

KARTAGENER'S SYNDROME: A CASE REPORT

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ABSTRACT

Kartagener's syndrome is a subset of primary ciliary dyskinesia, an autosomal recessive inherited disease, and is characterized by the triad of chronic sinusitis, bronchiectasis, and *situs inversus*. This paper reports the case of a 27-year-old female presenting with dyspnea on medium exertion, accompanied by chronic cough, non-productive or with clear expectoration. She had recurrent pneumonia until 15 years of age and underwent a lobectomy in the lower lobe of the left lung, probably due to bronchiectasis. Chest computed tomography showed *situs inversus totalis*, signs of previous surgical manipulation, and mild bronchial thickening. Computed tomography of the paranasal sinuses showed signs of chronic sinusitis due to a probable ciliary kinesia disorder. These findings suggest the diagnosis of Kartagener's syndrome. The prognosis reveals a slow rate of decline in lung function. However, repeated or chronic infections can negatively influence the quality of life of these patients.

Keywords: *Kartagener's syndrome; dextrocardia; bronchiectasis; sinusitis; ciliary motility disorders*

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INTRODUCTION

Kartagener's syndrome was first recognized in 1933 and is characterized by the triad of *situs inversus*, bronchiectasis, and chronic sinusitis, being the most frequent type of primary ciliary dyskinesia (PCD)¹, which is an autosomal recessive disorder characterized by immotile, dysmotile, or absent cilia². There is a considerable variation in clinical presentation, but the most common features are recurrent upper and lower respiratory diseases. *Situs inversus*, when present in patients with PCD, usually occurs as a complete inversion of the circulatory system and viscera; however, it does not add serious adverse health consequences³. The diagnosis of chronic sinusitis and bronchiectasis is suspected by clinical symptoms and can be confirmed through imaging tests such as plain radiography and computed tomography (CT)^{4,5}.

There is no gold standard diagnostic test established. A history of neonatal respiratory distress, persistent cough, chronic nasal congestion, rhinorrhea, otitis media, and signs of laterality indicating *situs inversus* should raise a strong clinical suspicion about the condition³. The prognosis reveals a slow rate of decline in lung function. The patients with the syndrome commonly lead an active life and have a normal life expectancy. However, repeated or chronic infections can negatively influence the quality of life of these patients⁶.

Normal ciliary function is required to determine the laterality of the heart and abdominal contents during fetal development; in the absence of ciliary motility, the laterality occurs randomly. Poor function of the nodal cilia during embryonic development explains why dextrocardia and *situs inversus* occur in approximately half of the patients in whom PCD is identified⁷.

The following case aims to highlight findings present in Kartagener's syndrome, both because of the difficulty in establishing its diagnosis and because of its rarity.

CASE PRESENTATION

A 27-year-old female presented with dyspnea on medium exertion, accompanied by chronic cough, non-productive or with clear expectoration. Pulmonary auscultation showed bilateral lung apex wheezing.

She had a long history of airway diseases, with recurrent pneumonia until 15 years of age. In 2012, she performed a lobectomy in the lower lobe of the left lung, but was unable to state the reason for the lobectomy; we infer to be areas of bronchiectasis. The patient was also asthmatic.

In the investigation, spirometry suggested mild obstructive ventilatory disorder with response to bronchodilators, confirming the diagnosis of asthma. Chest CT showed *situs inversus totalis* (Figure 1), signs of previous surgical manipulation, and mild bronchial thickening (Figure 2). CT of the paranasal sinuses showed signs of chronic sinusitis due to a probable ciliary kinesia disorder (Figure 2).

Clinical and tomographic findings, consisting of chronic sinusitis due to a probable ciliary kinesia disorder, *situs inversus totalis*, and bronchial thickening, suggested the diagnosis of Kartagener's syndrome.

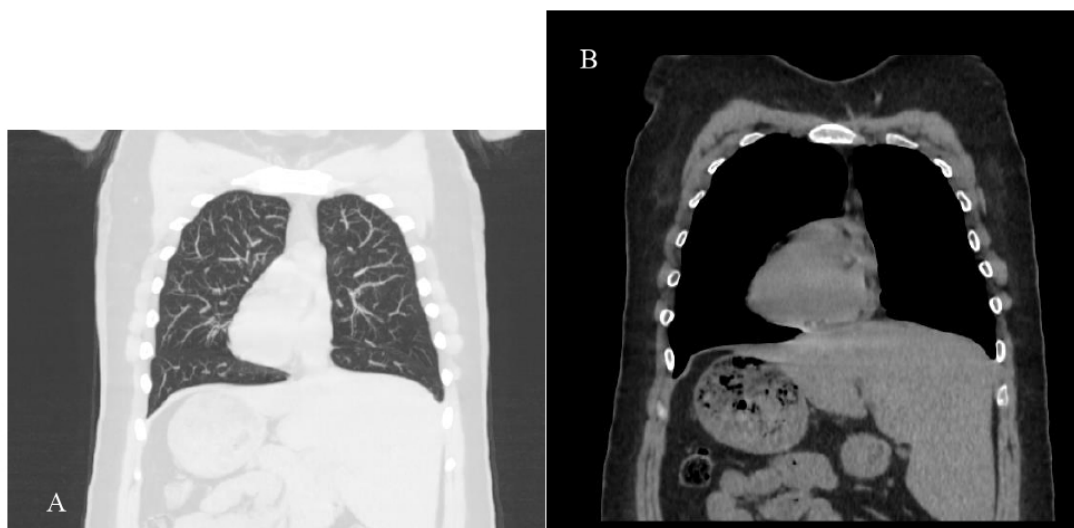


Figure 1: Chest computed tomography. A: Mediastinal window, coronal section; B: Parenchymal window, coronal section.

