

An 18-month-old boy is brought to the emergency department for evaluation of a productive cough and fever. He has had 8 office and emergency department visits over the past year, all of which required antibiotics for diagnoses of sinusitis and pneumonia. He was delivered vaginally without complications. His older brother has had similar recurrent infections throughout his life. Temperature is 38.9 C (102 F), blood pressure is 90/60 mm Hg, pulse is 120/min, and respirations are 30/min. The patient's pulse oximetry is 99% on room air. Examination of the oropharynx reveals mildly enlarged tonsils without erythema or exudate. Auscultation reveals rales and rhonchi in both lungs, and point of maximal impulse is displaced to the right. What is the most likely diagnosis?

- ☐ A. Common variable immunodeficiency
- ☐ B. Cystic fibrosis
- ☐ C. Foreign body
- ☐ D. Kartagener syndrome
- ☐ E. Normal childhood infections
- ☐ F. X-linked agammaglobulinemia

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- ☐ A. Common variable immunodeficiency [4%]
- ☐ B. Cystic fibrosis [12%]
- ☐ C. Foreign body [0%]
- ☒ D. Kartagener syndrome [70%]
- ☐ E. Normal childhood infections [0%]
- ☐ F. X-linked agammaglobulinemia [13%]

Proceed to Next Item

Explanation:

User Id: [redacted]

Features of primary ciliary dyskinesia	
Pathophysiology	Mutations in ciliary dynein arms lead to absent or dysmotile cilia & poor mucociliary clearance
Clinical manifestations	<ul style="list-style-type: none">• Recurrent sinopulmonary infections• Bronchiectasis• +/- Situs inversus (Kartagener syndrome)
Diagnosis	<ul style="list-style-type: none">• Low nasal nitric oxide levels• Bronchoscopy & electron microscopic visualization of ciliary abnormalities• Genetic testing

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This patient has recurrent sinopulmonary infections with a displaced point of maximum impulse (PMI). These are suggestive of **Kartagener syndrome** (KS), a subgroup of primary ciliary dyskinesia (PCD [immotile-cilia syndrome]). PCD is an autosomal recessive disorder characterized by **dysmotile cilia** that result from aberrant production or attachment of ciliary dynein arms. These mutations result in impaired ciliary function, poor mucociliary clearance of secretions, and chronic infections.

KS is characterized by a classic triad of **situs inversus**, **recurrent sinusitis**, and **bronchiectasis**. The typical radiographic finding is **dextrocardia** (apex of the heart is in the right chest), which can be detected on physical examination by displaced heart sounds and PMI to the right. Although there is no gold standard for diagnosis, KS can be diagnosed with a suggestive phenotype and demonstration of abnormal mucociliary transport.

(Choice A) Common variable immunodeficiency results from low immunoglobulin levels and typically presents in adolescence or adulthood. Affected patients exhibit recurrent sinopulmonary infections but not dextrocardia.

(Choice B) Cystic fibrosis is an autosomal recessive disorder characterized by inspissated bronchial secretions, abnormal ciliary function, and repeated respiratory tract infections. However, patients with CF do not have KS.

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(Choice B) Cystic fibrosis is an autosomal recessive disorder characterized by inspissated bronchial secretions, abnormal ciliary function, and repeated respiratory tract infections. It can be distinguished from KS by the presence of pancreatic insufficiency and absence of dextrocardia.

(Choice C) Foreign body aspiration can lead to endobronchial obstruction, retention of secretions, and bronchiectasis but would not present with dextrocardia.

(Choice E) Healthy young children who attend day care or have older siblings may experience up to 12 respiratory infections per year. The majority of these infections are viral in origin and do not require antibiotics. This patient's displaced PMI makes KS more likely.

(Choice F) X-linked agammaglobulinemia can present with recurrent respiratory tract infections, but patients characteristically will have no tonsillar and adenoidal tissue due to lack of mature B cells.

Educational objective:

Kartagener syndrome, a subgroup of primary ciliary dyskinesia, is characterized by a classic triad of situs inversus, recurrent sinusitis, and bronchiectasis.

References:

1. **Kartagener syndrome.**

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References:

1. [Kartagener syndrome.](#)
2. [Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome.](#)

Media Exhibit

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