ATİPİK KARTAGENER SENDROMU

Atypical Presentation of Kartagener's Syndrome

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ÖZET

Kartagener sendromu situs inversus ve solunum sisteminde ilerleyici hasarla görülen nadir, otozomal resesif genetik silier bir bozukluktur. Bu vaka takdiminde 53 yaşında bir kadın hastada görülen atipik Kartagener sendromu tartışılmıştır.

Anahtar kelimeler: *Kartagener sendromu; Bronşektazi; Situs inversus*

ABSTRACT

Kartagener's syndrome is a rare, autosomal recessive genetic ciliary disorder with progressive damage of the respiratory system and situs inversus. The present case discusses atypical Kartagener's syndrome in 53-year-old woman.

Key words: *Kartagener's syndrome; Bronchiectasis; Situs inversus*

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INTRODUCTION

Ciliary motility disorders may be congenital or acquired. Congenital disorders are named as primary ciliary dyskinesia (PCD). Kartagener's syndrome, a well-known triad of situs inversus, bronchiectasis and sinusit was encompasses about half of the subjects with PCD, which is a rare, typically autosomal recessive disorder and characterized by defective ciliary structure and function and chronic oto-sino-pulmonary symptoms (1, 2). The incidence of the process is 1–2/30 000 births (3). A defect in the coordinated ciliary motility leads to the disease complex which in cludes recurrent sinusitis, lower respiratory tract infections leading to bronchiectasis, otitis (4). Motile cilia on the embryonic node have an important role determining left right symmetry accounting for PCD patients having situs inversus (5). Male infertility due to impaired sperm motility, female infertility and ectopic pregnancy can occur (6). PCD can clinically manifest between 4 and 51 years of age; more frequently it is associated with nasal symptoms or chronic sputum production (7). Delay in recognizing this disorder is common. High index of suspicion and awareness for PCD should be in individuals with a compatible clinical presentation, along with a detailed history and meticulous physical examination.

Although treatment for patients with this syndrome has yet to be established, it is important to control chronic lung infections and declining lung function. We present a clinical case of atypical Kartagener's syndrome in a 53-year-old woman.

CASE REPORT

A 53-year-old woman with a past medical history of chronic obstructive pulmonary disease (COPD), situs inversus (she learned this condition at 48 years old after traffic accident), recurrent pulmonary infections, sinusitis presented to our hospital with pulmonary complaints. Her vital signs were stabil and chest physical examination was unremarkable. An examination of the repiratory system revealed inspratory ralles at the basal of the lungs. Her laboratory work-up wasWhite blood cell count: 13.0µl, C-reactive protein: 2.86/ ml. A chest X-ray revealed dextrocardia. Spirometry demonstrated restrictive pattern, FEV1, FEV1/FVC were 76 % and 92 %. High-resolution computed tomography (HRCT) thorax revealed bronchiectasis and situs inversus (Fig 1-3). A clinical diagnosis of Kartagener'ssyndrome was made. She was diagnosed as atypical presentation of Kartagener's syndrome.

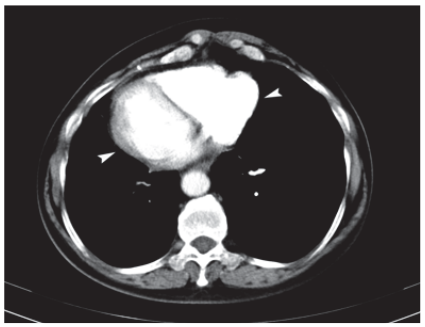

Figure 1. Thorax CT examination shows dextrocardia.

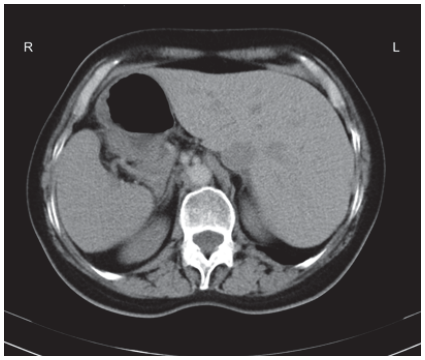

Figure 2. In the CT examination, Liver is seen in the left side

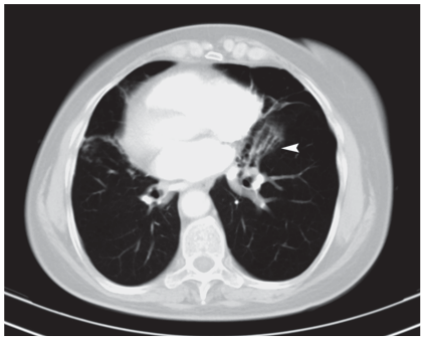

Figure 3. In left paracardiac area bronchectatic segment is seen.

DISCUSSION

The triad of recurrent sinusitis, bronchiectasis and situs inversus form the classical presentation of Kartagener's syndrome. The diagnosis of Kartagener's syndrome may be delayed because the syndrome, characterized by bronchitis, sinusitis and otitis, can be easily confused with common infections. The delayed diagnosis of the disease can translate into adverse consequences for the patient, including insufficient care or inappropriate treatment. But in our delayed case demonstrated a non-progressive course of the bronchiectasis as in another case report (8). Occasionally, Kartagener's syndrome may be associated with reversible airflow obstruction (9). Clinical progression of the disease is variable with lung transplantation required in severe cases.
The clinical characteristics of Kartagener's syndrome are productive cough, respiratory tract infections, sinusitis, otitis media and infertility.Our patient had all pulmonary symptoms but she was fertile and had two children.

In a study of 47 adult patients, with well defined ultrastructural disorders, the following changes were observed: cough (100 %), bronchiectasis (98 %), otitismedia (92 %), history of neonatal respiratory symptoms (65 %), sinusitis (47 %) and situs inversus (46 %) (10).

A systematic investigation of the underlying etiology in all bronchiectasis cases is very important, especially in those that have more specific treatment. In order to carry out the most effective tests, the initial assessment must be oriented by the clinical and family history.
Differential diagnoses for immotile-cilia syndrome in clude malignancy, interstitial lung diseases including idiopathic pulmonary fibrosis and idiopathic interstitial pneumonias, and other conditions associated with bronchiectasis which include acquired (foreign body aspiration, tumor, lymphadenopathy, chronic obstructive pulmonary disease, and mucoid impaction) and congenital bronchial obstruction (bronchomalacia, pulmonary sequestration, and yellow nail syndrome), recurrent infection (immunodeficiencies abnormal secretion disorder (cysticfibrosis), and other miscellaneous conditions (alpha-1 antitrypsin deficiency and connective tissue disease) (11).
Diagnosis of PCD has relied on identification of ciliary dysmotility and specific ciliary ultrastructural defects in outer dyneinarms, inner dyneinarms or central apparatus. But we should have not done these tests to our patient. Diagnosis was made clinically.

The objective of the treatment should be the prevention of chronic lung lesions and bronchiectasis (12). The two pillars of respiratory treatment are antibiotic therapy and thoracic physiotherapy. In general, antibiotics are used during exacerbations of the disease. Patient was followed up with regularly scheduled appointment severy 6 months, including additional visits during the exacerbations. The prognosis is generally considered favorable, and life expectancy is usually normal.
Although the management of patients with Kartagener's syndrome remains uncertain and evidence is limited, it is important to follow up these patientswith an adequate and shared care system.

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