Kartagener's Syndrome : A Rare Case

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Abstracts: Primary ciliary dyskinesia (PCD) is a syndrome characterised by productive cough with bronchiectasis and sinusitis early in life and reduced fertility later in life. PCD is a rare syndrome with an estimated incidence of 1: 20,000 to 30,000 . Primary ciliary dyskinesia is a genetic disorder which manifests early in life and that distinguish it from the acquired mucociliary disorders . Here we discuss one such case of Kartagener's syndrome that presented to us with bronchiectasis , recurrent sinusitis with primary infertility and situs inversus , all of which which fit into the triad of Kartagener's syndrome [Bavaliya M et al NJIRM 2012; 3(4) : 139-141]

Key Words: Primary ciliary dyskinesia , Primary infertility , Acquired mucociliary disorders.

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Introduction: Kartagener Syndrome's (KS) is an autosomal recessive disorder characterized by dextrocardia , bronchiectasis and sinusitis[1]. The basic defect lies in the abnormal defective movement of the cilia. Males are generally infertile because of immotile sperms however; some males have completely normal spermatozoas. Cases of semi-sterility in females have also been reported[2].

Case Report:_History - A 35 years old male patient presented with complaints of productive cough and haemoptysis since 15 days and headaches since 1 month . He had similar complains in childhood with episodic fever and worsening of symptoms . He has been married since 10 years but does not have any children .

Treatment history - His records showed that he received several courses of antibiotics , antihistamines , bronchodilators , inhaled and oral corticosteroids , and even anti-tuberculous drugs but there was only partial response to these medications with temporary relief .

On examination – The patient had 100° F temperature , normal pulse and blood pressure with a respiratory rate of 24/min . Apex beat was localised to right fifth intercostals space in the mid-clavicular line and on percussion , the liver dullness was present on the left side – all of these suggestive of situs inversus .

On auscultating the respiratory system , there were bilateral , lower zone , coarse leathery crepitations with occasionally wheeze present in inspiration as well as expiration.

Investigations – His previous records show that he had undergone a lot of investigations like repeated chest x-rays and examination of the sputum for tuberculosis . His investigations revealed that the patient had haemoglobin of 12 gm/dl and total leucocyte count of 12,000/cmm of blood , and ESR of 60 mm in 1st hour . Routine biochemical tests did not reveal any significant abnormality . Sputum culture was negative for acid-fast bacilli , but showed Gram-positive cocci . Chest radiography confirmed dextrocardia with the aortic arch lying on right side of the trachea with cysticbronchiectatic changes seen in both lower zones and midzones (Figure 1)

HRCT Chest shows B/L bronchiectactic changes in lower and middle lobes(Figure 2).Xray PNS show absent left frontal sinus.CT abdomen shows situs inversus (Figure 3)

Semen analysis : sperm count – 14 million/ml . Rest parameters are normal Pulmonary function tests (PFT) revealed FEV1/FVC to be 62% of predicted values , suggestive of obstructive pattern with minimal improvement in PFT after bronchodilators . The 'nasal saccharin transit time ' test results gave the mucocilliary clearance time of 60 minutes (Normal < 30 minutes)[3] . However , due to the lack of facility for electron microscopic study of the nasal cilia, we

were not able to study the ultrastructural defect in it.

Management - Kartagener's syndrome was diagnosed clinically and confirmed by various investigations . He was treated with intravenous antibiotics and mucolytic agents and chest physiotherapy . Majority of the symptoms rapidly improved and he was discharged on low dose antibiotics in rotation.

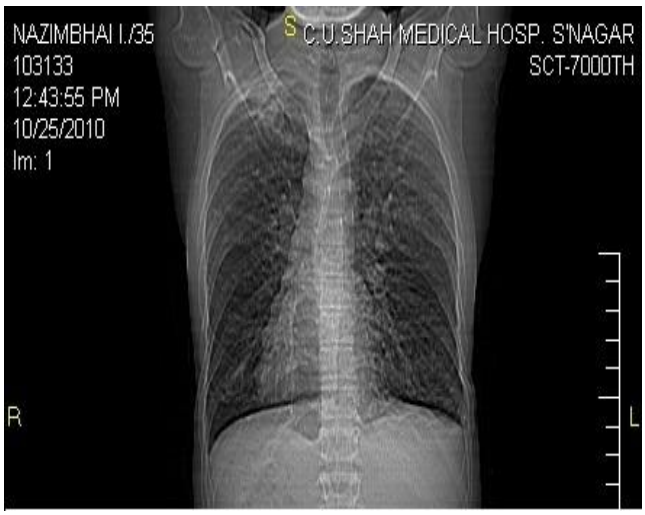

Figure 1 : chest x-ray showing dextrocardia and bronchiectactic changes

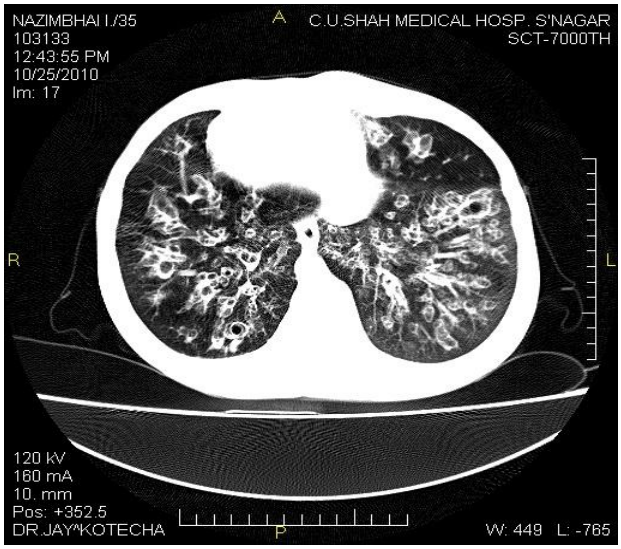

Figure 2: HRCT chest showing B/L bronchiectactic changes In lower and middle lobes

