reveal chronic mucoid rhinorrhoea. Nasal polyps are found in 30% of patients. Sinuses can be opacified with mucosal thickening. Ears have retracted tympanic membranes and middle ear effusions.[4] Many undergo repeated tympanostomy tube insertion, often complicated by chronic suppurative otitis media. Other associated otological disorders may include tympanosclerosis and cholesteatoma. Chest radiograph and CT Scan of the thorax reveals dextrocardia, bronchial wall thickening with bronchiectasis. Obstructive lung disease can also be present due to raised levels of local inflammatory mediators. There is diminished male and female infertility. Esophageal and congenital cardiac abnormalities may be present.

Mucociliary clearance can be tested by a saccharin test in which a small 1mm piece of saccharin is placed under the inferior turbinate and time taken to get the taste is normally 20 min. It is delayed by one hour or more in ciliary dyskinesia.

Other tests to study ciliary movement include direct video cinematography or oscillography and digital high speed video to study ciliary beat patterns. Nasal or tracheal biopsy can be done to study the structural abnormalities of cilia by electron microscope.

Uncontrolled bronchial asthma if coexistent can lead to airway inflammation and structural changes in the bronchial wall and add to the difficulty in expelling out thick viscid secretions. Bronchial hypersecretion is the consequence of hypertrophy and hyperplasia of submucosal glands and epithelial goblet cells. The replacement of ciliated cells by goblet cells contributes to airway remodeling.[6]

Effective management lies in the use of suitable antibiotics, mucolytics and chest physiotherapy. Patients should be explained the underlying problem and the importance of early and aggressive treatment of

infections to prevent bronchiectasis.[6] Newer therapies in management are beta adrenergic agonists, uridine-5-triphosphate, hypertensive saline and human recombinant DNAases.[7]

Summary: It is very important to recognize early the underlying defect of ineffective ciliary functions and the inability to efficiently remove secretions. Our patient had suffered from recurrent sinusitis and lower respiratory tract infections since childhood. She had a large polyp in right maxillary sinus. Because the patient was not prompt and did not receive early treatment for her infections, she developed severe impairment of her respiratory function. She was evaluated and placed on regular treatment of mucolytics, bronchodilators and physiotherapy and was explained about her problem and made to understand the importance of early initiation of antibiotic and a regular follow up.

References:

1. Kartagener M. Zur pathogene der Bronchiectasien: Bronchiectasien bei situs viscerum inversus. Beitr Klin Ertoversoh Tuberk Lungerke. 1933; 83: 489.

2. Eliasson R, Mossberg B, Camner P, Afzelius BA. The immotile-cilia syndrome. A congenital ciliary abnormality as an etiologic factor in chronic airway infections and male sterility. N Engl J Med. 1977 Jul 7;297(1):1-6. View Abstract

3. Leigh MW, Pittman JE, Carson JL, Ferkol TW, Dell SD, Davis SD, Knowles MR, Zariwala MA. Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome. Genet Med. 2009 Jul;11(7):473-87. View Abstract

4. Greenstone M, Stanley P, Cole P, Mackay I. Upper airway manifestations of primary ciliary dyskinesia. J Laryngol Otol. 1985 Oct;99(10):985-91. View Abstract

5. Mario Castro and Monica Kraft. Clinical Asthma. Pathophysiology of Asthma. Page-23-34.

6. Bush A, Cole P, Hariri M, Mackay I, Phillips G, O'Callaghan C, Wilson R, Warner JO. Primary ciliary dyskinesia: diagnosis and standards of care. Eur Respir J. 1998 Oct;12(4):982-8. View Abstract

7. Amirav I, Cohen-Cymberknoh M, Shoseyov D, Kerem E. Primary ciliary dyskinesia: prospects for new therapies, building on the experience in cystic fibrosis. Paediatr Respir Rev. 2009 Jun;10(2):58-62. Epub 2009 Apr 9. View Abstract