

Kartagener's syndrome; two cases in siblings

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Section: Chest imaging

Area of Interest: Respiratory system Abdomen

Procedure: Diagnostic procedure

Imaging Technique: Digital radiography

Imaging Technique: CT

Special Focus: Congenital Case Type: Clinical Cases

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Patient: 26 years, male

Clinical History:

Two brothers (26-year-old and 17-year-old) with clinical history of chronic cough with sputum, sometimes haemoptysis and dyspnoea since birth, were referred to our radiology department. The patients also had a history of headache with chronic nasal discharge.

Imaging Findings:

Case 1 (26-year-old): The chest radiography shows dextro-cardia and right-sided fundic air bubble. The chest CT revealed cystic bronchiectasis throughout both lungs predominantly in the right lower lobe and some of the dilated bronchi are filled with mucus. Significant numbers of opacities with tree-in-bud appearance in the lungs bilaterally with slightly increased in lungs volume. Dextro-cardia is also appreciated with right-sided aortic arch. Included sections from the abdomen demonstrate inverse position of the liver and spleen. Paranasal sinuses radiography shows significant mucosal thickening in the bilateral maxillary, frontal and ethmoid sinuses, representing sinusitis.

Case 2 (17-year-old): The Chest CT shows bilateral bronchiectasis with significant small pulmonary opacities with tree-in-bud appearance. Dextro-cardia is visualized with right-sided aortic arch. Included sections from the upper abdomen show inverse position of the liver and spleen. Mucosal thickening is noted in the maxillary sinuses bilaterally on radiography.

Discussion:

Kartagener's syndrome was first described in 1904 by Siewert as a condition characterized by a combination of bronchiectasis, chronic sinusitis and situs inversus. However, Manes Kartagener recognized this clinical triad as a distinct congenital syndrome in 1933. [1, 4, 5, 6]

Kartagener's syndrome is a subset of primary ciliary dyskinesias, characterised by ultrastructural genetic defect causing impaired ciliary motility resulting in recurrent chest and sinus infections and infertility. Although patients are symptomatic since childhood the age at which diagnosis is made is variable. [3, 5]

Kartagener's syndrome (KS) is a rare genetic condition, inherited via an autosomal recessive pattern [2, 4, 5, 6]. Its incidence is about 1 in 20, 000-30, 000 live births [2, 4, 5]. Male patients with this syndrome are almost invariably

infertile because of immotile spermatozoa. [4, 5]

The pathophysiology of primary ciliary dyskinesia is the absence and/or dysmorphic features of the dynein arms in airway cilia due to a variety of specific mutations in cilia coding genes. [1, 5, 6]

The symptoms of this condition are non-specific and common in many other more frequent respiratory conditions. Furthermore, clinical manifestations can easily be mistaken for commonly occurring infections. [1]

Patients with primary ciliary dyskinesia suffer from recurrent chest and sinus infections since childhood but diagnosis is usually delayed due to low index of suspicion.

Early diagnosis may help in improvement of lung function, quality of life and life expectancy.

It is more common in middle and lower lobes of the lungs. The frontal sinuses are hypoplastic and often nasal polyps are also seen. [2]

A high index of suspicion is required for early diagnosis to offer appropriate treatment for infertility and preservation of lung function. [3]

Screening tests include exhaled nitric oxide and saccharin test to assess nasal epithelial muco-ciliary function.

Biopsy of nasal or tubal mucosa and ciliary beat pattern & frequency analysis using video recording, and electron microscopic confirmation of the ultrastructural ciliary defect is considered diagnostic. In our cases these tests were not performed and the diagnosis was clinico-radiological as in various earlier case reports.

