

# KARTAGENER SYNDROME

Khalid Shakeel Babar<sup>1</sup>, Habibullah Khan<sup>2</sup>, Yasser Ismail<sup>2</sup>, Qaisar Azim<sup>2</sup>, Muhammad Fawad<sup>2</sup>  
Department of <sup>1</sup>Radiology and <sup>2</sup>Medicine, Gomal Medical College, D.I.Khan, Pakistan

## ABSTRACT

Kartagener syndrome is an autosomal recessive disorder, a subgroup of primary ciliary dyskinesia, characterized by a triad of bronchiectasis, sinusitis and situs inversus. In cases of recurrent lower respiratory tract infections Kartagener syndrome should be kept in mind to prevent unnecessary repeated admissions, investigations and inappropriate treatment. Social, psychological and fertility issues should be addressed once it is diagnosed to help these patients to live with Kartagener syndrome safely. Here we present a case of 37 year old female who presented with bronchiectasis, recurrent episodes of sinusitis and situs inversus which conforms to the triad of Kartagener syndrome.

**KEY WORDS:** Kartagener syndrome; Dextrocardia; Situs inversus; Bronchiectasis; Sinusitis.

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## INTRODUCTION

Kartagener syndrome is an autosomal recessive disorder, a subgroup of primary ciliary dyskinesia, characterized by a triad of bronchiectasis, sinusitis and situs inversus. Approximately one half of patients with primary ciliary dyskinesia are having Kartagener syndrome.<sup>1</sup> It has an estimated incidence of 1 in 20,000 to 30,000.<sup>2</sup>

Siewert first explained the combination of situs inversus, chronic sinusitis, and bronchiectasis in 1904. (Although,) Manes Kartagener first recognized this clinical triad as a distinct congenital syndrome in 1933 and described this syndrome in detail, so it bears his name.<sup>1,3</sup> We present a case of 37 years old female, who presented with bronchiectasis, recurrent episodes of sinusitis and had situs inversus. The correct diagnosis of this rare congenital autosomal recessive disorder is important for its proper management as many complications can be prevented if timely management is instituted.

## CASE HISTORY

A 37 year old female presented to us with chief complaints of shortness of breath and productive cough for one and a half months. Patient had episodes of shortness of breath, sinusitis and allergic rhinitis in the past. On further inquiry, she had been married for 15 years and gave birth to one male child with no history of using contraceptive measures. On chest examination, there were bilateral rhonchi in middle and lower lobes and heart sounds were

absent on left side and heard over the right side, rest of the examination was unremarkable. On routine investigation, chest x-ray (PA view) showed dextrocardia, increased broncho-pulmonary markings in left lower lobe and gastric bubble on left side. (Fig.1)



Figure 1: Chest x-ray (PA view) of patient with Kartagener syndrome, showing dextrocardia, increased bronchopulmonary markings in the left lower lobe and gastric bubble on left side.

## Corresponding Author:

Dr. Khalid Shakeel Babar  
Babar House, Darabun Chungi D.I.Khan, Pakistan  
e-mail: khalidshakeelbabar@gmail.com

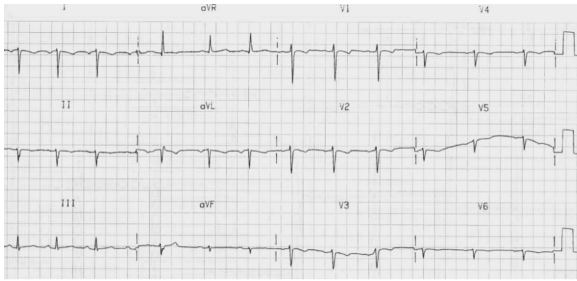


Figure 2: ECG of patient with Kartagener syndrome, showing signs of dextrocardia.

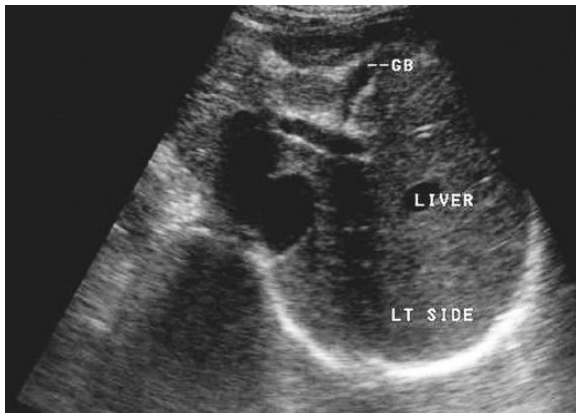


Figure 3: Ultrasound abdomen of patient with Kartagener syndrome showing situs inversus.

