

AMSER Case of the Month

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35-Year-Old Female Presenting with Dyspnea

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Patient Presentation

- **History of Present Illness:** A 35-year-old female presents to clinic with dyspnea, productive cough, and wheezing.
- **Medical History:** Patient recalls several bouts of pneumonia as a child.
- **Surgical History:** Patient mentions two sinus surgeries, adding that she has residual congestion and pressure.
- **Social History:** Patient formerly smoked ½ pack-per-day.
- **Physical Exam:** Chest auscultation reveals bilateral wheezing.

Pertinent Labs

- **Basic Metabolic Panel:** Results are within normal limits.
- **Complete Blood Count:** Results are within normal limits.
- **Microbiology Sputum Culture:** Final yeast and bacterial cultures are still pending.

What imaging should we order?

ACR Appropriateness Criteria

American College of Radiology
ACR Appropriateness Criteria®
Chronic Dyspnea-Noncardiovascular Origin

Variant 1: Chronic dyspnea. Unclear etiology. Initial imaging.

Procedure	Appropriateness Category	Relative Radiation Level
Radiography chest	Usually Appropriate	☼
CT chest without IV contrast	May Be Appropriate (Disagreement)	☼☼☼
CT chest with IV contrast	May Be Appropriate	☼☼☼
CT chest without and with IV contrast	Usually Not Appropriate	☼☼☼
FDG-PET/CT skull base to mid-thigh	Usually Not Appropriate	☼☼☼☼
MRI chest without and with IV contrast	Usually Not Appropriate	○
MRI chest without IV contrast	Usually Not Appropriate	○
US chest	Usually Not Appropriate	○

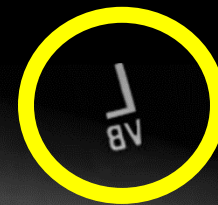
Imaging modality ordered by outpatient pulmonologist.



