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Kartagener Syndrome With Azoospermia : A Case Report

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Abstract

Kartagener Syndrome (KS), also known as Immotile Cilia Syndrome (ICS) is a rare, autosomal recessive genetic disorder that causes an impairment in the functioning of the cilia lining the respiratory tract (lower and upper, sinuses, Eustachian tube) and fallopian tube, and also of the flagella of sperm in males. In this report, we describe an unusual presentation of Kartagener syndrome with azoospermia in a 30-year-old male patient. The diagnosis was based on history of recurrent respiratory tract infection, bronchiectasis, maxillary sinusitis with polyp, dextrocardia with situs inversus totalis, etc. Semen analysis revealed azoospermia without any evidence of obstruction in epididymides or vas deference.

Keywords: Kartagener Syndrome, Bronchiectasis, Dextrocardia, Situs Inversus Totalis, Azoospermia

Introduction

Kartagener syndrome is a rare disorder in which there is a rotation of internal organs (situs inversus) in about 50% of cases with bronchiectasis, sinusitis and male infertility caused by immotile spermatozoa of normal morphology as revealed on semen analysis. All these abnormalities are due to an inherent defect in ciliary structure and function [1,2].

This report describes a rare case of Kartagener Syndrome with Azoospermia which is a variation from the usual presentation of Kartagener Syndrome.

Case Details

A 30-year-old male farmer came to our OPD for cough with purulent expectoration, occasional low-grade fever, nasal discharge, exertional breathlessness and headache for the last 15 days. He had history of similar illness off and on since early childhood. He gave history of bilateral hearing loss; more so on left side. He had undergone left ear surgery at the age of 2 months and surgery for nasal polyp at 24 years. He was a nonsmoker and had never consumed alcohol. He also denied history suggestive of gastrointestinal disease, cardiac disease, or genital infection.

There was no family history of recurrent sino-pulmonary infection. He was divorced from the last two years and had no issue despite history of normal puberty and libido.

On examination, the patient appeared pale with digital clubbing (grade II). The apex beat was in the right fifth intercostal space in midclavicular line with area of cardiac dullness on the right side. The liver dullness percussed on the left side, with tympanic stomach resonance detected on the right side. Respiratory system examination revealed tenderness over paranasal sinuses. He appeared eugonadal with normal-sized gonads and right testis placed at a lower level than the left. Ear examination showed bilateral chronic serous otitis media.

His investigations revealed hemoglobin of 9.1 g% with normal blood counts, blood biochemistry, urine analysis, etc. Serological test for HIV was nonreactive. Sputum smear microscopy was negative for acid-fast bacilli. Pulmonary function tests revealed mild obstructive type of ventilatory defect. Semen analysis revealed a low volume of semen (1 ml) with complete azoospermia on three occasions. Audiometry revealed bilateral conductive hearing loss. X-ray chest, PA view, revealed right-sided heart [Figure 1]. X-ray of paranasal sinus showed bilateral rhinosinusitis with maxillary polyp [Figure 2]. On CT scan thorax, there was situs inversus with complete inversion of bronchial, atrial and visceral situs with dextrocardia. This was accompanied by complete collapse of left middle lobe with cylindrical and bilateral bronchiectasis; and infectious Bronchiolitis. These findings were consistent with Kartagener syndrome [Figure 3&4].

Figure 1: X-ray chest (PA view) showing right-sided heart

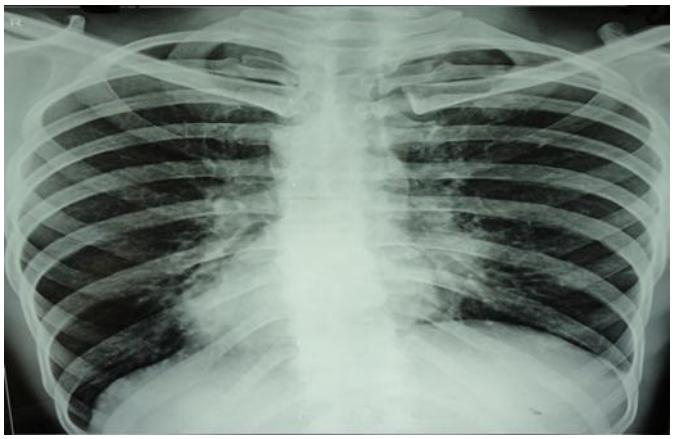

Figure 2: X-ray of paranasal sinus showing bilateral rhinosinusitis with maxillary polyp

