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# A case of Kartagener's syndrome with combined aplasia of frontal and sphenoid sinuses and hypoplasia of maxillary and ethmoid sinuses

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## Abstract:

Kartagener's syndrome (KS), characterized by a triad of bronchiectasis, chronic sinusitis, and situs inversus, is a subset of an autosomal recessive hereditary disorder of primary ciliary dyskinesia. We report the case of a 35-year-old male who presented with a history of intermittent episodes of productive cough, breathlessness, and cold since childhood. High resolution computed tomography of chest revealed bronchiectatic changes, dextrocardia, and right-sided aortic arch. Computed tomography (CT) scan of the abdomen revealed situs inversus. CT of the paranasal sinuses revealed combined aplasia of bilateral frontal and sphenoid sinus with sinusitis. Based on these findings, a diagnosis of KS was made. There was no complaint of infertility, which usually accompanies KS, even though an analysis of his seminal fluid revealed reduced count and reduced motility of sperms. The uniqueness of our case is that our patient was a male aged 35 years; besides, the third decade is an unusual age for presentation of combined aplasia/agenesis of bilateral frontal and sphenoid sinuses with hypoplasia of maxillary and ethmoid sinuses. Even though these findings have been reported in children and young adults, there are very few case reports of such a presentation in adults in literature.

## Keywords:

Aplasia, bronchiectasis, Kartagener's syndrome, paranasal sinus, situs inversus

## Introduction

A subset of primary ciliary dyskinesia (PCD) is known as Kartagener's immotile cilia syndrome or Kartagener's syndrome (KS). In 1975, Camner *et al.*, first observed that ciliary dyskinesia was the cause of KS. In 1977, in order to categorize infertility with chronic sinopulmonary infections, Eliasson *et al.*, first coined the word "immotile cilia syndrome" for KS.<sup>[1,2]</sup> The classical triad of bronchiectasis, sinusitis, and situs inversus constitutes Kartagener's syndrome. It is an autosomal recessive disease characterized by the irregular ciliary structure or function,

leading to impaired mucociliary clearance. With the disordered ultra structure of cilia in the body, there is sluggish and uncoordinated ciliary beating. The awkward ciliary movement contributes to low mucociliary transport in the respiratory system, leading to sinusitis and persistent bronchiectasis, and infertility due to the immotility of sperms. The immotile sperms have flagellar mutations with their tails in which the axonemes lack dynein arms with spoken heads. Dynein is an ATPase protein associated with the development of ciliary movement energy. During the embryonic stage, synchronized ciliary activity causes spontaneous intestinal rotation, resulting in situs inversus in about half of the affected individuals.

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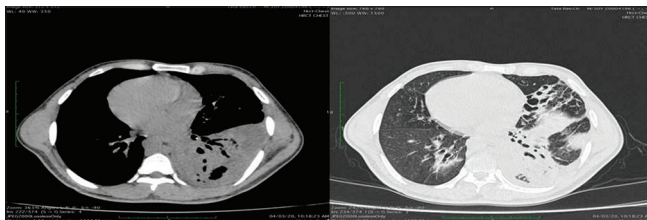
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We present the case of a 35-year-old male patient who was referred to the department of radio-diagnosis to undergo high resolution computed tomography (HRCT) of the chest and computed tomography (CT) of paranasal sinuses, with a history of recurrent episodes of productive cough, breathlessness, and cold since childhood. He had been treated symptomatically on and off for his complaints but only had temporary relief.

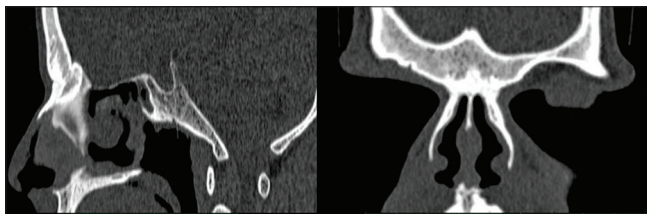
### Case Report

The patient was of moderate built with no evidence of pallor, cyanosis, or clubbing. No other family member had similar problems. He was married with two male children aged 8 years and 10 years who have no such complaints. The patient had a history of symptomatic treatment but no history of any radiographic investigations such as chest X-ray or ultrasound.

Routine blood investigations were normal, and sputum was negative for acid-fast bacilli. HRCT scan of the chest revealed varicose type of bronchiectatic changes in the left upper lobe and right middle and lower lobe segments, with areas of consolidation in the basal segments of the left lower lobes, and mild left pleural effusion, with no associated lymphadenopathy, mass lesion, or ground glassing [Figure 1]. Dextrocardia was seen with right-sided aortic arch. CT scan of the abdomen revealed situs inversus, with the liver on the left and spleen on the right side with no other abnormality [Figure 2]. CT paranasal sinuses revealed complete aplasia/agenesis of frontal and sphenoid sinus and hypoplastic maxillary and ethmoid sinuses with features of sinusitis [Figures 3 and 4].