In conclusion, primary ciliary dyskinesia and Kartagener's syndrome should be considered as a possible diagnosis in young patients with lower and upper respiratory tract infections. [2]

Differential Diagnosis List: Kartagener's syndrome, Cystic Fibrosis, Primary ciliary dyskinesia

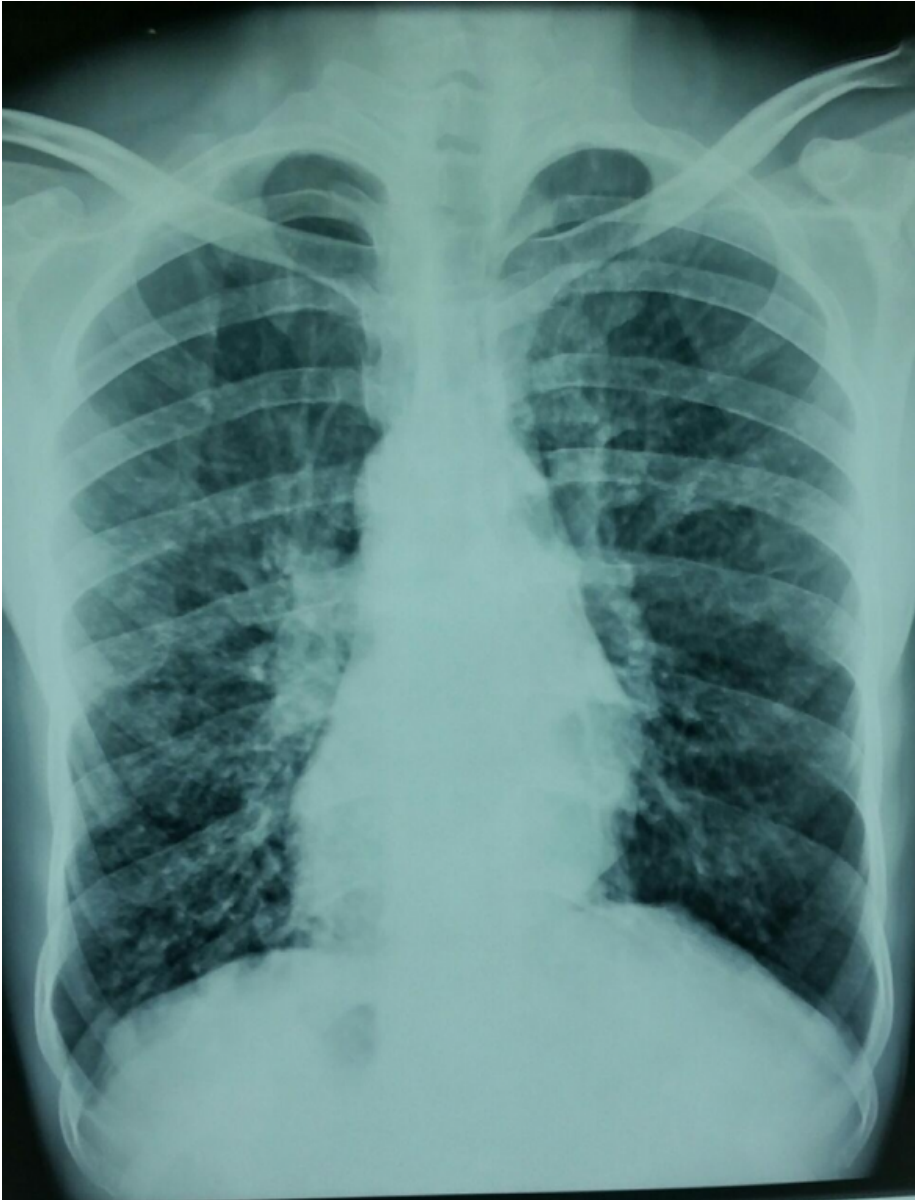
Final Diagnosis: Kartagener's syndrome

References:

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Figure 1

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Description: Chest radiograph; showing dextrocardia and fundic bubble on the right side. **Origin:** Radiology Department, FMIC, Kabul, AF