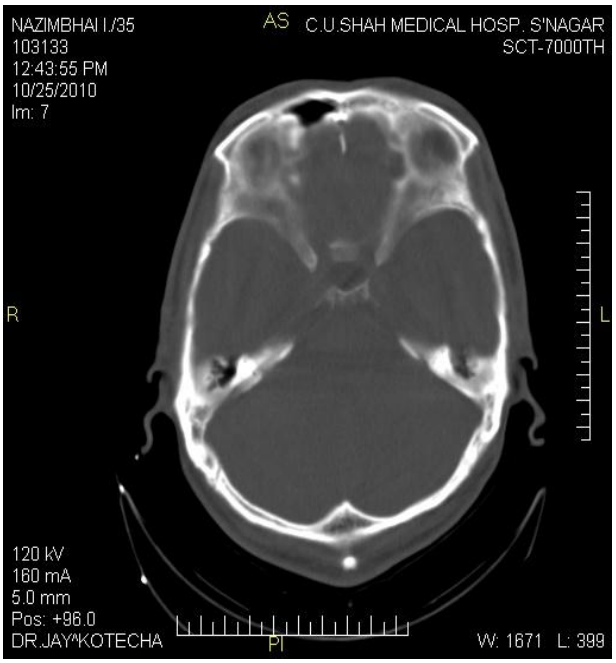

Figure 3 : x ray skull showing absent left frontal sinus

Discussion: The triad of bronchiectasis , sinusitis , and situs inversus was first described by Siewert in 1903, although its usual eponym is Kartagener's syndrome – after the Swiss paediatrician who described four cases with similar features in 1933[4] . By 1960 , 300 cases had been reported and the concept of a disease with congenital and generalised non-functioning Of the cilia evolved . About 50% of the people affected with primary ciliary dyskinesia have situs inversus , so they fit in the criteria for Kartagener's syndrome.

The clinical features of PCD have been described in primary ultrastructural defects in cilia . Ultrastructurally , cilia and spermatozoa tail are similar . The axoneme is the key component of the cytoskeleton and has a characteristic nine plus two arrays of microtubules .The nexin links and spokes seem to provide structural rigidity to the axoneme . Dynein arms extend from one side of a doublet in a clockwise direction , when viewed from the tip of the cilium . They contain most of the ATPase activity of the axoneme , and are important in

releasing energy for sliding and bending of microtubules and ciliary motion[5].

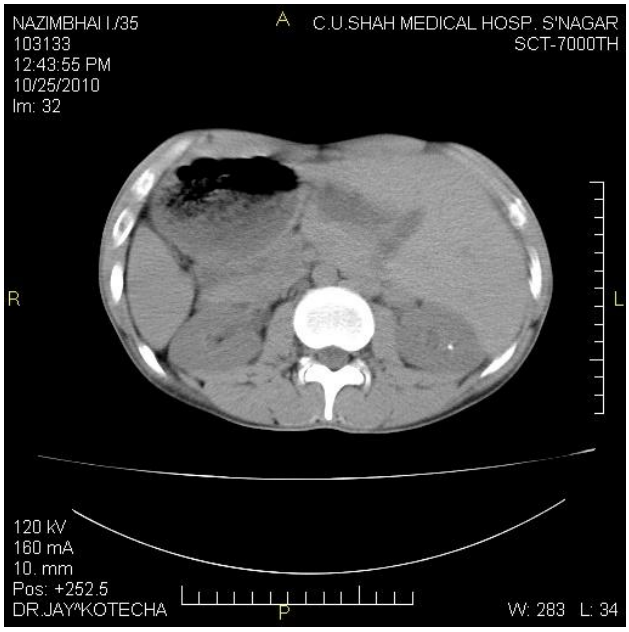

Figure 4 : CT abdomen showing situs inversus

The tracheobronchial tree is ciliated upto the level of the respiratory bronchioles , each ciliated cell having about 200 cilia . Mucociliary transport in the respiratory tract is important for normal respiratory function and resistance to respiratory infectioin as ciliary movements propels out the foreign materials e.g.microborganisms[6].

The typical clinical picture of PCD is a chronic productive cough which can usually be traced back to early childhood or infancy , chronic rhinitis often with nasal polyposis , chronic or recurrent maxillary sinusitis , and frequent ear infections in childhood . Bronchiectasis does not present at birth , but may develop early , sometimes as early as in childhood .

The most common respiratory pathogens are *Haemophilus Influenzae* and *Streptococcus pneumonia* . Most males are sterile , but many females have a lowered fertility . About 50% of patients have situs inversus viscerum[7] .

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