

# Pulmonology Case Presentation

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UNC  
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26-year-old female presenting  
with a lifelong history of  
productive cough and sinusitis



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## Patient history

A 26-year-old Hispanic female presented to clinic with a lifelong history of cough with copious production of purulent sputum, recurrent sinusitis, and occasional hemoptysis and chest pain. Additionally, she and her partner have been unable to conceive despite approximately seven years of unprotected intercourse; neither of them has been worked up for infertility. She has previously been diagnosed with “asthma”. She has no history of pancreatitis or liver disease. She has no known family history of specific pulmonary disease, although she does note that her older sister died of pneumonia as an infant. She has never smoked, although her husband smokes and her mother smoked when she was young.

# Imaging studies obtained

- What studies should be ordered for this patient?

# Imaging studies obtained

- Chest x-ray PA and lateral
- Chest CT without contrast

Last review date: 2012

## American College of Radiology ACR Appropriateness Criteria®

**Clinical Condition:** Chronic Dyspnea — Suspected Pulmonary Origin

**Variant 1:** Any age.

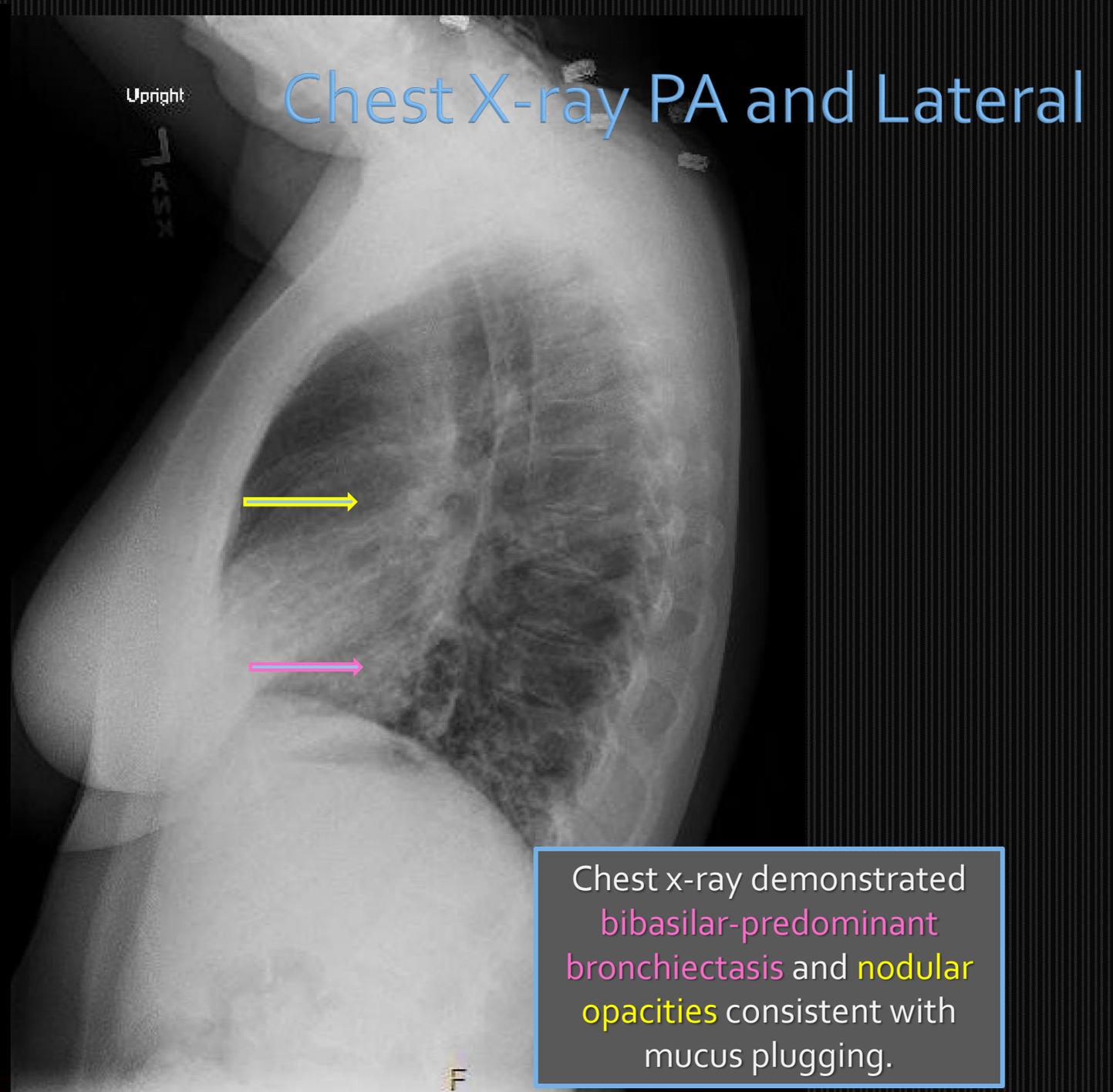
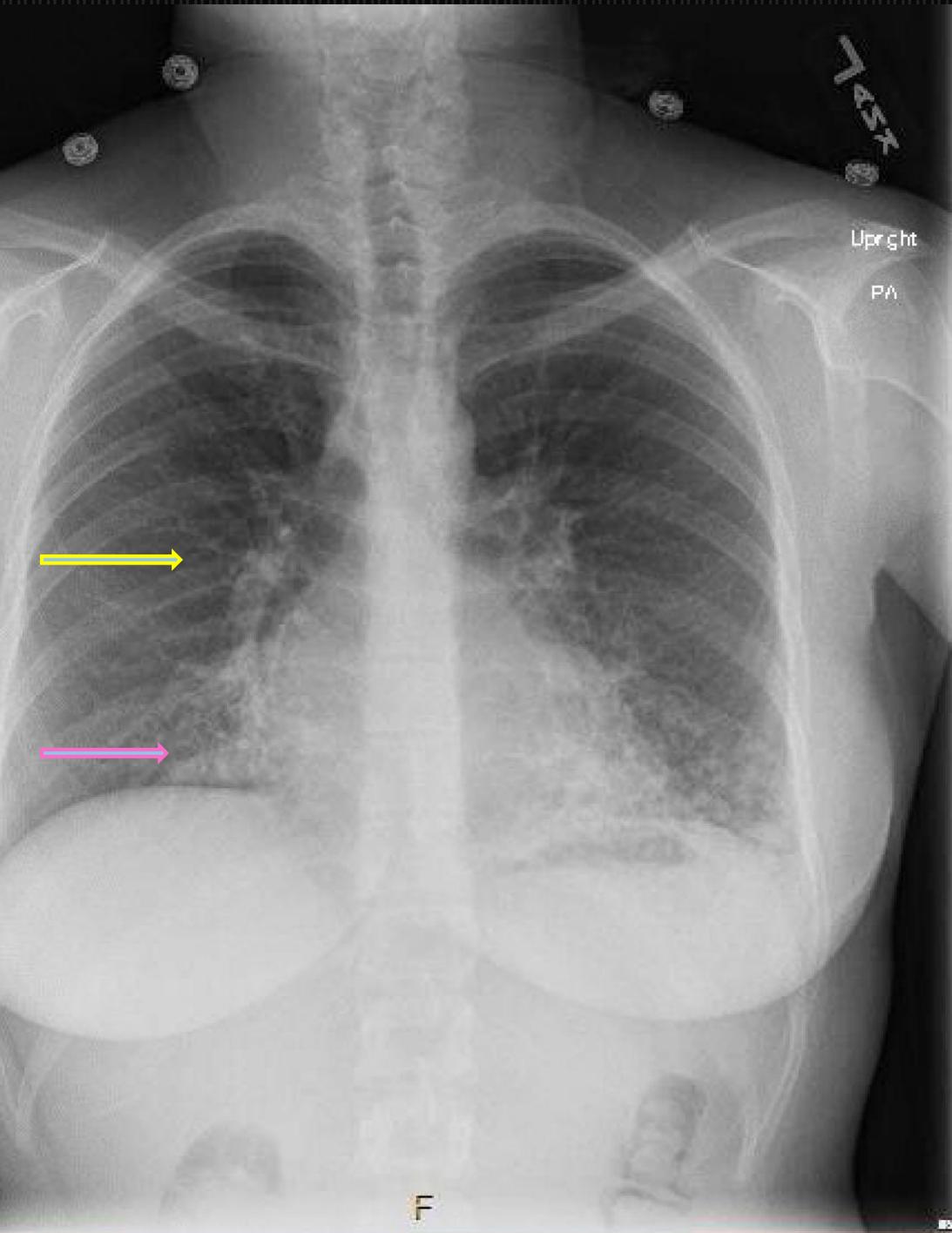
Radiologic Procedure	Rating	Comments	RRL*
X-ray chest	9	A negative chest radiograph does not exclude diffuse disease.	☼
<b>Rating Scale:</b> 1,2,3 Usually not appropriate; 4,5,6 May be appropriate; 7,8,9 Usually appropriate			<b>*Relative Radiation Level</b>

