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Original Research Article

Unraveling the Puzzle: A Case Report on Kartagener Syndrome with Young's Syndrome

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Abstract:
Primary ciliary dyskinesia is an autosomal recessive disorder associated with abnormal ciliary structure and function, which results in retention of mucus and bacteria in respiratory tract, leading to chronic oto-sino-pulmonary disease, situs abnormalities and abnormal sperm motility. Kartagener syndrome is rare genetic disorder characterized with classical triad of situs inversus, chronic sinusitis and bronchiectasis whereas Young's syndrome is characterized by chronic rhinosinusitis, bronchiectasis and azoospermia. We present the case of 19 year old male patient with recurrent sinusitis and respiratory infection. His semen analysis revealed azoospermia without any evidence of obstruction in epididymis or vas deferens. He was diagnosed with both kartagener syndrome and young syndrome based on history, clinical presentation and radiological features. He was treated with oral antibiotics, mucolytics and chest physiotherapy. He was symptomatically better with the treatment and was put on long term low dose antibiotic. An early diagnosis of this syndrome is crucial to avoid future complications and improve the quality of life of patient.

Keywords: Azoospermia, Bronchiectasis, Dextrocardia, Kartagener Syndrome, Primary Ciliary Dyskinesia, Situs Inversus Totalis, Young Syndrome.

Introduction

Primary ciliary dyskinesia (PCD) is a genetic disorder that affects the structure and function of cilia, leading to impaired mucociliary clearance and various respiratory and other symptoms. Situs inversus occurs in about 50% of PCD cases. It is thought to affect 1 in 30000 live births. [1] Kartagener syndrome, a subtype of primary ciliary dyskinesia (PCD), typically involves a triad of symptoms: situs inversus (reversed internal organs), bronchiectasis (permanent dilation of the bronchi), and chronic sinusitis. Additionally, male infertility is common due to immotile spermatozoa, although their morphology is typically normal. [2] Young's syndrome, also known as primary ciliary dyskinesia (PCD) with bronchiectasis, sinusitis, and infertility, is indeed considered a variant of immotile cilia syndrome. Young's syndrome typically follows an autosomal recessive inheritance pattern, meaning that both copies of the responsible gene must be mutated for the disorder to manifest. The defective genes involved in Young's syndrome affect the structure or function of cilia in the respiratory tract and reproductive organs, leading to the characteristic clinical features. [3] Diagnosis of Young's syndrome requires ruling out other conditions that may present with similar symptoms, such as cystic fibrosis, which affects both genders equally, and immotile cilia syndrome (ICS), which shares some features with Young's syndrome. [4] Here we represent an unusual case of Kartagener Syndrome with Young syndrome presented with azoospermia which is a variation from the usual presentation of Kartagener Syndrome.

Case Detail: A 19 year old male patient second year college student, presented to Respiratory Medicine Department with chief complains of cough with

greenish expectoration since last 15 days, increased sputum production increased in morning with no any postural variation, left sided chest pain generalized to all region which was dull aching in nature since one month. He also had complain of fever since one week, continuous in nature, not associated with chills and rigors and relieved by medications. Generalized weakness was present since one week. He had no complain of blood in sputum, shortness of breath, decreased appetite, or burning micturition. He had a history of travelling outside the state one month back. He had a persistent complain of nasal blockage during childhood, which led to the diagnosis of nasal polyp, and operated for same before ten years. He had a history of multiple times lower respiratory tract infections since childhood. There was no history of pulmonary tuberculosis, trauma, injury, any addiction, any known allergy or any known co-morbidities including thyroid, diabetes mellitus and hypertension.

On general examination, he was afebrile with a heart rate of 110/min, oxygen saturation of 96% on room air, blood pressure of 112/66 mmHg, respiratory rate of 22 breaths per min. Clubbing was present on fingers of both hands which was of grade II. There was no pallor, icterus, cyanosis, pedal edema, cervical or axillary lymphadenopathy. On examination, auscultation of chest revealed bilateral air entry present with left sided crepitations present over inter-scapular region and infra scapular region.

