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RESEARCH ARTICLE

CLINICAL MANIFESTATIONS AND DIAGNOSTIC CHALLENGES OF KARTAGENER SYNDROME

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Abstract

Kartagener syndrome (KS) is an uncommon autosomal recessive genetic disorder characterized by the triad of bronchiectasis, chronic rhinosinusitis, and situs inversus. It is a specific phenotype within the spectrum of primary ciliary dyskinesia (PCD), a condition involving dysfunctional ciliary motility. Early diagnosis is crucial for effective management of the associated respiratory complications.

Introduction:

Casereport

A 16-year-old female presented with a persistent asthmatic cough in the absence of dyspnea and fever.
She reported experiencing these symptoms since childhood but noted a significant worsening over the past year. His medical history included multiple hospitalizations for respiratory infections.

Chest X-ray and chest CT were performed : Extensive bronchiectasis was observed, with mucus plugging and peribronchial thickening (Figure 1 (A,B). A right-sided aortic arch was also observed, along with situs inversus viscerum characterized by the hepatic shadow on the left and the gastric shadow on the right (Figure3).

An abdominal CT scan confirmed situs inversus totalis, revealing a left-sided liver and a right-sided spleen (Figure 4), while a sinus CT demonstrated pansinusitis (Figure 5).

This case illustrates the prototypical presentation of Kartagener Syndrome, characterized by bronchiectasis, chronic rhinosinusitis, and situs inversus. The patient's history of recurrent respiratory infections, coupled with distinctive radiological findings, was crucial in establishing the diagnosis of KS.

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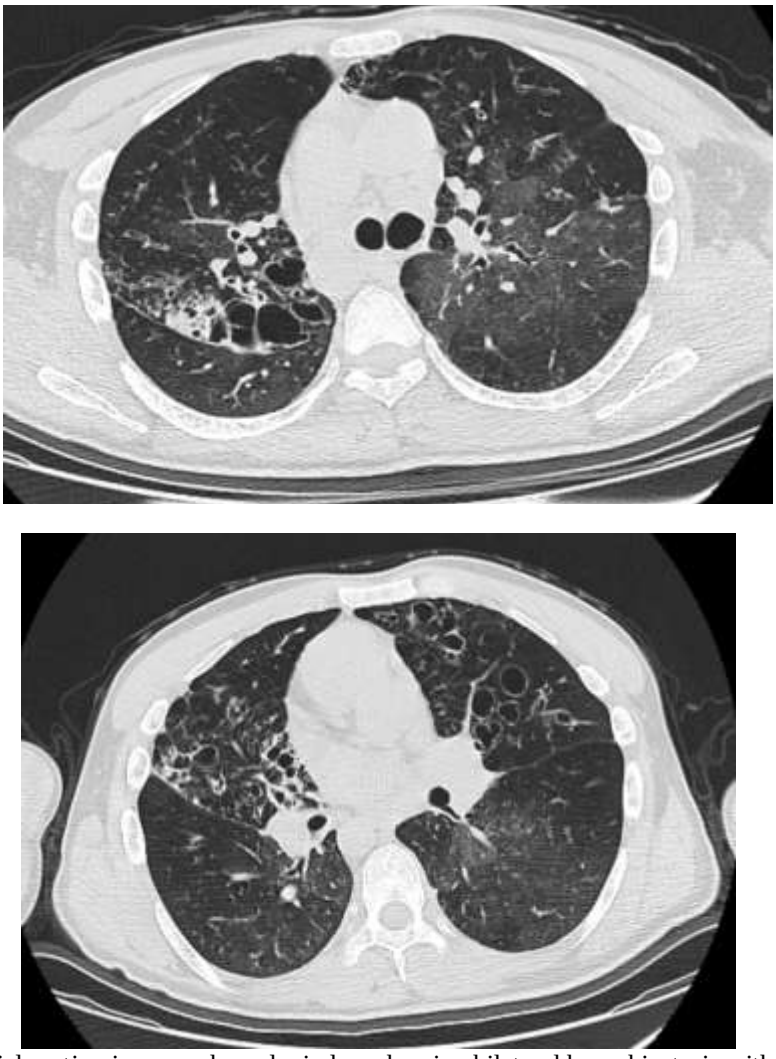


Figure 1:- CTaxial section in parenchymal window showing bilateral bronchiectasis with some mucoide impactions.

