

AMSER Case of the Month

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HPI 23-year-old female with history of pneumonia presenting with chronic dyspnea

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Patient Presentation

HPI: 23-year old female presenting with chronic dyspnea

PMH: prior pneumonias throughout childhood

Physical Exam: Bilateral wheezing

Labs: Noncontributory

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

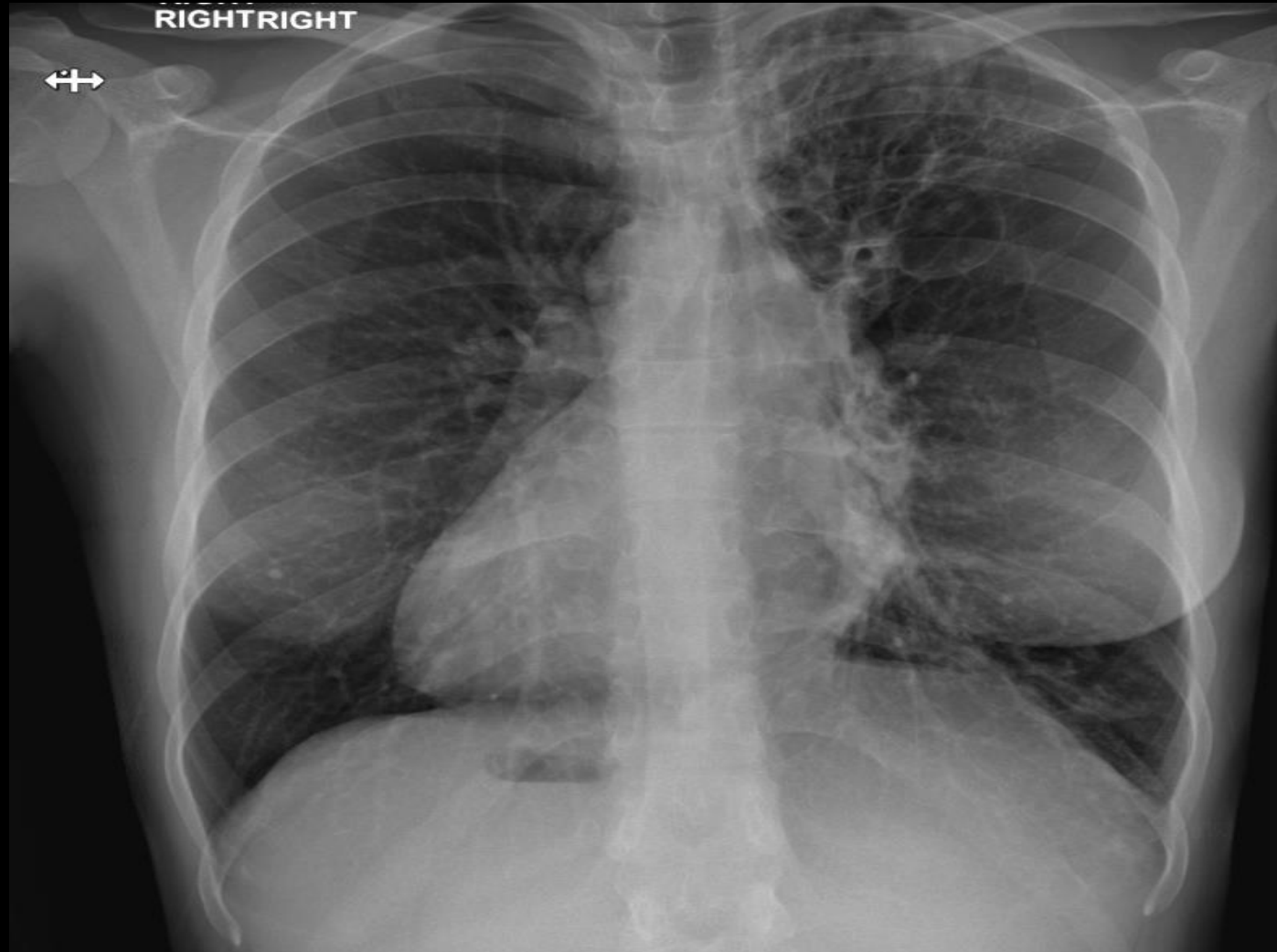
Variant 1: **Chronic dyspnea. Unclear etiology. Initial imaging.**