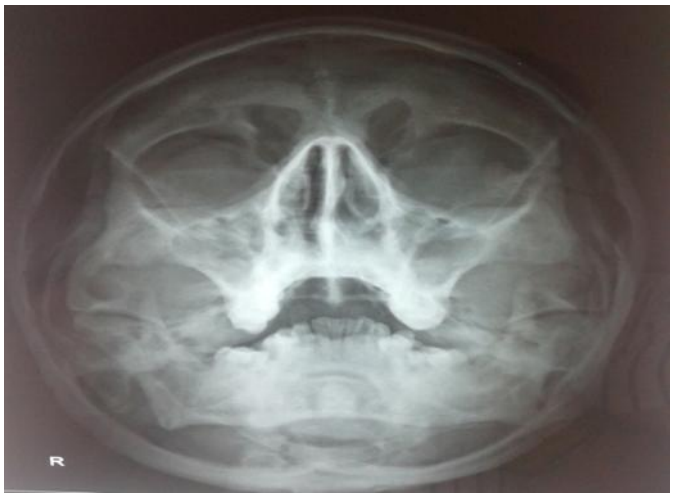

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Figure 3: CT scan thorax showing situs inversus totalis with complete inversion of visceral situs

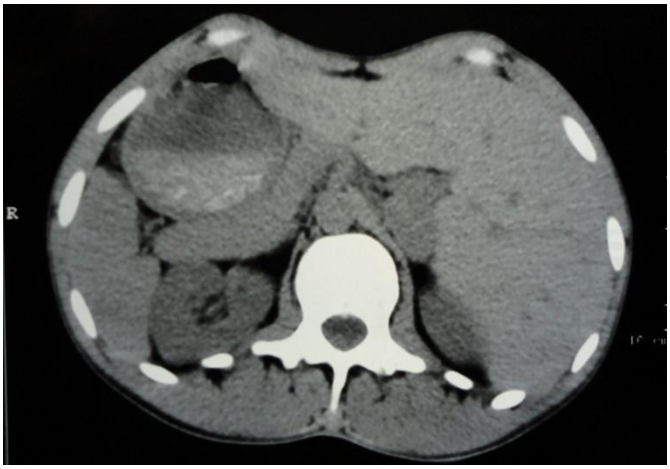
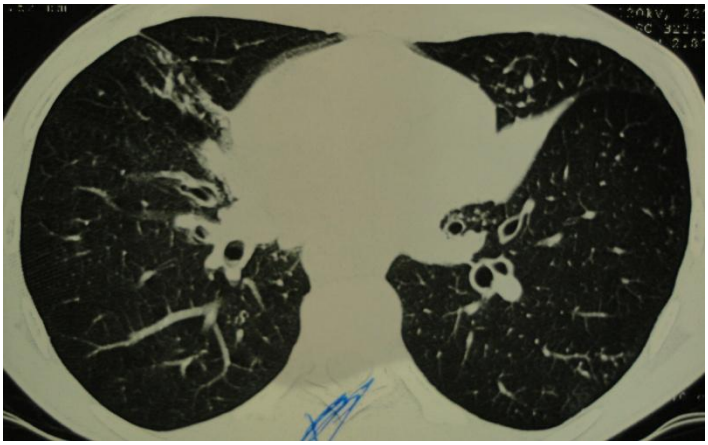


Figure 4 : CT scan thorax showing cylindrical, bilateral bronchiectasis



Discussion And Conclusion

The triad of situs inversus, bronchiectasis, and sinusitis has carried Kartagener's name since he reported four cases in 1933 [3]. Although Siewart first described this condition in 1904, the credit of recognizing the etiological correlation between the elements of the triad goes to Kartagener, whose name has been associated with this condition [4]. Up to 1955, only 104 cases of this syndrome were reported in the literature; subsequently, more cases of this condition were recognized, including a few from our country [5, 6].