All routine blood investigations were done which were as follow: Hb-12.4 gm/dl, WBC-9820/cumm, Platelets-3,34,200/cumm, peripheral blood smear showed normal morphology of all cells, S.creatinine-0.8mg/dl, Sgpt-16, S.FSH-6.8mIU/ml (ref value for adult male:2.1-18.6 mIU/ml), S.LH-3.6mIU/ml (ref value for adult male:1.7-11.2mIU/ml) and urine analysis was within normal limits. Sputum examination for culture and sensitivity showed growth for E.coli and Acinetobacter species.

The chest X-Ray postero-anterior view showed ill-defined heterogeneous density lesion involving left middle zone with cardiac shadow and aortic knuckle present over right hemithorax with gastric bubble seen over right side, suggestive of dextrocardia with situs inversus.

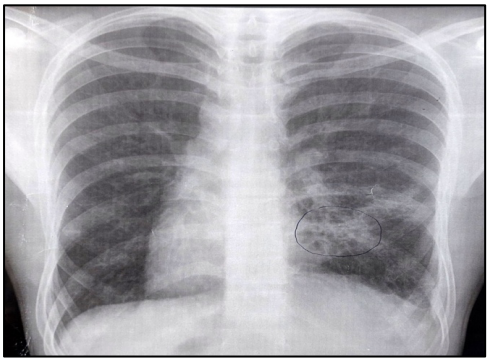

Figure 1: Chest X Ray PA view suggestive of heterogeneous density over left mid zone with cardiac apex and aortic knuckle over right side.

Electrocardiogram of both right and left sided precordial leads revels sinus tachycardia with normal axis and no ST-T waves abnormality.

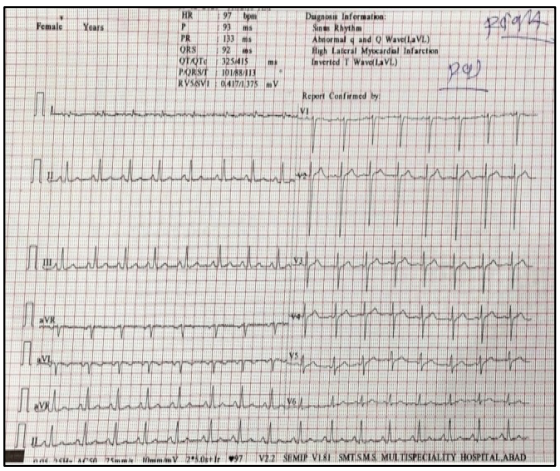

Figure 2 (a): Right sided ECG

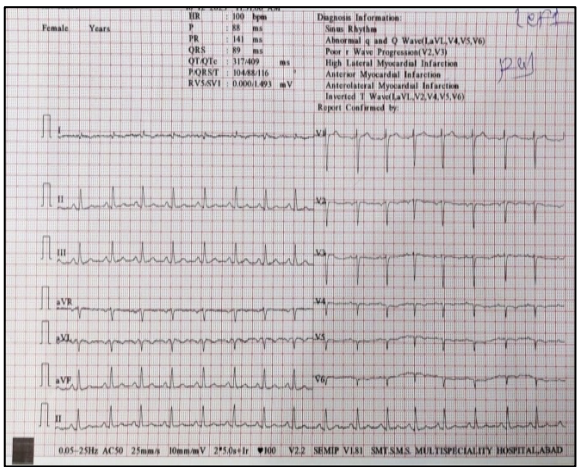

Figure 2 (b): Left sided ECG

Ultrasound sonography of abdomen pelvis showed few sub-centimeter to centimeter sized lymph nodes in LIF region, largest measuring 12×5 mm. Situs Inversus Totalis was noted with heart and spleen on the right, liver on the left and descending colon was seen on right side and terminal ileum, ileo-caecal junction, caecum and ascending colon were on left side.

X-Ray of para-nasal sinuses was suggestive of mild loss of pneumatization involving both maxillary sinuses.