ECG showed right axis deviation, positive QRS complexes (with upright P and T waves) in aVR, inversion of all complexes in lead I, and absent R-wave progression in the chest leads (dominant S waves throughout). (Fig. 2)

Ultrasound abdomen confirmed situs inversus i.e. abdominal viscera on the opposite side. (Fig. 3)

The provisional diagnosis of Kartagener syndrome was made on the basis of clinical picture and investigations.

## DISCUSSION

In Kartagener syndrome sinusitis, bronchiectasis, and male infertility are attributed to abnormal ciliary motility. Cilia may be immotile or may show uncoordinated and inefficient movement patterns. Camner & coworkers first suggested ciliary dyskinesia as the cause of Kartagener syndrome in 1975. They described two patients with Kartagener syndrome who had immotile cilia and immotile spermatozoa. These patients had poor mucociliary clearance because the cilia that lined their upper airways were not functioning.<sup>1,4</sup> Most males are sterile, but many females have a lowered fertility.<sup>2</sup>

Kartagener syndrome is part of the larger group of disorders referred to as primary ciliary dyskinesias. Approximately one half of patients with primary ciliary dyskinesia have situs inversus and, thus, are

classified as having Kartagener syndrome. Afzelius proposed that normal ciliary beating is necessary for visceral rotation during embryonic development. In patients with primary ciliary dyskinesia, organ rotation occurs as a random event; therefore, half the patients have situs inversus and the other half have normal situs.<sup>5</sup>

It is associated with mutations in genes, mainly affecting DNAI1 on 17 chromosome 9p21-p13, DNAI2, DNAH5 14 chromosome 5p15p14, DNAH11 on 18 chromosome 7p21, thioredoxin domain containing 3 (*TXNDC3*), radial spoke head 9 homologue (*RSPH9*) and 4 homologue A (*RSPH4A*), which results in numerous defects including structural abnormalities of the dynein arms, radial spokes, and microtubules of the cilia.<sup>2,6,7</sup>

In some patients with the clinical phenotype of primary ciliary dyskinesia (PCD), the ultrastructure of the cilia appears normal, but the cilia are not normally orientated with respect to one another. This disorientation defect can occur secondary to infection or as a primary defect.<sup>8</sup>

Patients with Kartagener syndrome may have either situs solitus i.e dextrocardia only or situs inversus totalis where all the viscera are on the opposite side, including left sided appendix.<sup>9</sup> The patient in this case was having situs inversus totalis.

Tests measuring nasal nitric oxide and mucociliary clearance are useful for screening, but require confirmation with tests of ciliary function and ultrastructure.<sup>10</sup>

Demonstration of abnormal ciliary movement needs electron microscopic studies of biopsies obtained from the nasal mucosa or trachea. However these procedures are invasive and available only at specialized centers, therefore the diagnosis of Kartagener syndrome in this case was clinical, supported by imaging studies.

The condition should be kept in mind in a patient presenting with; recurrent sinusitis and bronchiectasis, asthma like symptoms and signs responding poorly to conventional treatment, recurrent lower respiratory tract infections causing fever, sweating and weight loss, tempting the physician to give a trial of antituberculous drugs.<sup>2,9</sup> Ectopic pregnancy and subfertility in women, male infertility (immotile sperm, but 50% of PCD males are fertile).<sup>11</sup>

Differential diagnosis is from cystic fibrosis (an inherited disorder), allergic bronchopulmonary aspergillosis, a fungal infection and alpha-1 antitrypsin deficiency, an inherited disease seen in white population.<sup>2</sup>

Appropriate medical therapy has been shown to prevent deterioration in lung function. They

should be looked after only in a setting where multi-disciplinary review is available. Management is not evidence based. Respiratory management consists of; regular respiratory monitoring, airway clearance by combinations of physiotherapy and physical exercise, and aggressive treatment of upper and lower airways infections.<sup>11</sup>

Early diagnosis and treatment is important to prevent long term complications and problems associated with it.<sup>12</sup> Genetic, social, psychological and fertility issues should be addressed once it is diagnosed and help these patients to live with Kartagener syndrome safely.

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CONFLICT OF INTEREST  
Authors declare no conflict of interest.  
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