The issue of fertility was not addressed in the initial published reports of patients with Kartagener syndrome, until Arge reported three male patients with this syndrome having immotile spermatozoa and sterility [7]. In 1976, Afzelius noted a lack of dynein arm in the sperm and cilia of four subjects; out of them, three had Kartagener syndrome [8]. In 1977, Eliasson and associates proposed that, what they called the immotile-cilia syndrome (ICS), was responsible for chronic airway infection and male sterility in a number of persons [2]. Following these original reports, a new syndrome, called ICS, was recognized and applied to many patients with or without Kartagener syndrome [9].

The cause of this condition was found to be a congenital defect in the ultra-structure of the cilia in the form of lack of dynein arms. In addition, the definition was broadened to include patients with other ciliary abnormalities, including transposition of ciliary microtubules, defective radial spokes, and absence of inner but preservation of outer dynein arms [10]. Other electron microscopic abnormalities that are reported include missing nexin link, compound cilia, supernumerary, absent or incomplete

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microtubules, lack of ciliary orientation, and abnormal attachment of the dynein arms [11].

In the recent years, the term Kartagener syndrome has been replaced by the term primary ciliary dyskinesia. This replacement reflects the findings that the cilia of many patients do move, albeit in an uncoordinated fashion [12]. Whatever may be the defect, whether immotility or inappropriate motility (insufficient beating pattern), because cilia occurs mainly in the respiratory tract and genital tract, the clinical symptoms of Kartagener syndrome are mostly chronic sinusitis, bronchitis, bronchiectasis, and male sterility. The situs inversus, which occurs in approximately 50% of patients with this disorder, is possibly caused by an inability of embryonic cilia to shift the heart to the left side; however, the exact genetic link between ciliary dyskinesia and situs inversus is still not clear [10]. It is also suggested that the absence of dynein in the axoneme is probably part of a diffuse genetic defect, which may extend to cytoplasmic, nonaxonemal dynein and leads to a disturbance of various microtubule-dependent cell activities. This also seems true as there are reports of ICS associated with hepatic parenchymal steatosis, retinitis pigmentosa due to abnormal cilia of retinal pigment epithelium, polysplenia, extra-hepatic biliary atresia etc., suggesting a single dysmorphogenetic process [13, 14, 15].

The suggested criteria used for diagnosis of Kartagener syndrome include clinical signs of chronic bronchitis and rhinitis from early infancy, combined with one or more of the following additional symptoms: 1) situs inversus in the patient or in sibling, with similar clinical signs; 2) living but immotile spermatozoa of normal appearance on semen analysis; 3) transbronchial clearance absent or markedly depressed and 4) cilia with characteristic ultra-structural defect [16]. Our case fulfills the diagnostic criteria for Kartagener syndrome with variation in view of ‘complete azoospermia’. This situation is extremely rare and has been reported rarely in literature. Bashi et al., reported a similar case in 1988 from Riyadh, Saudi Arabia [17]. Gill et al., reported a case of Kartagener syndrome along with left empyema thoracis and azoospermia in 1996 from Varanasi, India [18]. In the latter, the authors attributed azoospermia in their case to asthenozoospermia, oligozoospermia, or oligoasthenozoospermia. The other contributing factors could be lack of contraction of myoids in the walls of tubules and muscular elements in tunica albugenia.

The diagnosis of Kartagener syndrome in our case was based on clinico-radiological features. The clinical presentation with chronic cough, bronchiectasis, maxillary sinusitis, and situs inversus along with infertility was sufficient enough to suspect Kartagener syndrome. Further, it is also observed that none of the ciliary defects observed in Kartagener syndrome are specific. They may all occur as secondary lesions or sporadically as varieties in otherwise healthy subjects [11]. In conclusion, Kartagener syndrome should be suspected on clinical grounds in individuals presenting with chronic cough, rhinorrhea, sinusitis, otitis media and obstructive lung disease along with male sterility and with or without situs inversus.

Patient Consent

The authors, Dr. Nitin Tangri and Dr. Priyanka Tangri declare that:

They have obtained written, informed consent for the publication of the details relating to the patient(s) in this report.

All possible steps have been taken to safeguard the identity of the patient(s).

This submission is compliant with the requirements of local research ethics committees.

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