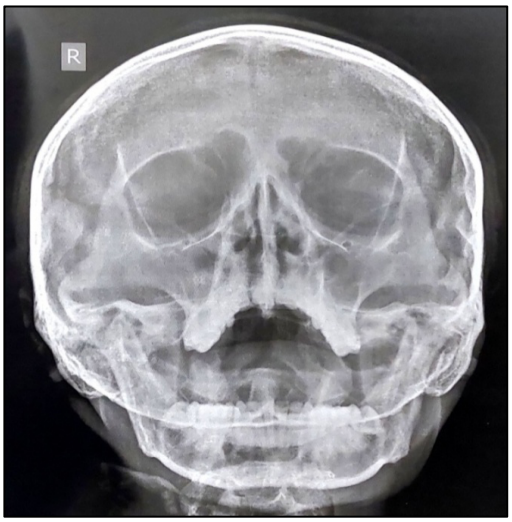

Figure 3: X-ray of para-nasal sinuses suggestive of mild loss of pneumatization of both maxillary sinuses.

High Resolution CT Thorax was suggestive of centrilobular nodular density with tree in bud opacities surrounding ground glass opacities in both upper lobes and lower lobes and right middle lobe. Small patchy areas of consolidations were noted in posterior basal segment of both lower lobes. Mild bronchial wall thickening was noted in bilateral lower lobes. These findings suggest possibility of infective bronchiolitis, Koch's etiology. Bronchiectasis was noted in right middle lobe, lingular segment and both lower lobes with distal mucous plugging with secondary infection. Cardiac apex was noted on right side, Visualized abdomen section showed viscero-vascular inversion-situs inversus totalis. These findings suggest possibility of kartagener syndrome.

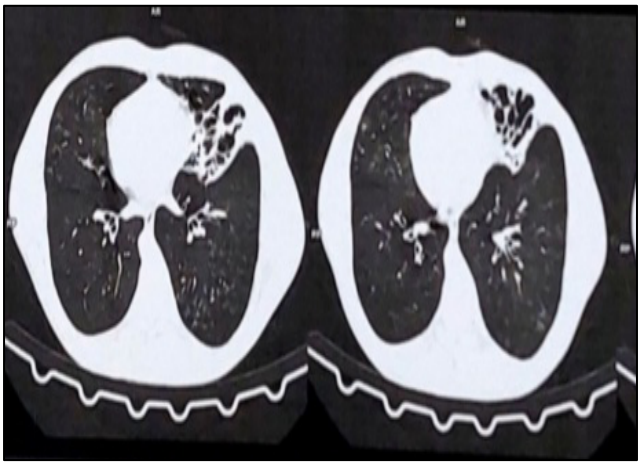

Figure 4: HRCT Thorax suggestive of bronchiectasis over lingular segment of left lung.

2 dimensional Echo-cardiography showed dextro-version, left ventricular ejection fraction 60% with rest normal findings.

Semen analysis done suggestive of whitish color, opaque in appearance, seminal odor with normal viscosity. The chemical examination revealed alkaline nature of semen with positive fructose test. A microscopic examination revealed no spermatozoa present in semen.

ENT reference done for nasal polyp, zero degree nasal endoscopy done suggestive of antero-inferior septal perforation, right sided nasal polyposis, sinusitis present with bi-lateral crusting present. Left sided examination were within normal limits.

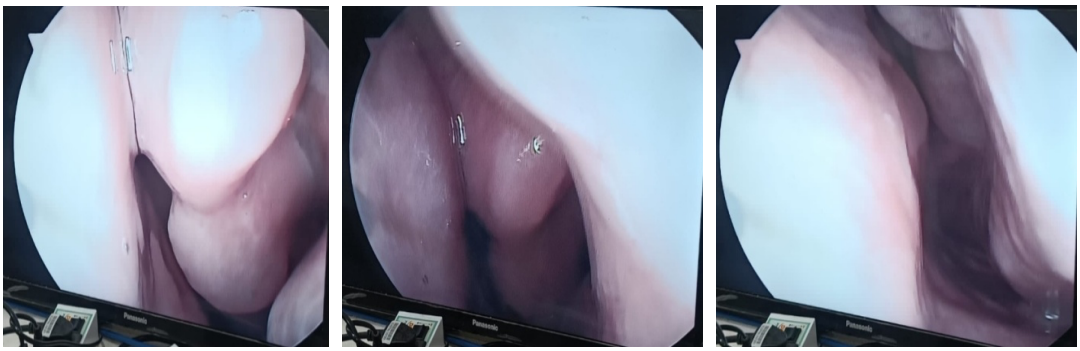

Figure 5(a) **Figure 5(b)** **Figure 5(c)**

Fig 5(a), 5(b) and 5(c) suggestive of right sided nasal polyposis with antero-inferior septal perforation.

