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Case Report

Situs Inversus Totalis: The Association of Kartagener's Syndrome With Diffuse Bronchiolitis and Azoospermia☆

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ABSTRACT

Kartagener's syndrome (KS) is characterized by bronchiectasis, paranasal sinusitis and situs inversus totalis (SIT). Association of diffuse bronchiolitis (DB) with KS has been documented from Japan only. Fourteen patients with SIT were seen in one unit over 7 years. All patients underwent a similar work up which included high resolution computed-tomography (HRCT) of thorax and CT-paranasal sinuses. Semen analysis was done in 2/3 adult males. Eleven patients (6 males and 5 females) had KS while 3 adult females had SIT alone. HRCT-thorax detected bronchiectasis in 10/11 patients with KS. HRCT-thorax confirmed DB in 6/11 patients with KS. One adult male had total sperm count of 2.5 million/ml without sperm motility while the other had no sperms.

This series documents the largest number of patients with SIT/KS from India and highlights for the first time, the association of DB with KS from India.

Situs inversus total: asociación de síndrome de Kartagener con bronquiolitis difusa y azoospermia

Palabras clave:
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Discinesia ciliar primaria
Situs inversus total

RESUMEN

El síndrome de Kartagener (SK) se caracteriza por bronquiectasias, sinusitis paranasal y situs inversus total (SIT). La asociación de la bronquiolitis difusa (BD) con el SK se ha documentado únicamente en Japón. En una unidad se identificaron 14 pacientes con SIT durante un periodo de 7 años. Todos los pacientes fueron objeto de un estudio diagnóstico similar que incluyó una tomografía computarizada (TC) de alta resolución (TCAR) del tórax y TC de senos paranasales. Se realizó un análisis de semen en 2/3 de los varones adultos. Once pacientes (6 varones y 5 mujeres) tenían un SK, mientras que en 3 mujeres adultas había tan solo un SIT. La TCAR de tórax detectó bronquiectasias en 10/11 pacientes con SK. La TCAR de tórax confirmó la BD en 6/11 pacientes con SK. Un varón adulto presentó un recuento espermático total de 2,5 millones/ml, sin motilidad espermática, mientras que en los otros no hubo espermatozoides.

Esta serie documenta el mayor número de pacientes con SIT/SK descrito en la India y resalta por primera vez la asociación de BD con SK en este país.

Introduction

Situs inversus total (SIT) entails a mirror-image reversal of all the asymmetrical structures of the body and it is estimated to occur in 1/8000–1/25 000 newborns.[1] Most patients with SIT lead a completely normal life but, in approximately 20%–25% of patients, associated primary ciliary dyskinesia (PCD) may also be observed.[1] PCD, also known as "immotile ciliary syndrome", is a genetic disease caused by defects of the structure and function of the cilia that lead to abnormal mucociliary clearance which leads to a clinical disease of the sinus and pulmonary regions. Kartagener's syndrome[2] (KS), which is currently classified as a subgroup of PCD, is characterized by the triad of bronchiectasis, paranasal sinusitis and SIT.

We present a review of 14 patients with SIT, 11 of whom had KS. In 6 of these patients, there were signs of diffuse bronchiolitis (DB) on thoracic CT. This manifestation associated with KS was first reported in Japan,[3] and it still has not been widely accepted. The

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presence of oligo/azoospermia was observed in 2 of our patients with KS. As far as we know, this is the largest patient series of SIT cases in India.

Case Descriptions

Over the course of a 7-year period, between April 2003 and March 2010, approximately 11 361 new patients (1944 of them from the pediatric age group) with respiratory symptoms were remitted to a unit at our institute. 14 of them (6 males and 8 females; 6 adults and 8 pediatric patients) were diagnosed with SIT. Out of these 14 patients, 11 presented SIT with KS, while 3 only presented SIT (all adult females). The diagnosis of KS was based on the demonstration of the classic triad of the syndrome, consisting of bronchiectasis, paranasal sinusitis and SIT. All patients underwent a similar diagnostic study that included sputum culture for aerobic microorganisms, including *Mycobacterium tuberculosis*, electrocardiogram, complete spirometry, abdominal ultrasound, computed tomography (CT) with contrast as well as thoracic high resolution CT (HRCT) and CT of paranasal sinuses (PNS). By means of CT, the severity of the bronchiectasis[4] and sinusitis[5] was evaluated with previously described methods. A semen analysis was done in 2/3 of the adult males who gave their consent.

The clinical characteristics of these 14 patients are indicated, by order of presentation, in Table 1.