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	○

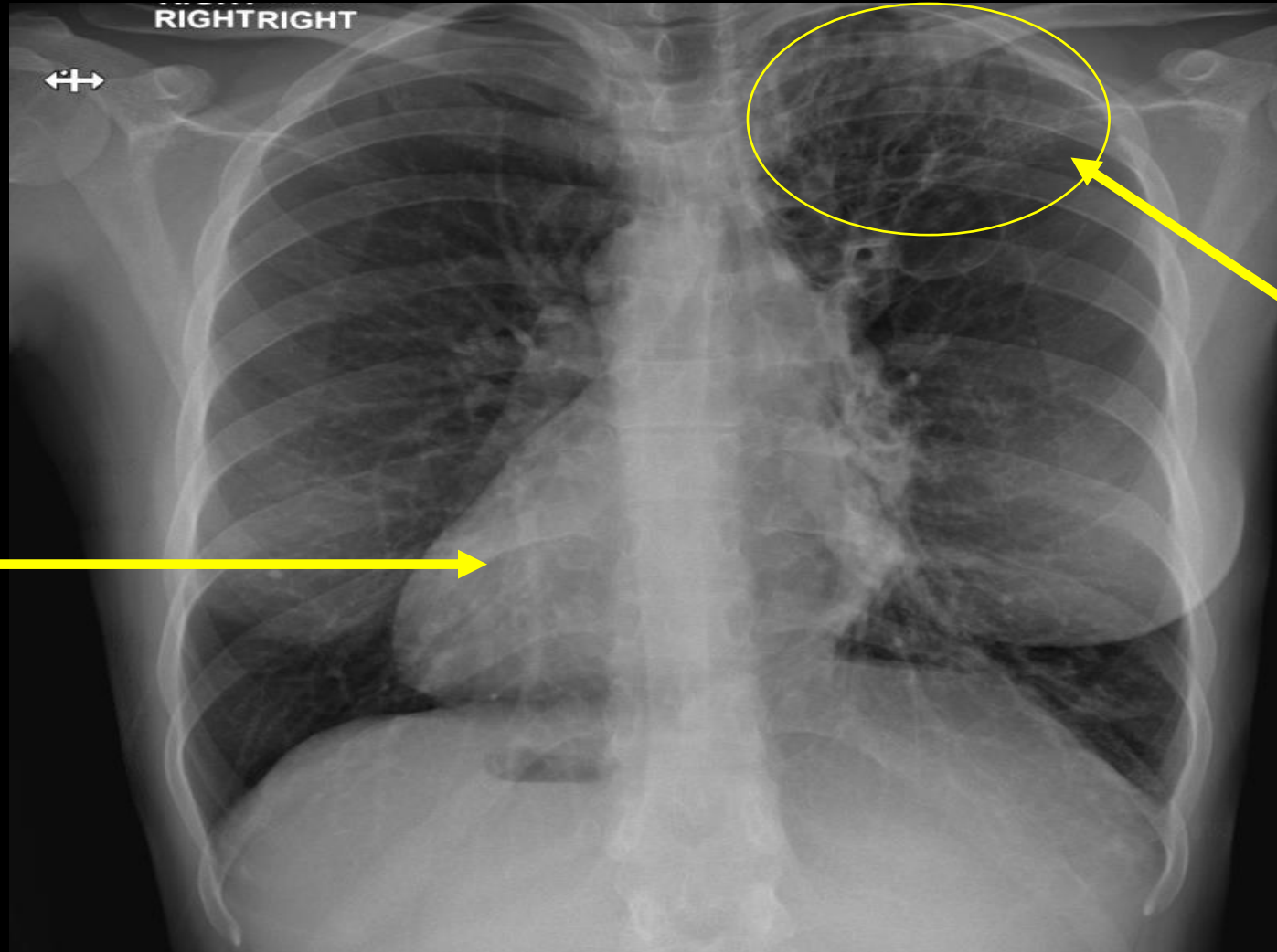
This imaging modality was ordered by the ER physician



CXR Findings (unlabeled)



CXR Findings: (labeled)