**Figure 1:** High-resolution computed tomography thorax showing consolidation in the basal segments of the left lower lobe. Varicose type of bronchiectasis in the left upper lobe and right middle and lower lobe segments. Mild left pleural effusion

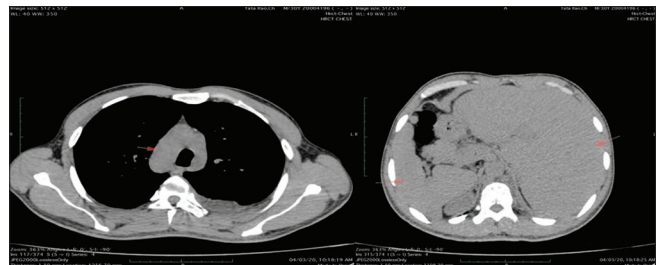


**Figure 3:** Computed tomography of paranasal sinuses showing sagittal image of aplasia of sphenoid and coronal image showing aplasia of frontal sinuses

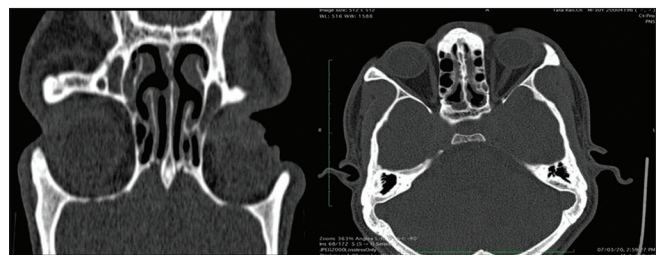
With the findings of bronchiectasis, sinusitis, dextrocardia, and situs inversus, a diagnosis of KS was considered. The patient's semen analysis revealed reduced count and motility of sperms. He had two male apparently normal children. We report this case because of the rare presence of combined aplasia of frontal and sphenoid sinus and hypoplastic maxillary and ethmoid sinuses.

### Discussion

Kartagener's syndrome, a rare (1 in 30,000) clinical triad of bronchiectasis, chronic sinusitis, and situs inversus, is an autosomal recessive hereditary disorder of defective ciliary movement.<sup>[3]</sup> It is one of the subsets of PCD. Efficient clearing of mucous and other secretions from the respiratory tract and paranasal sinuses is facilitated by the coordinated and synchronous beating of the cilia, lining their walls. Abnormal ciliary beatings result in bronchiectasis and sinusitis.<sup>[2]</sup> Ciliary dysmotility in the embryonic stage results in the failure of the heart and other organs to move to the left side, resulting in dextrocardia and situs inversus, respectively.<sup>[4,5]</sup> The infertility seen in few cases results from the defective motility of spermatozoa and impaired ciliary motility in the Fallopian tubes.<sup>[6]</sup> Patients with KS usually present with recurrent lower respiratory tract infections, chronic productive cough, chronic sinusitis from childhood, and sometimes infertility in adults. Afzelius recognized the relation between KS and male infertility.<sup>[7]</sup>



**Figure 2:** Mediastinal window showing right-sided aortic arch and axial image of the abdomen showing liver on the left and spleen on the right



**Figure 4:** Computed tomography paranasal sinuses showing coronal image of hypoplasia of the bilateral maxillary sinuses and axial computed tomography showing hypoplastic ethmoid air cells

Although sinusitis is a common presentation, aplasia/agenesis of more than one sinus and/or associated hypoplasia of other sinuses is rare. Under any circumstances, the aplasia/agenesis of more than one sinus group of paranasal sinuses is an unusual or rare presentation. Though unilateral agenesis of frontal sinus is relatively common, bilateral agenesis of frontal sinus is relatively rare and occurs only in 4%–10% of the population. Agenesis of the sphenoid sinus is extremely rare, seen in only 1%–1.5% of cases. Impaired mucociliary clearance results in frequent respiratory tract, paranasal sinus, and ear infections; chronic nasal congestion; and infertility. Recurrent lower respiratory tract infection leads to bronchiectasis. The dilated bronchi are prone to colonization by *Haemophilus influenzae*, *Staphylococcus aureus*, and *Pseudomonas* species.<sup>[8]</sup> Most persons commonly seek medical help because of infertility. Situs inversus and dextrocardia result from disorganized ciliary beating in the embryonic stage, which results in random rotation of the primitive organ precursors to either left or right. Therefore, dextrocardia is not found in all cases of KS.