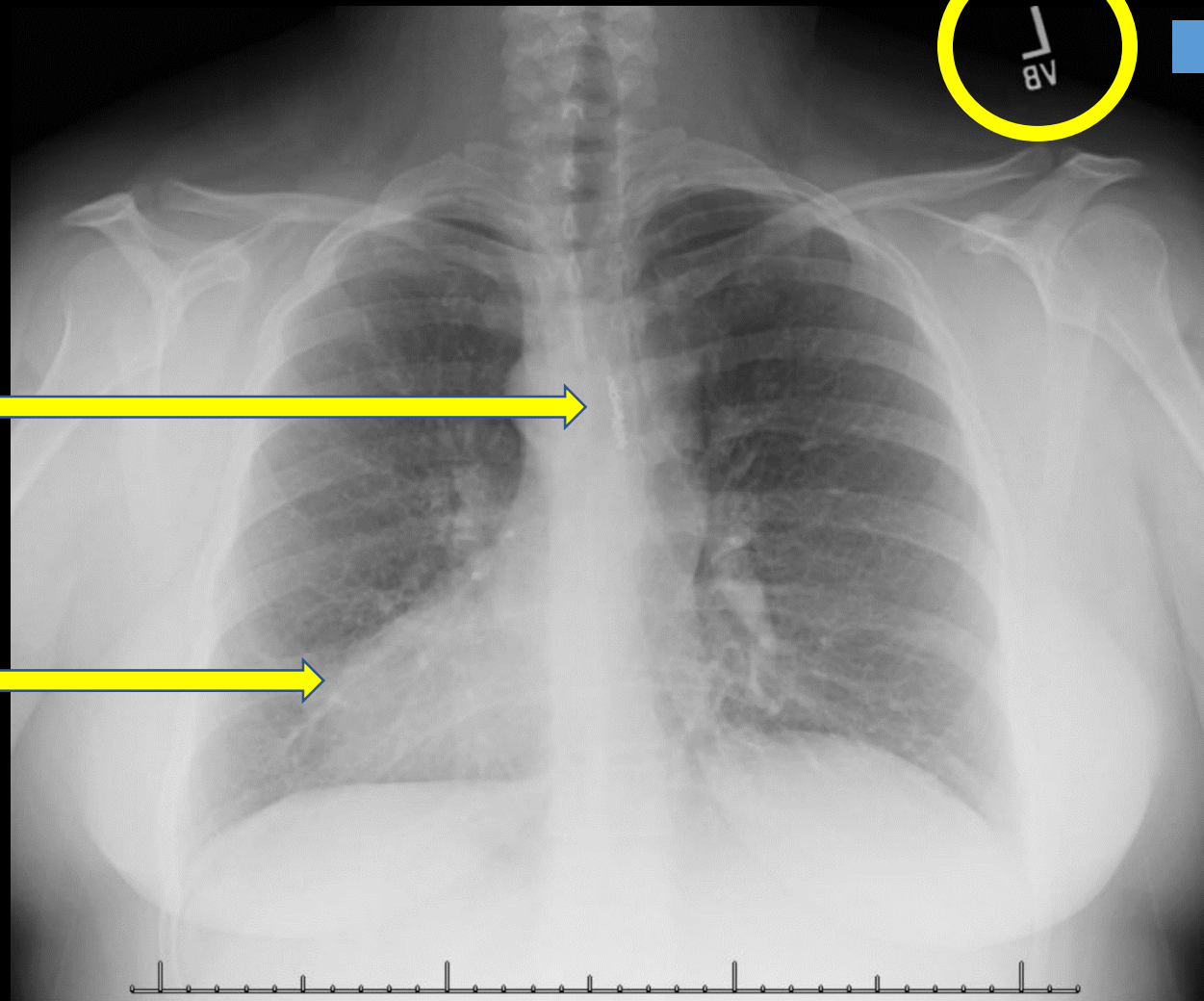
Findings



Findings



Correct Orientation



Embolization Coils



Dextrocardia



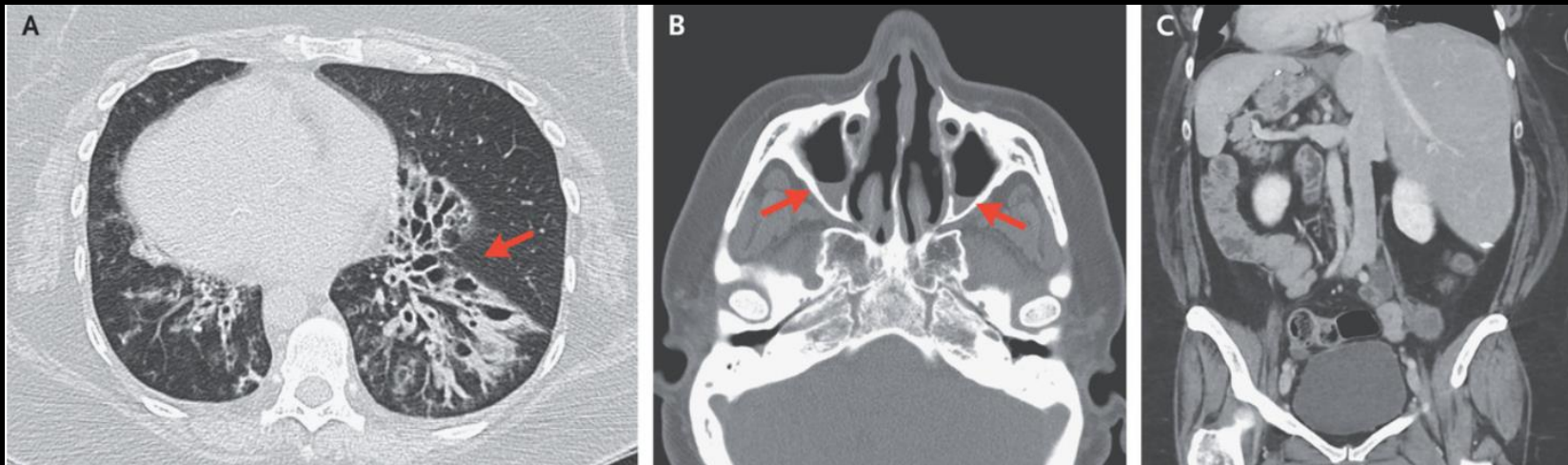
Situs Inversus

Final Diagnosis

Kartagener's Syndrome

Case Discussion: Kartagener's Syndrome

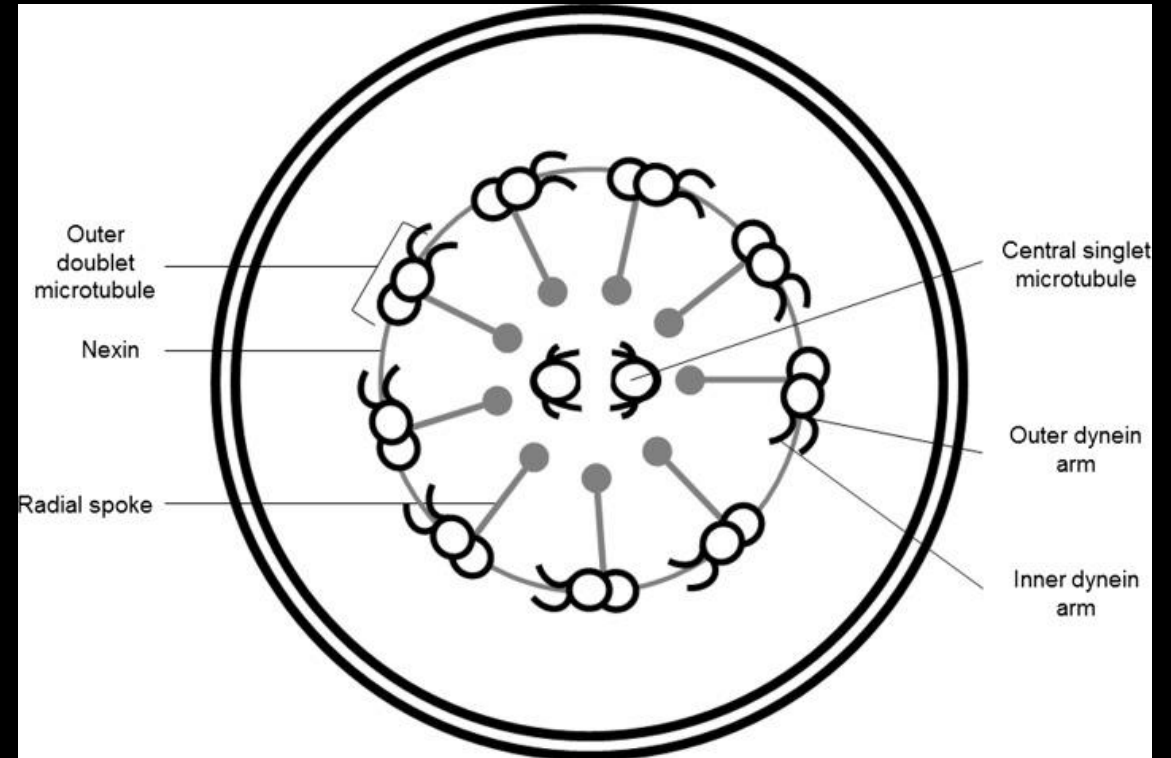
- Kartagener's syndrome is a subtype of primary ciliary dyskinesia
- It presents as a characteristic triad
 - A. Bronchiectasis
 - B. Chronic sinusitis
 - C. Situs inversus



N Engl J Med 2021; 384:e45 DOI: 10.1056/NEJMicm2028152

Case Discussion: Pathophysiology of Primary Ciliary Dyskinesia (PCD)

- Caused by abnormal ciliary beating
 - Most often caused by structural defects, especially dynein arms
- Motile cilia are also present in the lungs, middle ear, paranasal sinuses, reproductive organs, and brain ependyma
 - Therefore, most patients will also have non-chest symptoms, including subfertility
- Motile cilia in the embryonic node help create left-right body symmetry
 - Thus, ~50% of PCD patients will also have situs inversus



Arch Dis Child; 2014 Sep;99(9):850-6. DOI: 10.1136/archdischild-2013-304831.

Case Discussion: Diagnosis and Treatment

Diagnosis

- Current diagnostic guidelines are limited
- A low index of suspicion is recommended for Kartagener's syndrome given risk of irreversible lung damage