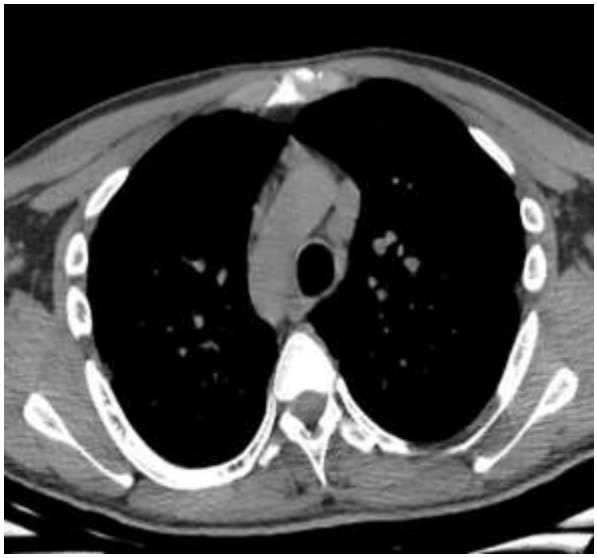


Figure 2:- Chest CT scan, axial slice in mediastinal window showing aortic arch oriented to the right.

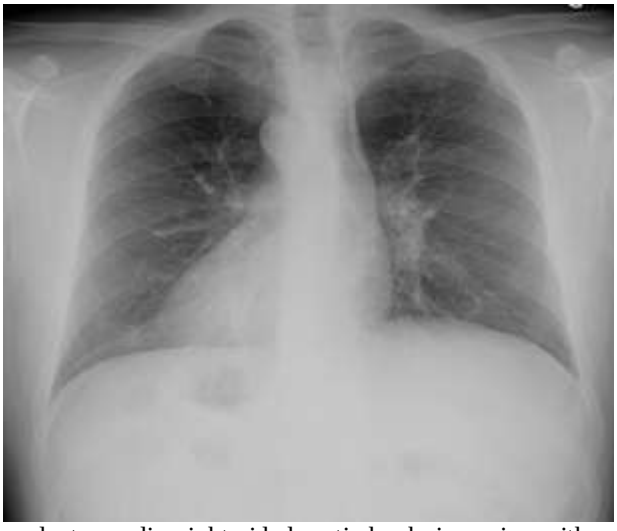


Figure 3:- Frontal chest X-ray: dextrocardia, right-sided aortic knob, inversion with mirror image positioningof the hepatic opacity on the left and the gastric air bubble on the right."

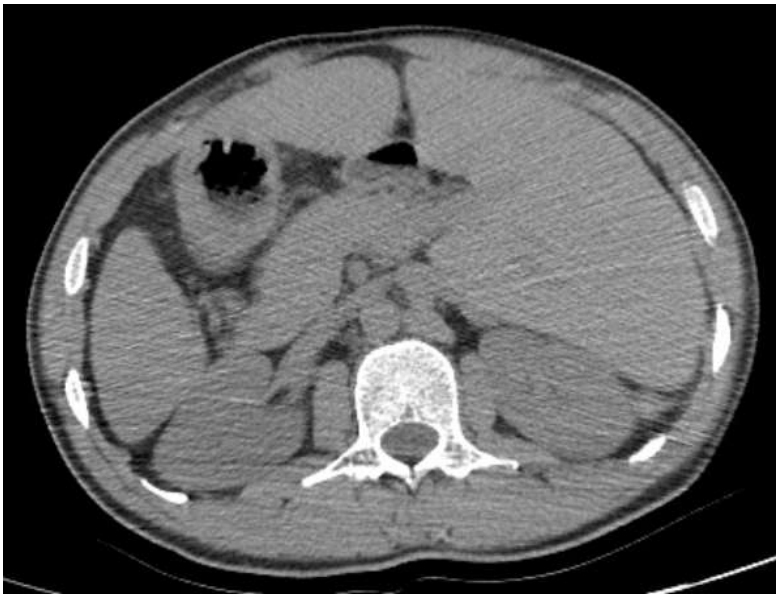
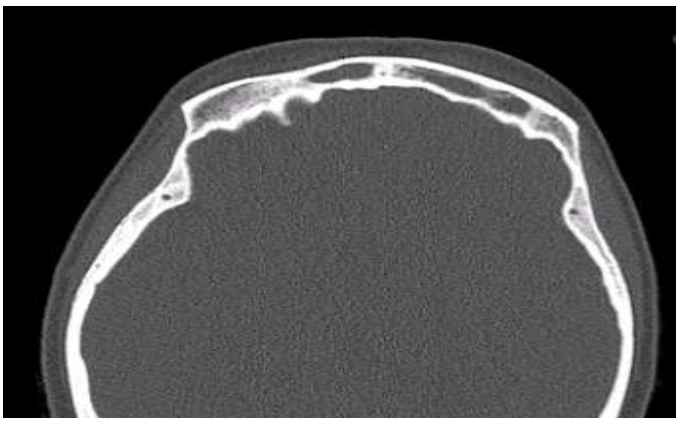
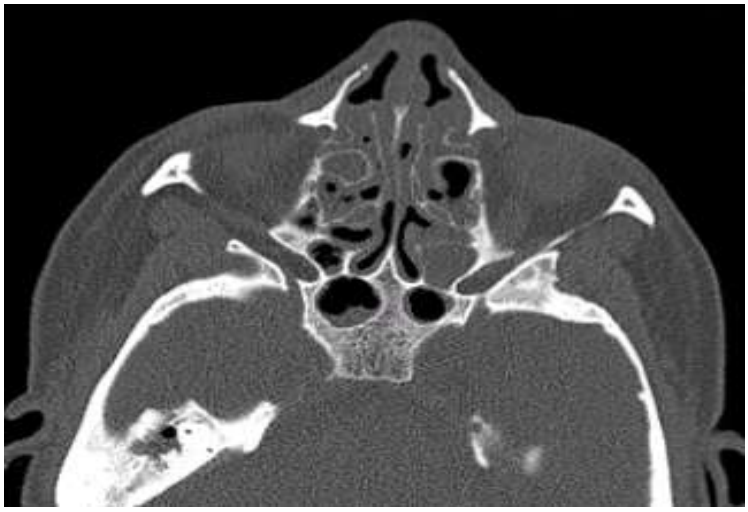


