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Kartagener's Syndrome Incidentally Discovered During an Etiological Workup: A Case Report

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Abstract **Case Report**

Kartagener syndrome is a peculiar entity among primary ciliary dyskinesias (PCD) characterized by a clinical triad: chronic sinusitis, bronchiectasis and complete or incomplete situs inversus. It is a rare genetic ciliary disease with autosomal recessive inheritance. The fundamental problem lies in defective movement of the cilia, leading to recurrent chest infections. We report a case of Kartagener syndrome discovered incidentally during an etiological work-up for wheezing dyspnea with chronic bronchorrhea.

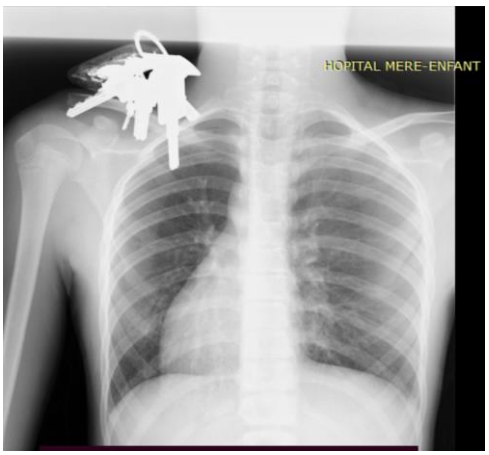
Keywords: Bronchiectasis, Kartagener syndrome, sinusitis, situs inversus, imaging.

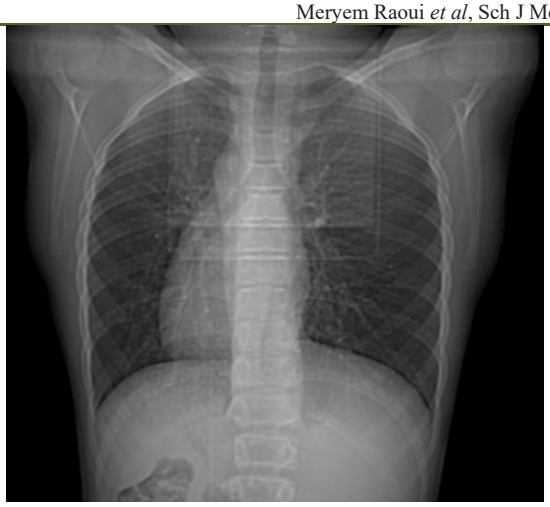
INTRODUCTION

- Kartagener syndrome (KS) is a subset of a larger group of ciliary motility disorders known as primary ciliary dyskinesias (PCD).
- It is a genetic disorder with autosomal recessive inheritance, comprising a triad of situs inversus, bronchiectasis and sinusitis[1,2].
- Although Siewart first described the disease in 1904, it was Kartagener who recognized the etiological correlation between the elements of the triad, reporting four cases in 1933[2].
- In KS, the ultrastructural genetic abnormality results in impaired ciliary motility, leading to recurrent chest, ear, nose and throat (ENT) and sinus infections, as well as infertility in adult subjects.
- Furthermore, although unproven, it seems likely that early diagnosis is important for preserving lung function, quality of life and life expectancy in this disease [3], but this has not been confirmed and further large-scale prospective studies are needed.

PATIENT AND OBSERVATION

- This is a 12-year-old boy presenting with wheezing dyspnea with chronic bronchorrhea. A thoracic radiograph was first requested, which showed a situs inversus. **(figure 1)**

