

## A rare association of Kartagener's syndrome with mixed connective tissue disorder

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### Abstract:

We hereby, present a female with Kartagener's syndrome associated with mixed connective tissue disorder. Kartagener's syndrome is a rare congenital malformation comprising triad of sinusitis, situs inversus and bronchiectasis. Mixed connective tissue disorder is an autoimmune disorder comprising of scleroderma, systemic lupus erythematosus, myositis and presence of antibody to ribonucleoprotein called U1-RNP.

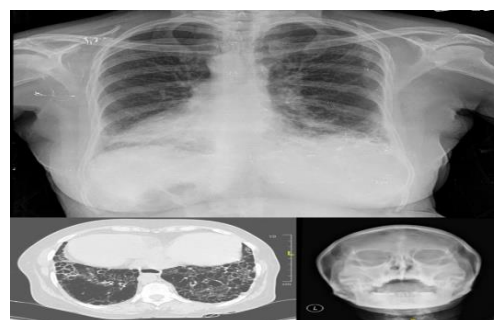
**Keywords:** Bronchiectasis; Kartagener's Syndrome; Mixed connective tissue disorder; Usual interstitial pneumonitis.

### Introduction:

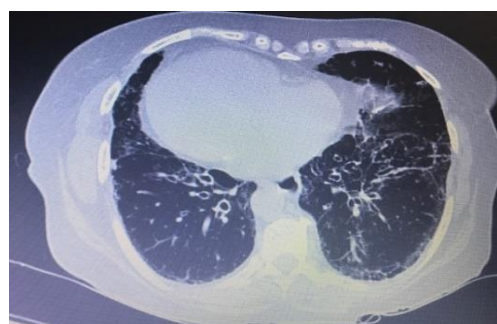
Kartagener's syndrome is an autosomal recessive condition characterized by triad of chronic sinusitis, situs inversus and bronchiectasis [1]. It is a subset of immobile ciliary disorder called primary ciliary dyskinesia. It was first described by Siewart in 1904 [2], but the etiological correlation was established by Kartagener. The prevalence of primary ciliary dyskinesia is 1 in 30,000 populations, but as such only few case series of Kartagener disease has been mentioned in literature studies [2]. There is recurrent chest infection, sinusitis and infertility due to ultrastructural defect in cilia in Kartagener's disease. Mixed connective tissue disease was recognized by Sharp in 1972 with overlapping clinical features of scleroderma, systemic lupus erythematosus, myositis, rheumatoid arthritis and the presence of an antibody against ribonucleoprotein called U1-RNP [4]. Association of Kartagener's disease and mixed connective tissue disease is rare entity. This case represents an unusual association of two rare conditions and thus is of clinical importance to the treating physicians.

### Case Report:

A 52-year-old married female presented with complaints of cough with expectoration on and off since childhood, dyspnoea, dry mouth, epigastric pain and occasional fever. She was a known diabetic and hypertensive on medications. Her chest X ray showed dextrocardia, stomach bubble in the right side of abdomen with bilateral lung lower zone reticular shadows (Figure. 1). Ultrasound abdomen was done which was suggestive of situs inversus totalis with reversal of arrangement of all organs. X-ray PNS showed maxillary sinus haziness (Figure. 1). ECG was also consistent with dextrocardia. HRCT showed bilateral honeycombing and traction bronchiectasis of the bilateral lower lung fields which was suggestive of a usual interstitial pneumonia pattern (Figure. 2).



**Fig. 1: Chest X-ray showing dextrocardia with bronchiectasis, High resolution CT thorax showing Bronchiectasis, X- Ray PNS showing haziness of the maxillary sinuses**



**Fig. 2: HRCT thorax showing usual interstitial pneumonitis pattern of interstitial lung disease**

Sputum for AFB was negative. Other serological investigations were done which showed anti-nuclear autoantibody positive, anti-U1 ribonucleoprotein autoantibody positive and anti-Smith autoantibody positive suggestive of mixed connective tissue disease. Her serological marker for rheumatoid arthritis was found to be negative. Salivary scintigraphy showed impaired uptake and clearance function of bilateral sub-mandibular and right parotid salivary glands. Pulmonary function test with diffusion capacity of lung was done which showed severe restriction.

**Discussion:**

Ciliary motility disorders can be primary or secondary. About 50% of primary ciliary dysfunction (PCD) has situs inversus and they form the group called Kartagener's syndrome. PCD is a heterogeneous condition where there is defect in the ultrastructure of the cilia affecting the dynein arms and thereby the functional mobility [1,2]. The immobility of the cilia leads to stasis of secretion causing bronchiectasis, recurrent sinusitis and infertility. Both male and female are affected equally but females present at a later age than males [3]. Female fertility is reduced, however majority of females were able to reproduce [5]. Kartagener's syndrome is diagnosed due to situs inversus, but PCD remains mostly undiagnosed [6].

Mixed connective tissue disease (MCTD) is a systemic autoimmune disease where pulmonary involvement is not seen clinically in the early course of the disease. In a study done in 1976, pulmonary disease was present in 80% of MCTD patients while 69% of them were clinically asymptomatic [7]. Recently, it has been seen that fibrosing alveolitis and pulmonary hypertension has become the major complications of MCTD. In a recent study, about 48% of MCTD presents with interstitial lung disease [7].

The association of MCTD with primary bronchiectasis is found to be about 12% with majority of cases being cystic fibrosis [8]. The radiological presentation of MCTD on HRCT thorax is varied ranging from ground glassing, nodular shadows to interstitial fibrosis. Kozuka et al. 2001, did a radiological evaluation of HRCT thorax of 41 patients and found the predominant findings are that of ground glassing attenuation followed by subpleural micronodules and non-septal linear opacity with lower lobe peripheral predominance. The less common findings observed was honeycombing, traction bronchiectasis, interlobular septal thickening with airspace consolidation [8].

Our case had an association of MCTD with fibrosing alveolitis (UIP) pattern. The association of primary bronchiectasis with MCTD presenting as UIP pattern is very rare and is of clinical importance since management of MCTD requires steroid and immunosuppressant which might exacerbate the infection in bronchiectasis.

**Conclusions:**

Kartagener's syndrome associated with interstitial lung disease can complicate the clinical course of the patients. Kartagener's syndrome can present as bilateral bronchiectasis, which is a usual manifestation. But other coexistent lung diseases like ILD (UIP) can also be present and can lead to accelerated lung function decline in the patients. Hence a very high index of suspicion is needed to diagnose the presence of other coexistent lung pathologies.

**Conflicts of interest:** None declared

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