Figure 2

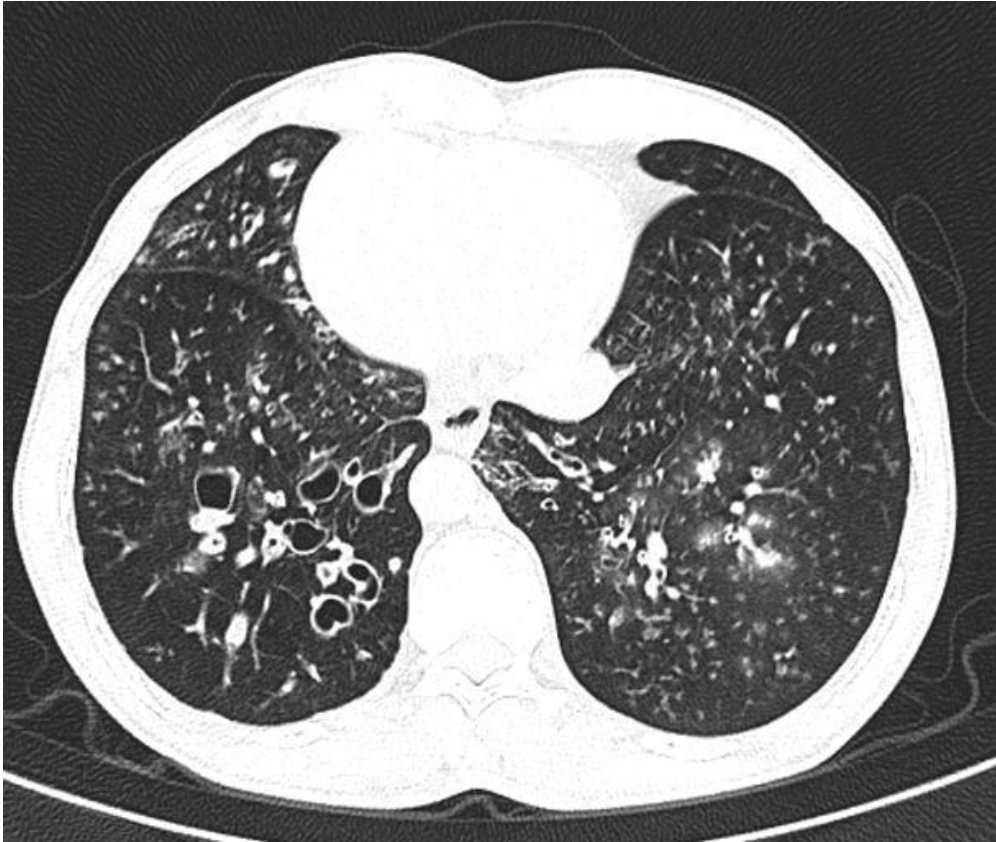
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Description: Chest topogram; shows dextrocardia and right side gastric air bubble. **Origin:** Radiology Department, FMIC, Kabul, AF

Figure 3

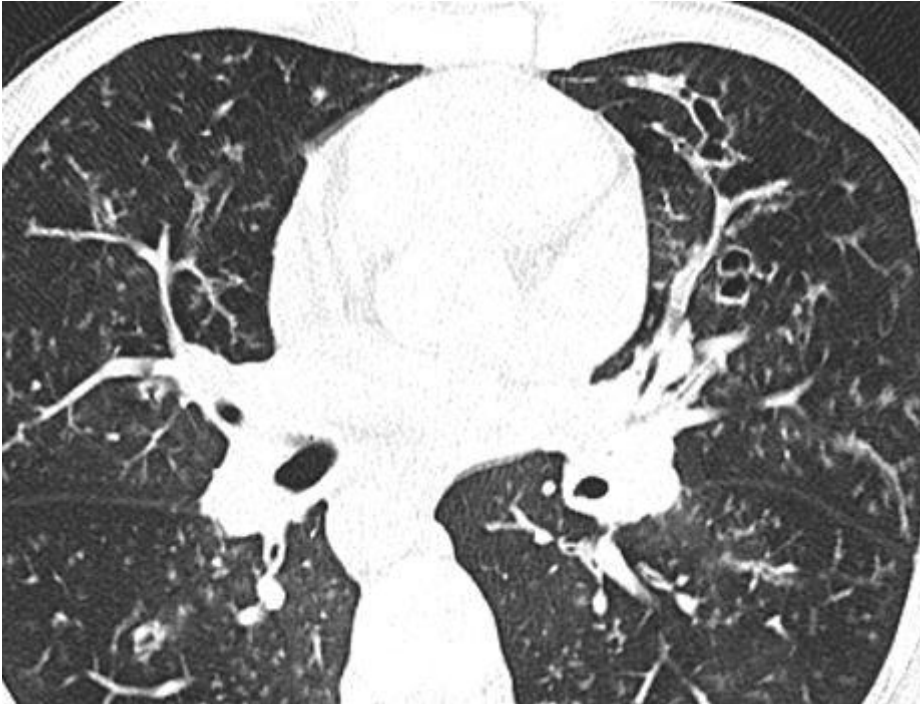
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Description: Axial CT section; shows bronchiectasis in the right lower lobe. **Origin:** Radiology Department, FMIC, Kabul, AF