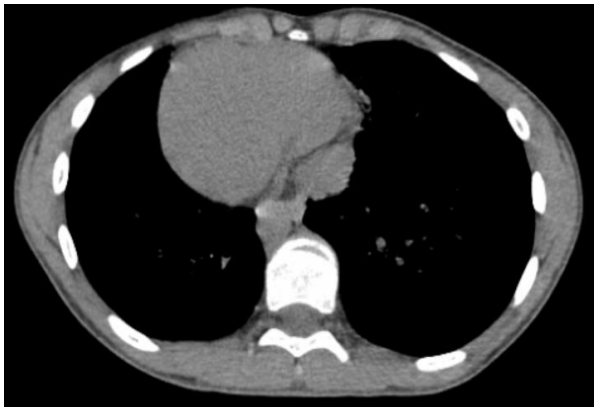




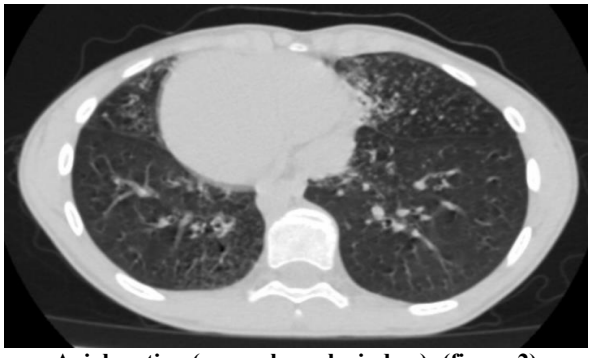
Front chest X-ray:
(figure 1)
-Situs inversus
-Placement of key on patient's right shoulder to confirm diagnosis

- A complementary thoracic CT scan was ordered, which revealed: **(figure 2)**
  - Des foci of cylindrical bronchiectasis in the lower lobar bilaterally, middle lobar and lower lingular, with local mucoid impactions, associated with peribronchial thickening.
  - Micronodules involving the same areas described above, branching out to form a bud-tree appearance.
  - Situs inversus: aortic arch on the right, heart apex on the right.

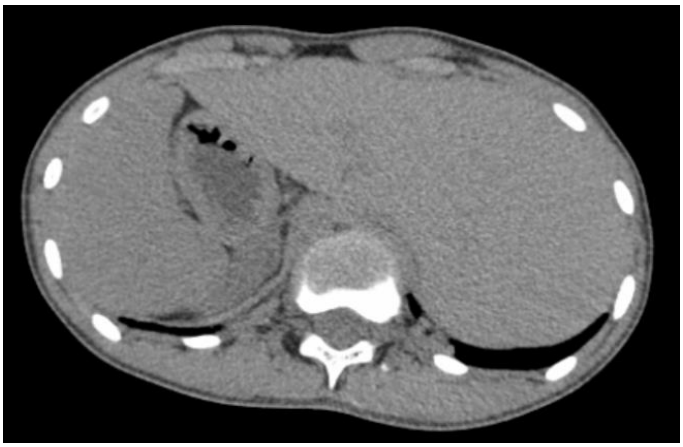
- Basithoracic sections show the stomach and spleen on the right and the liver on the left. **(figure 3)**



Axial section (mediastinal window): (figure 2)
Situs inversus: aortic arch on right, apex of heart on right