Radiological Exploration

Chest HRCT was able to detect bronchiectasis in 10/11 patients with KS. It was observed in a minimum of 2 lobes and in a maximum of 5, but in the majority of the patients there were 4 affected lobes. The left lower lobe was the most affected, with signs in 8/11 patients. This was followed in frequency by the right lower and middle lobes and lingula (7/11 patients each). Bronchiectasis was observed in the upper lobes in 3/11 patients. The overall severity score of the bronchiectasis[4] ranged between 0 and 13 out of a total maximum score of 18, with a mean of 5.81±2.75 (Tables 1 and 2). In 6 patients with KS, small diffuse centrilobular nodules were observed (2 mm in diameter) with an appearance of "tree in bud" pattern predominantly in the middle lobes, lingula and lower lobes that would suggest diffuse bronchiolitis. In 3/6, we also observed air trapping, tram-tracking and bronchiolectasis. In 4/6, there were grouped thick-walled cystic spaces (bronchi), mainly in the middle lobes and lingula that were suggestive of atelectasis or consolidation (Table 2). In the 3 patients without KS, chest HRCT showed normal lung parenchyma and the CT of the PNS was also within the limits of normality. Table 2 details the radiological manifestations of the patients with KS.

Table 2
Radiological Tests.

Structural characteristics on thoracic HRCT	Patients with SK (n=11)
Bronchiectasis (any lobe)	10
Overall severity score of bronchiectasis[18]	5.81±2.75
Affected lobe	
Upper left lobe	3 (patients 1, 3, 4)
Middle left lobe	7 (patients 2, 5, 6, 8, 10, 12, 13)
Lower left lobe	8 (patients 2, 4, 5, 6, 8, 10, 12, 13)
Right upper lobe	3 (patients 4, 5, 8)
Right lingula	7 (patients 3, 5, 6, 8, 10, 12, 13)
Lower right lobe	7 (patients 1, 4, 5, 6, 8, 12, 13)
Number of lobes affected	
None	1 (patient 7)
One	0
Two	3 (patients 1, 2, 3)
Three	1 (patient 10)
Four	4 (patients 4, 6, 12, 13)
Five	1 (patients 5, 8)
Six	0
Thoracic HRCT: other signs	
Small centrilobular nodules with Tree-in-bud pattern	6 (patients 2, 5, 6, 7, 8, 10)
Hyperinflation (air trapping)	3 (patients 5, 8, 10)
Consolidation/Colapse of the middle lobe and/or lingula	4 (patients 2, 6, 7, 10)
Pectus carinatum	1 (patient 6)
TC de PNS structural characteristics	Patients con SK (n=7/11)
Sinusitis (any)	7
Overall severity score[19]	15.37±1.99
Hypoplasia/agenesis of the frontal sinus	6 (patients 1, 6, 7, 8, 12, 13)

CT, computed tomography; HRCT, high resolution computed tomography; PNS, paranasal sinuses.

Semen Analysis

Two of the 3 adult males consented to a semen analysis. The 2 patients were married but did not have any children. In one patient, the sperm count was 2.5 million/ml with no sperm motility, and in the other no spermatozoids were observed (Table 1). In this latter case, the level of fructose in semen was higher than 100 mg/dl.

Table 1
Demographic Characteristics and Clinical Profile.

Patient Number	Age (Years)/Sex	Married/Infertility/Semen Analysis (Total Count [millions/ml] and Motility [%])	Respiratory Symptoms[a]	Nasal Symptoms[b]	Quantity of Sputum, ml/day	Sputum Culture for Microorganisms Aerobios
1	6/M	N	1,2	1,2	None	Lack of sputum
2	17/F	N	1,2,3,4	1,2	20	*Haemophilus influenzae*
3	16/F	N	1,2,6	1,2,5,6	100	*Streptococcus pneumoniae*
4	22/M	N/2,5y0	1,2	1,5,6	100	NOP
5	28/M	Y/Y/0y0	1,2,6	1,2	100	NOP
6	16/M	N	1,2	1,2	100	NOP
7	10/M	N	1,2	1,2,6	10	*Pseudomonas aeruginosa*
8	50/M	Y/Y/Unrealized	1,2,3,6	1,5,6	100	*Pseudomonas aeruginosa*
9	54/F	Y/N	1,2,3,4	1,2,4	10	NOP
10	13/F	N	1,2	1,2,6	10	NOP
11	77/F	Y/N	1,2	N	5	Lack of sputum
12	15/F	N	1,2,3	1,2	100	NOP
13	17/F	N	1,2	1,2	100	NOP
14	35/F	Y/N	1,2,3,4	N	5	NOP

M, males; F, females, N, no; Y, yes; NOP, no pathologic organism.

[a] Respiratory symptoms: 1, cough; 2, sputum; 3, dyspnea; 4, wheezing; 5, thoracic pain; 6, hemoptysis.

[b] Nasal symptoms: 1, rinorrea; 2, nasal obstruction; 3, nasal prurito; 4, sneezing; 5, postnasal drip; 6, repetitive clearing of the throat.