Figure 4:- Abdominal scan, axial slice in parenchymal window showing the spleen and stomach on the right, the liver is located in the left hypochondrium.





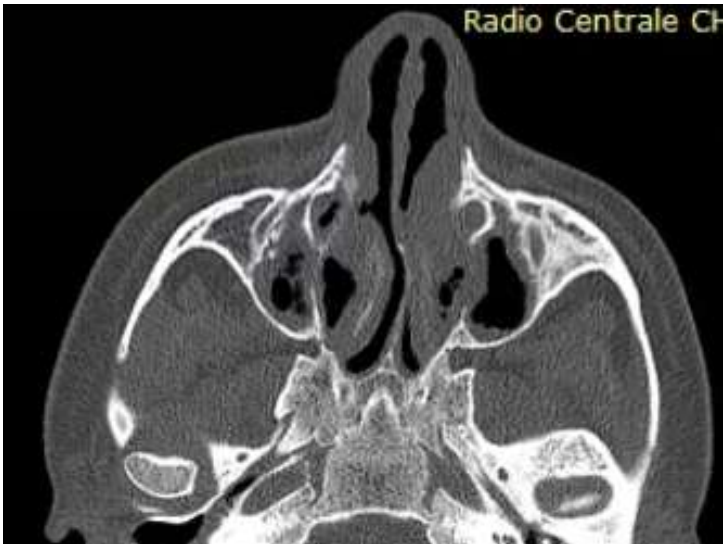


Figure 5 (A,B,C):
CT appearance suggestive of pansinusitis Total opacification of the frontal sinus and partial opacification of someethmoidalcells."
Partial opacification of the maxillary sinus."
Partial opacification of the sphenoidal sinus."
Thickening of the superior, middle, and inferiorturbinates."
"Slightdeviation of the nasal septum."
"Irregularerosion of the anteriorwall of the right maxillary sinus

Discussion:

The clinical presentation and radiological findings in this case are consistent with those described in the literature.

According to O'Callaghan et al. (2010) (1), the triad of bronchiectasis, chronic sinusitis, and situs inversus is pathognomonic of KS. Our patient's HRCT findings of extensive bronchiectasis and sinus CT findings of chronic sinusitis are typical for KS, as noted by Kennedy et al. (2007).(3)

Diagnosing KS can be challenging due to its overlap with other respiratory conditions. In this case, the presence of situs inversus provided a critical diagnostic clue. Genetic testing confirmed the diagnosis, highlighting the importance of considering genetic evaluation in suspected cases of PCD/KS. (4)

Early diagnosis of KS is essential for implementing appropriate management strategies to prevent disease progression and improve quality of life. This includes regular monitoring, prompt treatment of infections, and physiotherapy to enhance mucus clearance. (3)

Further research is needed to explore the genetic basis of KS and the development of targeted therapies. Studies focusing on long-term outcomes of patients with KS and the efficacy of various management strategies are also warranted. (3)

Kartagener Syndrome, although rare, should be considered in patients with recurrent respiratory infections and situs anomalies. Radiological imaging plays a crucial role in the diagnosis, and genetic testing can confirm the diagnosis. Early and appropriate management is vital for improving patient outcomes. (5)

Radiological imaging contributes to the diagnosis and management of Kartagener syndrome. Early recognition of characteristic imaging findings, such as situs inversus and bronchiectasis, facilitates timely intervention and improves patient outcomes. Further research is necessary to refine imaging protocols and enhance therapeutic strategies for patients with this rare genetic disorder.(6)

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