Other abnormalities found in KS include nasal polyps, rhinitis, corneal abnormalities, and conductive deafness. Radiological investigations include chest X-rays and HRCT chest, which can reveal dextrocardia, bronchiectasis, and infiltrates. X-ray of the paranasal sinuses can show features of sinusitis in the form of haziness in the sinuses. Ultrasonography of the abdomen can be used to determine the presence of situs inversus. HRCT of the chest and CT scan of the paranasal sinuses remain the modality of choice<sup>[9]</sup> to determine bronchiectasis and sinusitis, respectively. Semen analysis of postpubertal males may reveal either abnormal sperm motility or aspermia. Echocardiography can also be used to confirm dextrocardia with normal atrioventricular concordance without any structural abnormality. Routine blood count reveals no abnormality.

The uniqueness of our case lies in the findings of combined aplasia of bilateral frontal and sphenoid sinus with hypoplasia of ethmoid and maxillary sinuses in a male patient aged 35 years, an unusual age for such presentations, because previously reported cases have been among children and young adults. A study by Eggesbø *et al.*, on 126 patients, with abnormalities of paranasal sinuses in cystic fibrosis and noncystic fibrosis, revealed that in 4% of cystic cases there was frontal sinus aplasia, but with no case of sphenoid sinus aplasia.<sup>[10]</sup> Similar studies were reported by Lobo *et al.*,<sup>[11]</sup> Jayashankar *et al.*,<sup>[12]</sup> Hailu *et al.*,<sup>[13]</sup> and Tadesse *et al.*

<sup>[14]</sup> The uniqueness of our case is that the patient was a 35 year old male who had combined aplasia of bilateral frontal and sphenoid sinus with hypoplastic ethmoid and maxillary sinuses while previously reported cases were found only in children and young adults.

## Conclusion

Unusual and rare findings in patients diagnosed with KS, such as aplasia of multiple sinuses and/or hypoplasia of one/more sinuses, should not be overlooked or undervalued. Knowledge of these findings is vital for the prevention of any intraoperative complications in patients who undergo surgical procedure for sinus pathologies, in future.

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## Declaration of patient consent

The author certifies that all appropriate consent forms were obtained from the patient's parents for the publication of the case report. Consent was also given for images and other clinical information to be reported in the journal. They were assured that the patients' names and initials will not be published and though every effort will be made to conceal the patient's identity, anonymity cannot be guaranteed.

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Nil.

## Conflicts of interest

There are no conflicts of interest.

## References

1. Camner P, Mossberg B, Afzelius BA. Evidence of congenitally nonfunctioning cilia in the tracheobronchial tract in two subjects. *Am Rev Respir Dis* 1975;112:807-9.
2. Eliasson R, Mossberg B, Camner P, Afzelius BA. The immotile-cilia syndrome. A congenital ciliary abnormality as an etiologic factor in chronic airway infections and male sterility. *N Engl J Med* 1977;297:1-6.
3. Greenstone M, Cole PJ. Primary ciliary dyskinesia. *Arch Dis Child* 1984;59:704-6.
4. Wanner A. Clinical aspects of mucociliary transport. *Am Rev Respir Dis* 1977;116:73-125.
5. Katsuhara K, Kawamoto S, Wakabayashi T, Belsky JL: Situs inversus totalis and Kartagener's syndrome in a Japanese population. *Chest* 1972;61:56-61.
6. Samuel I. Kartagener's syndrome with normal spermatozoa. *JAMA* 1987;258:1329-30.
7. Afzelius BA. A human syndrome caused by immotile cilia. *Science*

- 1976;193:317-9.
8. Rosen MJ. Chronic cough due to bronchiectasis: ACCP evidence based clinical practice guidelines. *Chest* 2006;129:122-31.
  9. Aydinlioğlu A, Kavakli A, Erdem S. Absence of frontal sinus in Turkish individuals. *Yonsei Med J* 2003;44:215-8.
  10. Eggesbø HB, Søvik S, Dølvik S, Eiklid K, Kolmannskog F. Proposal of a CT scoring system of the paranasal sinuses in diagnosing cystic fibrosis. *Eur Radiol* 2003;13:1451-60.
  11. Lobo LJ, Zariwala MA, Noone PG. Ciliary dyskinesia: Primary ciliary dyskinesia in adults. *Eur Respir Mon* 2011;52:130-49.
  12. Jayashankar CA, Somasekar DS, Perugu PK, Reddy KV, Prakash B, Santosh KV. Kartagener's syndrome: A case report. *Scholar J Med Case Rep*. 2014;2:7-10.
  13. Hailu SS, Amerga ED, Gorfu Y, Zewdineh D. Kartagener's syndrome: A case report. *Ethiop Med J* 2016;54:91-4.
  14. Tadesse A, Hailemariam A, Mezgebu S, Gebrewold Y. Kartagener's syndrome: A case report. *J Med Case Rep* 2018;12:5.