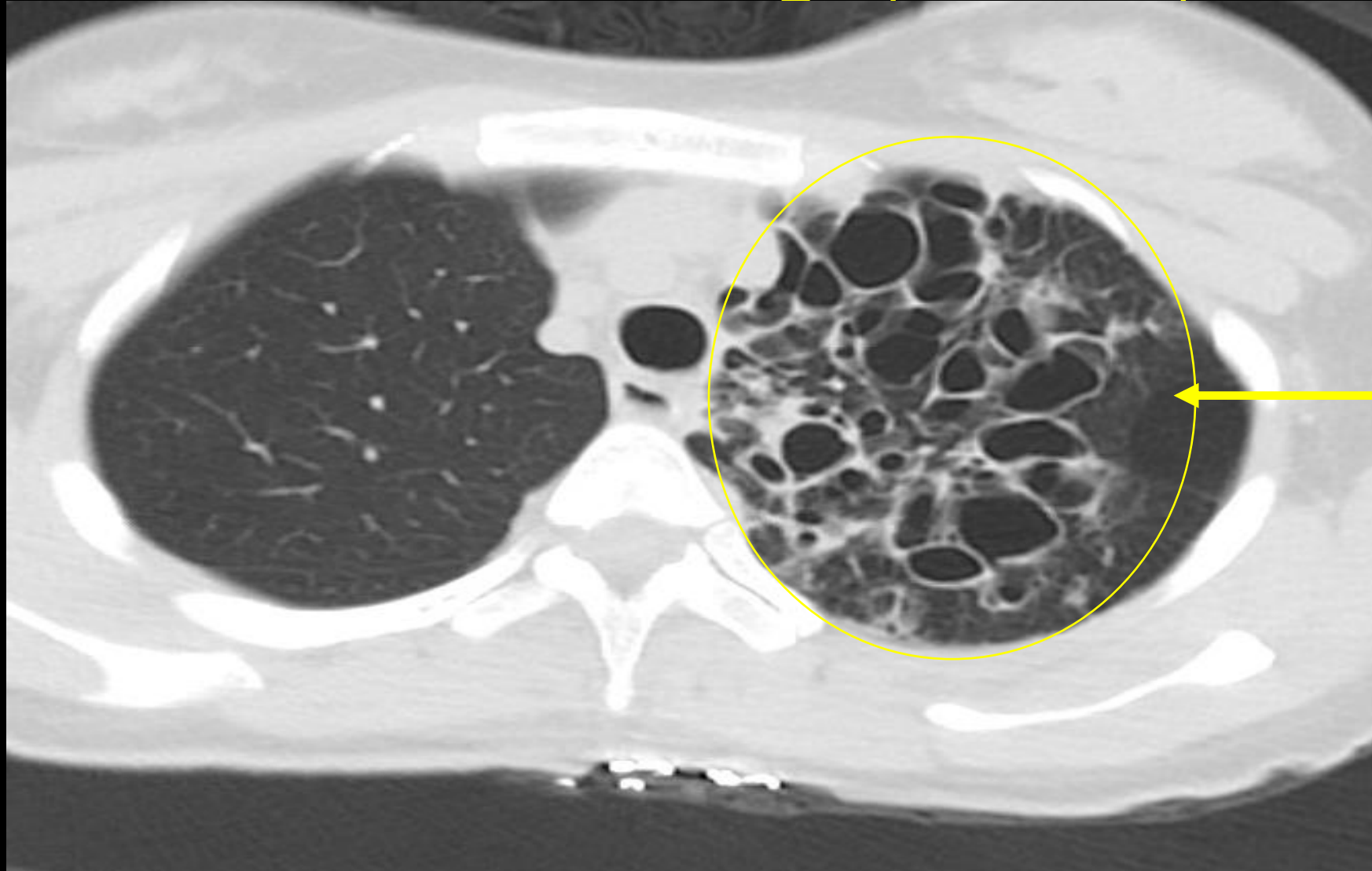
Dextrocardia

Bronchiectasis
with
surrounding
fibrosis

CT Chest Findings (unlabeled)



CT Chest Findings (labeled)



Severe
bronchiectasis
with
surrounding
fibrosis

Final Dx:

Bronchiectasis secondary to Kartagener Syndrome

Case Discussion

- Patho-Physiology: Gene mutation of dynein protein that leads to dysmotility of cilia
- Clinical Presentation
 - 50% present with dextrocardia
 - Frequent sinopulmonary infections due to decreased muco-ciliary clearance
 - Bronchiectasis
 - Infertility in men and women due to immotile sperm and decreased ciliary activity in fallopian tubes
- Diagnosis can be made through
 - Low levels of nasal nitrous oxide
 - Bronchoscopy + Electron microscope to visualize dysmotility of cilia
 - Genetic testing of DNAL1 gene Axonemal Dynein Intermediate chain gene

References:

- ACR Appropriateness Criteria® Evaluation of the Chronic Dyspnea without clear Etiology. Available at <https://acsearch.acr.org/docs/69448/Narrative/>. Accessed July 23, 2022.
- Geremek, M., Schoenmaker, F., Zietkiewicz, E. *et al.* Sequence analysis of 21 genes located in the Kartagener syndrome linkage region on chromosome 15q. *Eur J Hum Genet* **16**, 688–695 (2008). <https://doi.org/10.1038/ejhg.2008.5>
- Leigh MW, Pittman JE, Carson JL, Ferkol TW, Dell SD, Davis SD, Knowles MR, Zariwala MA. Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome. *Genet Med*. 2009 Jul;11(7):473-87. doi: 10.1097/GIM.0b013e3181a53562. PMID: 19606528; PMCID: PMC3739704.
- Skeik N, Jabr FI. Kartagener syndrome. *Int J Gen Med*. 2011 Jan 12;4:41-3. doi: 10.2147/IJGM.S16181. PMID: 21403791; PMCID: PMC3056330.