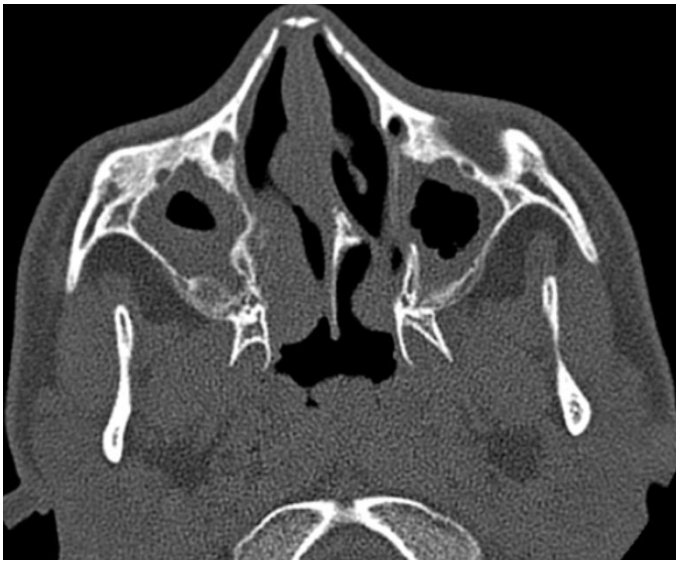


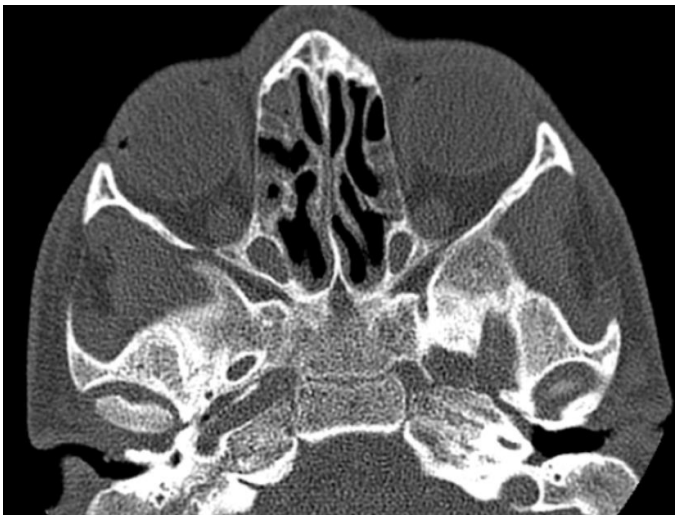
Axial section (parenchymal window) :(figure 2)
Foci of bronchiectasis
Micronodules



Axial section
(figure 3)

- To complete the workup, a CT scan of the sinuses was ordered, showing the presence of pansinusitis with signs of chronicity, associated with nasal polyposis **(figure 4)**.





Axial section (bone window) :(figure 4)
Pansinusitis
Nasal polyposis

- In view of the situs inversus, bronchiectasis and chronic sinusitis, the diagnosis of Kartagener syndrome was first suggested.

DISCUSSION

- Kartagener syndrome is a specific form of a disease called primary ciliary dyskinesia.
- This is a rare genetic disorder affecting the structure and function of the motile cilia present on cells in certain parts of the body, including the respiratory tract, fallopian tubes (women) and sperm flagella (men)[4].

- Kartagener's syndrome is defined by a classic **clinical triad** [4]:
  1. **Bronchiectasis:** Abnormal and irreversible dilatation of the bronchi, leading to mucus accumulation and recurrent respiratory infections.
  2. **Chronic sinusitis:** Persistent sinus infections due to poor mucus clearance.
  3. **Dextrocardia (or situs inversus totalis):** Inversion of the thoraco-abdominal organs from their normal position (the heart is on the right side instead of the left).

- **Main symptoms include:**
  - Infections recurrent breathing due to inability of cilia to clear mucus.
  - Toux chronic and frequent expectoration.
  - Rhinite chronic or persistent nasal congestion.
  - Otites
  - Situs inversus totalis.

- The severity of symptoms and the age at which the disease is diagnosed vary widely, even if symptoms are present from birth.
- Diagnosis of Kartagener's syndrome is based on several tests:
  - Imagerie medical: X-ray or CT scan to observe bronchiectasis and dextrocardia.
  - Biopsie of the nasal mucosa: to analyze cilia under the electron microscope and detect structural abnormalities.
  - Test saccharin: to assess the speed of mucus transport in the nose.
  - Analyse genetics: Identification of disease-causing mutations.

- There is no cure for Kartagener's syndrome, but there are steps you can take to reduce symptoms and prevent complications [3,6]:
  - Kinésithérapie respiratory: to drain mucus from the lungs.
  - Antibiotiques: To treat recurrent respiratory infections.
  - Vaccination: To prevent respiratory infections
  - Surveillance ENT: To treat ear infections, sinusitis and other infections.
  - Procréation Medically assisted reproduction (MAP).

- The clinical course of the disease is variable.
- With appropriate medical follow-up, people with Kartagener's syndrome can lead a normal life.
- However, recurrent respiratory infections can lead to long-term lung damage (such as pulmonary fibrosis).

CONCLUSION

- KS patients are frequently confronted with repeated infectious episodes for which they need to consult, which largely explains their morbidity.

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