**Variant 2:** Any age, nonrevealing or nondiagnostic clinical, standard radiography, and laboratory studies.

Radiologic Procedure	Rating	Comments	RRL*
CT chest without IV contrast	9	In the setting of chronic dyspnea, the most appropriate imaging study is a thin-section high-resolution chest CT with prone imaging when appropriate. In patients with obstructive or mixed PFTs, the inclusion of expiratory imaging is important to evaluate air trapping and possible tracheobronchomalacia.	☼☼☼
CT chest with IV contrast	5	Usually not indicated unless suspect mediastinal or hilar adenopathy or fibrosing mediastinitis as cause for dyspnea. If a patient has dyspnea not clearly of pulmonary origin, other entities such as chronic or acute pulmonary embolism may need to be excluded. In that setting, a chest CTA is appropriate. See the ACR Appropriateness Criteria® topic on " <a href="#">Acute Chest Pain — Suspected Pulmonary Embolism.</a> "	☼☼☼
MRI chest without IV contrast	2	May be useful in characterizing pleural and chest wall masses, but its use in diffuse lung disease is currently limited to research.	○
MRI chest without and with IV contrast	2	May be useful in characterizing pleural and chest wall masses, but its use in diffuse lung disease is currently limited to research.	○
CT chest without and with IV contrast	1		☼☼☼
FDG-PET/CT chest	1	May be useful in characterizing pleural and chest wall masses, but its use in diffuse lung disease is currently limited to research.	☼☼☼☼