Figure 4

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Description: Axial CT section; shows bronchiectasis in the left upper lobe. **Origin:** Radiology Department, FMIC, Kabul, AF

Figure 5

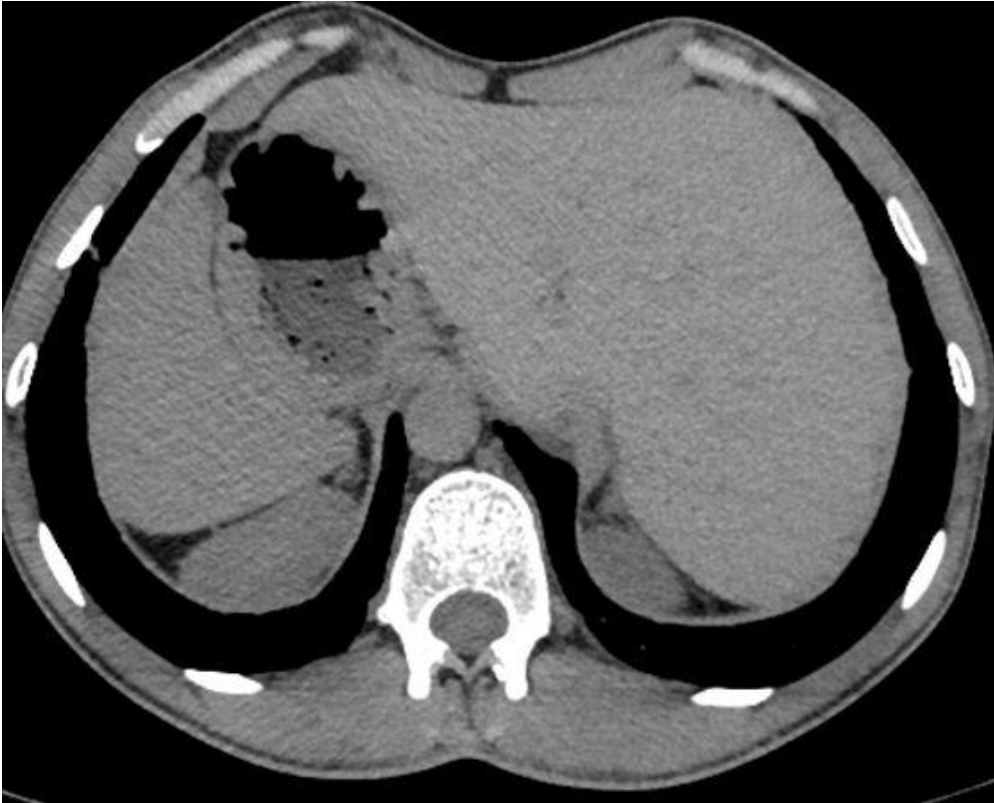
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Description: CT abdomen axial section; showing transposition of viscerae (spleen in the right side and liver in the left side). **Origin:** Radiology Department, FMIC, Kabul, AF

Figure 6

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Description: CT abdomen axial section; shows right-sided spleen and left-sided spleen and stomach, suggesting Situs Inversus. **Origin:** Radiology Department, FMIC, Kabul, AF

Figure 7

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Description: PNS radiograph; shows chronic sinusitis. **Origin:** Radiology Department, FMIC, Kabul, AF

Figure 8

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Description: PNS radiograph; shows chronic maxillary sinusitis with hypoplasia of frontal sinuses.

Origin: Radiology Department, FMIC, Kabul, AF