Discussion

Kartagener syndrome (KS) and Young's syndrome, which is a rare genetic disorder falling under the broader category of Primary Ciliary Dyskinesia. KS is characterized by a clinical triad of chronic sinusitis, bronchiectasis, and situs inversus.[5] KS is caused by the absence of dynein arms, a congenital abnormality in the ultrastructure of cilia.[2] These structural abnormalities are typically caused by mutations in genes such as DNAI1 and DNAH5.[1] The consequences of impaired ciliary motility include chronic recurrent sinopulmonary infections and infertility.[5] Treatment is symptomatic and includes oral or intravenous antibiotics. Bronchiectasis and pneumonia should be treated with bronchodilators, mucolytics, oral corticosteroids, and chest physiotherapy. Influenza and Pneumococcal vaccines are necessary to prevent frequent infections.[1]

Young's syndrome is an autosomal recessive disorder characterized by immobility or low motility of cilia in the airways and sperms. It may be a variation of immotile cilia syndrome.[3] The structural abnormality causing decreased ciliary movement is typically identified by an electron microscope appearance that reveals a dynein or microtubule doublet deficiency. The similar deficiency is seen in cilia from respiratory epithelia and sperm tails; however the pulmonary presentation might not be very severe. Sperm in primary ciliary dyskinesia are hypomotile and exhibit reduced mucus clearance ultra structurally, making them more vulnerable to recurrent sinopulmonary infections.[3]

Table 1: Comparison of our case with other cases of Kartagener syndrome and Young's syndrome.

Sr no	Citation	Age	Gender	clubbing	Sinusitis	Nasal polyp	Bronchiectasis	Dextrocardia	Situs inversus	Semen analysis	Pulmonary function test	Kartagener's syndrome	Young's syndrome
1	Ibrahim et al.[1]	3 yr	Male	_	+	_	_	+	+	_	_	+	
2	Tangri et al.[2]	30 yr	Male (Married)	+	+	+	Bi-lat-eral	+	+	Azoo-spermia	Mild ob-structive abnor-mality	+	
3	Alduihi et al.[6]	31 yr	Male (Married)	+	+	_	Bi-lat-eral	+	_	Sperms +	Severe restric-tive ab-normal-ity	+	
4	Parihar et al.[3]	30 yr	Male (Un-married)	+	+	_	bi-lat-eral	_	_	Oligo-spermia with	Mixed pattern		+

										immo-tile sperms			
5	Lau KY et al.[7]	64 yr	Male (Married)	+	+	_	Bi-lat-eral	_	_	Azoo-spermia	Moder-ate ob-structive abnor-mality		+
6	Tadesse et al.[5]	24 yr	Male	_	+	+	bi-lat-eral	+	+	_	_	+	
7	Cihan-beylerden M et al.[4]	28 yr	Male (Married)	_	+	+	Bi-lat-eral	_	_	Azoo-spermia	Severe obstruc-tive ab-normal-ity		+
8	Sahu S et al.[8]	22 yr	Female (Unmar-ried)	+	+	_	bi-lat-eral	+	+	_	_	+	
9	Present Case	19 yr	Male (Unmar-ried)	+	+	+	Bi-lat-eral	+	+	Azoo-spermia	Moder-ate re-strictive abnor-mality	+	+

The diagnosis of Kartagener syndrome and young syndrome in our case was based on clinico-radiological features. The clinical presentation with chronic cough, rhino sinusitis, bronchiectasis and situs inversus along with infertility was sufficient enough to suspect Kartagener syndrome and young syndrome.

Conclusion:

Patient with kartagener and young's syndrome exists with recurrent respiratory tract infections, chronic rhinosinusitis. Genetic counselling and fertility issues should be explained to the patient. The early diagnosis and management is essential in overall prognosis, as late diagnosis may lead to reduced quality of life which can prevent long term complications.

Declaration of patient consent: The authors certify that they have taken patient's consent. The patient had also given consent for case information to be reported in the journal for educational and research purpose. Patient's name and identity will not be displayed.

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