Source: The authors.

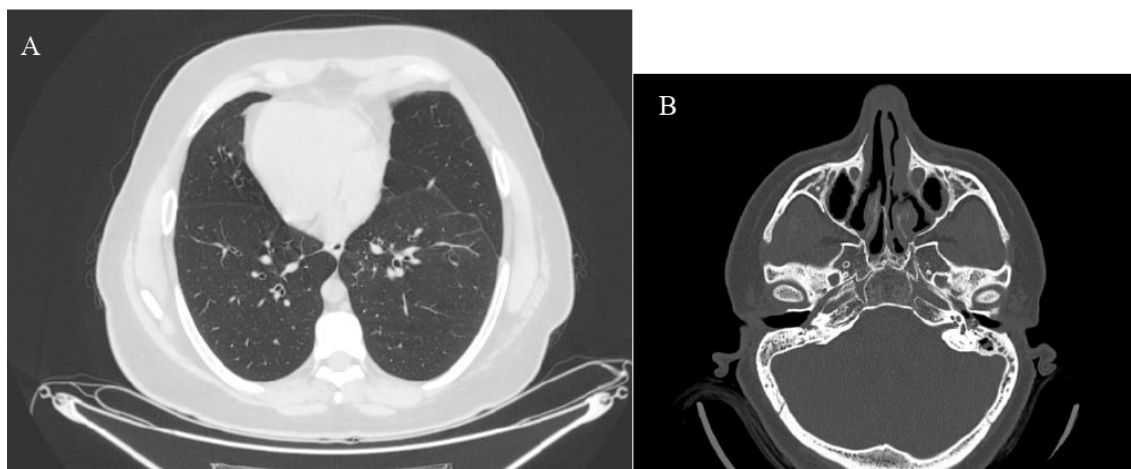


Figure 2: A: Chest computed tomography, parenchymal window, axial section; B: Computed tomography of the paranasal sinuses, axial section.

Source: The authors.

DISCUSSION

PCD is a genetic disease of an autosomal recessive character, characterized by ciliary dysfunction, which changes the frequency and/or pattern of ciliary beat, resulting in impaired mucociliary clearance, leading to chronic infections and inflammation^{8,9}. PCD has subgroups, one of which consists of chronic sinusitis, bronchiectasis, and *situs inversus*, components of the triad that characterizes Kartagener's syndrome. The *situs inversus* is explained by the need for ciliary function in the embryological development of cardiac laterality, occurring in 50% of cases in patients with PCD^{2,10}.

The upper respiratory tract is commonly involved with rhinosinusitis and otitis media, which account for much of the associated morbidity in childhood. In the lower respiratory tract, symptoms manifest as chronic productive cough, an attempt to compensate for the lack of effective mucociliary clearance; if this compensation does not occur, the patient is susceptible to recurrent episodes of pneumonia or bronchitis. As the disease progresses, new clinical manifestations arise, one of which is bronchiectasis. Over time, there may be a decline in lung function, showing a decrease in forced expiratory volume in 1 second (FEV1), leading to chronic respiratory failure and even lung failure, requiring a transplant. Regarding fertility, men, due to the deficiency in the motility of the sperm, end up infertile; women may have reduced fertility, due to ciliary abnormalities in the fallopian tubes, which hinder the passage of egg cells, predisposing to ectopic pregnancy⁸.

There are several differential diagnoses that make early diagnosis of PCD difficult. Among them are respiratory distress neonatal, laterality defects, cystic fibrosis, asthma, allergic

rhinitis, immunodeficiencies, interstitial lung disease, and idiopathic bronchiectasis⁸.

Regarding the triad of Kartagener's syndrome, the diagnosis of chronic sinusitis and bronchiectasis is suspected by clinical symptoms and can be confirmed through imaging exams, which can show: mucosal thickening or partial or complete opacification of the paranasal sinuses on plain radiography and polyps in the nasal cavity or middle meatus on CT^{4,5}. *Situs inversus* does not have serious adverse consequences of its own and is generally not detected until a chest X ray is obtained. This finding becomes a very useful sign when associated with PCD¹¹. Generally, the diagnosis of Kartagener's syndrome is late, mainly because bronchiectasis is not present at birth and is less prevalent in young people, making it difficult to suspect the condition. Treatment and support for chronic sinusitis, otitis media, infertility, and the other manifestations of PCD are individualized. For exacerbations of chronic lung disease, management is based, in part, on the experience of patients with cystic fibrosis and other forms of bronchiectasis¹². The prognosis demonstrates a slow decline in lung function, although recurrent or chronic infections negatively impact the quality of life⁶.

While there is no gold standard diagnostic test, physicians can rely on ancillary tests to guide clinical suspicion. Through the prophylaxis and treatment of complications, a better quality of life is possible for the patients. Finally, the maturation of promising new diagnostic methods is still awaited.

Conflicts of Interest

The authors declare that there is no conflict of interest.

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