

# Kartagener's Syndrome: A Case Report in Adult Southeast Asian Woman with Unspecified Respiratory Symptoms

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**Abstract:** *Kartagener's syndrome (SK) is a rare chronic genetic disorder characterized by a triad of chronic sinusitis, bronchiectasis, and situs inversus. This case report presented a case of a 30-year-old woman with complaints of nausea, chronic cough, and a history of chronic sinusitis since 10 years ago. On chest physical examination, crackles were heard in both lung fields, and a dominant heart sound was heard in the right hemithorax. Laboratory examination revealed increased leukocytes with an increase in neutrophils. Chest X-ray showed a situs inversus with bronchiectasis. The patient underwent a waters x-ray examination, and sinusitis was found in the left maxillary sinus. The patient then received antibiotic therapy, nebulization, and other supportive drugs. Kartagener's syndrome diagnosis is often delayed because less typical symptoms are often found in children and adolescents. Early diagnosis and treatment are necessary to prevent the more severe and progressive condition.*

**Keywords:** Kartagener, Kartagener's, bronchiectasis, sinusitis, situs inversus, PCD, ciliary

## 1. Introduction

Kartagener's syndrome (KS) is part of a larger group of ciliary motility disorders called Primary Ciliary Dyskinesias (PCD), which are genetic conditions with rare autosomal recessive inheritance. This syndrome includes the clinical triad of chronic sinusitis, bronchiectasis, and *situs inversus*.

<sup>1</sup>Although Siewart first described this condition in 1904, it was Kartagener who recognized the clinical condition, the etiological correlation between the elements of the triad, and reported four cases in 1933. The estimated prevalence of PCD is about 1 in 30,000, although it can range from 1 in 12,500 to 1 in 50,000. A normal ciliary function is essential in the respiratory system and sperm motility and ensures proper visceral orientation during embryogenesis. In KS, gene mutations in DNAI1 and DNAH5 cause impaired ciliary motility, leading to recurrent sinopulmonary infections, infertility, and errors with left-right body orientation. <sup>2</sup>Early diagnosis is important for preserving lung function, increasing quality of life, and life expectancy in this disease. <sup>3</sup>We reported a case of a 30-year-old woman with Kartagener's syndrome.

## 2. Case Report

A 30-year-old woman was hospitalized at the Wangaya Hospital, Denpasar, Bali on June 5, 2021, with complaints of nausea and vomiting. Vomiting occurred with every food or fluid intake. The symptoms occurred 6 times in the last 8 hours prior to admission. The patient said that her body felt weak. The patient also complained of a long-lasting cough with thick green phlegm that comes and goes, especially in the morning. The patient said that her breath was often heavy, but she denied shortness of breath. For the patient's previous medical history, she had a history of sinusitis for approximately 10 years, since the patient was 20 years old. The patient had an allergy to the antibiotic levofloxacin. There was no history of other diseases.

On physical examination, the patient's blood pressure was 100/60 mmHg, pulse 107x/minute, respiratory rate

24x/minute, body temperature 37°C, and 95% oxygen saturation in room air. On the chest physical examination, auscultation revealed vesicular breath sounds in both lung fields, and there were crackles in both lung fields, especially in the basal area, but no wheezing was found. On cardiac examination, heart sounds 1 and 2 were normal, single, with no murmur, but the heart sound was dominant in the right hemithorax. Laboratory examination revealed increased leukocytes up to 21,810/microliter, and the complete blood count showed increased neutrophils, with a fairly high neutrophil/lymphocyte ratio of 10.58, indicating the possibility of bacterial infection. Chest X-ray showed dextrocardia with bronchiectasis. The patient underwent a waters x-ray examination, and sinusitis in the left maxillary sinus was found.

The patient then received antibiotic therapy of Cefoperazone 2x1 g intravenously, nebulized with combivent 1 resp. every 8 hours, N – a cetylcysteine 1x1 fls intravenously and extra injection of methylprednisolone 62.5 mg iv, if dyspnea was present.



Figure 1: Waters X-ray Examination



Figure 2: Thorax X - ray Examination

### 3. Discussion

Primary ciliary dyskinesia / Kartagener's syndrome is a genetically inherited disorder. The structures that make up the cilia, including the inner and/or outer dynein arms, central apparatus and radial, etc., are missing or not functioning properly, making the ciliary structures unable to move. Defects in the ciliary component cause abnormal ciliary movement, resulting in impaired mucociliary clearance and manifest as recurrent and/or persistent synopulmonary infections.<sup>4</sup> Primary ciliary dyskinesia is a chronic disease. Its symptoms can be experienced from birth, during childhood, and adolescence. There is a relatively small impact on patients who are healthy and they can usually function normally. However, in the mid - 20s, there is a persistent and progressive increase in respiratory symptoms.<sup>5, 12 - 15</sup>

The diagnostic criteria for Kartagener syndrome are clinical features showing recurrent chest infections, bronchitis, and rhinitis since childhood, with one or more of the following: (1) *situs inversus* in the patient/sibling; (2) live but immobile spermatozoa; (3) reduced or absent transbronchial mucociliary clearance; and (4) cilia showing characteristic of ultrastructural defects on electron microscopy.<sup>6, 14</sup>

A striking and biologically interesting aspect of Kartagener's syndrome is that half of the patients have *situs inversus*. However, the occurrence of *situs inversus* is independent of chronic respiratory symptoms.<sup>7</sup> Bronchiectasis in patients with Kartagener's syndrome is defined as localized and irreversible dilatation of parts of the bronchial tree. The involved bronchi are dilated, inflamed and easily folded, resulting in airflow limitation, obstruction and impaired clearance of secretions, which can easily lead to respiratory infections, contributing to the purulent expectoration commonly observed in these patients. The results are bronchial injury, dilation, impaired clearance of secretions, recurrent infection, and further bronchial damage. In clinical practice, this condition is most commonly characterized by coughing and daily

mucopurulent sputum production lasting from months to years. The classic triad of chronic cough, excessive purulent sputum production, and recurrent infections is seen in most patients.<sup>8</sup>

The next component of the triad of Kartagener's syndrome is chronic sinusitis, an acquired condition that occurs in nearly 100% of patients with this disorder. Accumulation of secretions in the paranasal sinuses occurs following the abnormal ciliary movement. Occasionally, Kartagener's syndrome may be associated with reversible airflow obstruction. The clinical progression of the disease varies, and lung transplantation may be required in severe cases.<sup>9, 15</sup>

In addition to the criteria above, two types of tests are performed for the diagnosis – screening tests (measurement of nasal nitric oxide, which is usually low in PCD, and saccharin test to assess mucociliary function of the nasal epithelium) and diagnostic tests (analysis of ciliary pulse pattern and frequency using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect). To examine the ciliary motility and ultrastructure, samples for this test can be obtained by biopsy of the nasal mucosa.<sup>10</sup>

Treatment of Kartagener's syndrome is based on preventing recurrent infections that can worsen bronchiectasis, resulting in more severe airflow limitation and a more rapid decline in lung function, along with disfiguring conditions due to possible severe pneumonia with risk of sepsis. At diagnosis, the goals of respiratory management are improvement of lung function and restriction of disease progression.<sup>11</sup>

### 4. Conclusion

Kartagener's syndrome is a rare chronic genetic disease characterized by sinusitis, bronchiectasis, and *situs inversus*. Diagnosis is often delayed because of the symptoms often found in children and adolescents, which causes late diagnosis, usually at the conditions of severe lung disorders. Therefore, early diagnosis and treatment are necessary to prevent more severe and progressive disease conditions.

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