- Nasal nitric oxide is often used as a screening tool; diagnosis is then confirmed with higher specificity test, like electron microscopy of nasal or bronchial biopsy
- Other available tests include high-speed videomicroscopy analysis, cell culture of ciliated cells, sperm motility, genetic testing, inhalation of colloid albumin tagged with ^{99}Tc , and saccharin testing

Treatment

- There is a lack of evidence-based medicine in the management of PCD
- Most often, patients are managed empirically
- Consider chest physiotherapy for bronchiectasis and antibiotic treatment for exacerbations
- Chronic sinusitis and otitis media can be managed medically or surgically
- Patients experiencing subfertility can be referred for in vitro fertilization
- Patients generally have normal life expectancy when connected to multidisciplinary care

References

- Lucas JS, Burgess A, Mitchison HM, Moya E, Williamson M, Hogg C; National PCD Service, UK. Diagnosis and management of primary ciliary dyskinesia. *Arch Dis Child*. 2014 Sep;99(9):850-6. doi: 10.1136/archdischild-2013-304831. Epub 2014 Apr 25. PMID: 24771309; PMCID: PMC4145427.
- Mishra M, Kumar N, Jaiswal A, Verma AK, Kant S. Kartagener's syndrome: A case series. *Lung India*. 2012 Oct;29(4):366-9. doi: 10.4103/0970-2113.102831. PMID: 23243352; PMCID: PMC3519024.
- Zurcher K, Kawashima A. Kartagener's Syndrome. *N Engl J Med*. 2021 Mar 25;384(12):e45. doi: 10.1056/NEJMicm2028152. Epub 2021 Mar 20. PMID: 33764707.