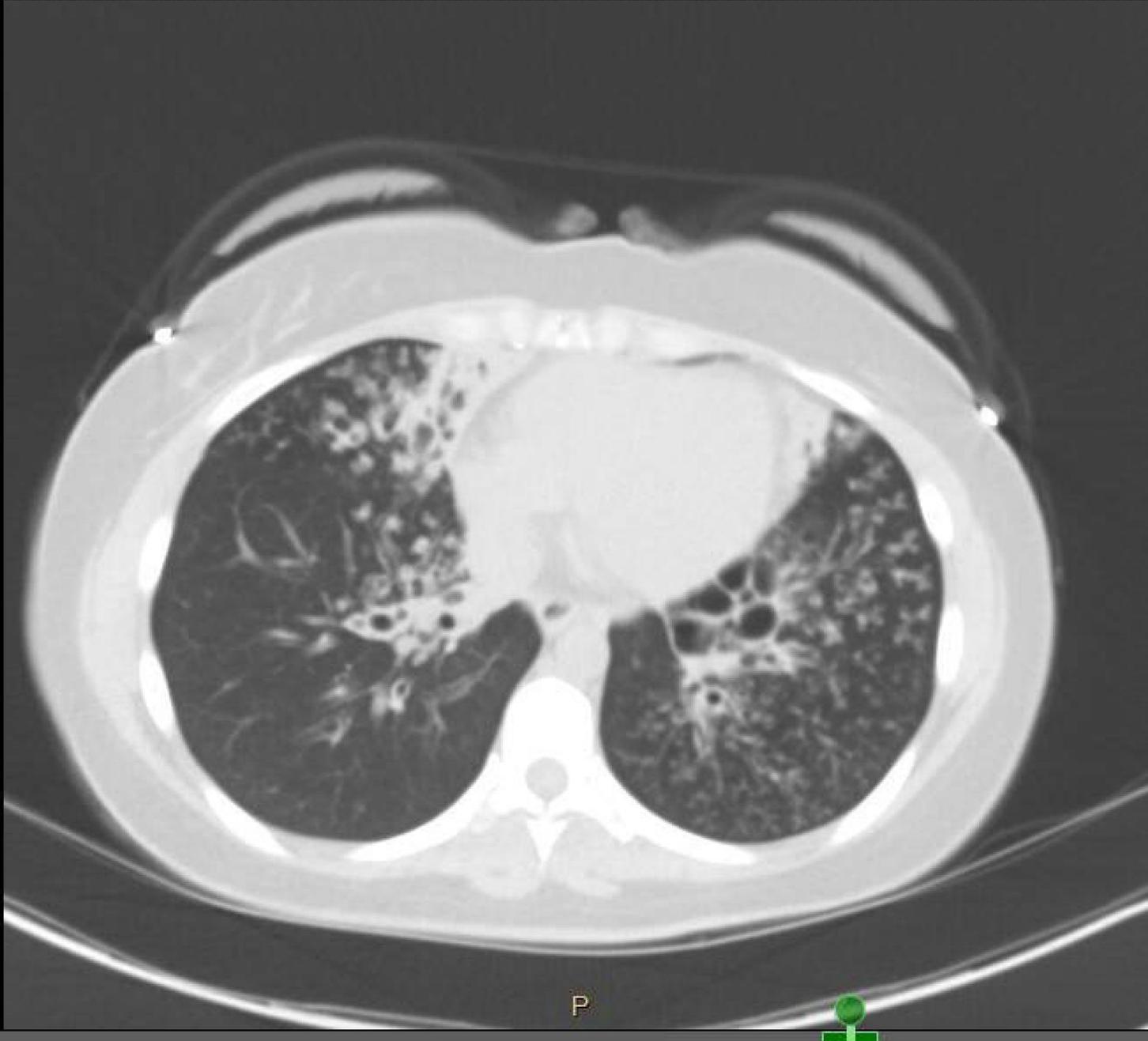
**Rating Scale:** 1,2,3 Usually not appropriate; 4,5,6 May be appropriate; 7,8,9 Usually appropriate

**\*Relative Radiation Level**



Chest x-ray demonstrated bibasilar-predominant bronchiectasis and nodular opacities consistent with mucus plugging.

## Chest CT without contrast



Chest CT without contrast demonstrated moderate to severe bronchiectasis within the right middle and lower lobes and left lower lobe, with bronchiectatic segments demonstrating mucus plugging. Within regions of bronchiectasis, there are associated centrilobular groundglass nodules in a tree-in-bud pattern.

# Diagnosis

Given her **lifelong** history of pulmonary symptoms **including hemoptysis** as well as **recurrent sinusitis**, possible **subfertility/infertility**, possible **family history**, and **imaging findings of bronchiectasis with a lower lobe predominance**, **primary ciliary dyskinesia** was highly suspected. She was found to have **low nasal nitric oxide**, consistent with this diagnosis. The diagnosis of primary ciliary dyskinesia was ultimately confirmed via **genetic testing**<sup>1,4</sup>.

(Cystic fibrosis, while less likely, was also ruled out with negative CFTR mutation analysis and normal sweat test.)

# Patient treatment and outcome

- Sputum culture grew fully-susceptible *Pseudomonas aeruginosa*
- Admitted to the hospital for IV antibiotic treatment with ceftazadime + tobramycin, as well as education and airway clearance with hypertonic saline + bronchodilators
- Continued on two weeks of IV antibiotics via PICC line following hospital discharge
- At clinic follow-up, azithromycin prophylaxis was added given that several sputum samples had proven stain and culture negative for acid-fast bacilli; continued airway clearance was also encouraged
- Over the past two years, has had periodic exacerbations treated with antibiotics but overall, has done well

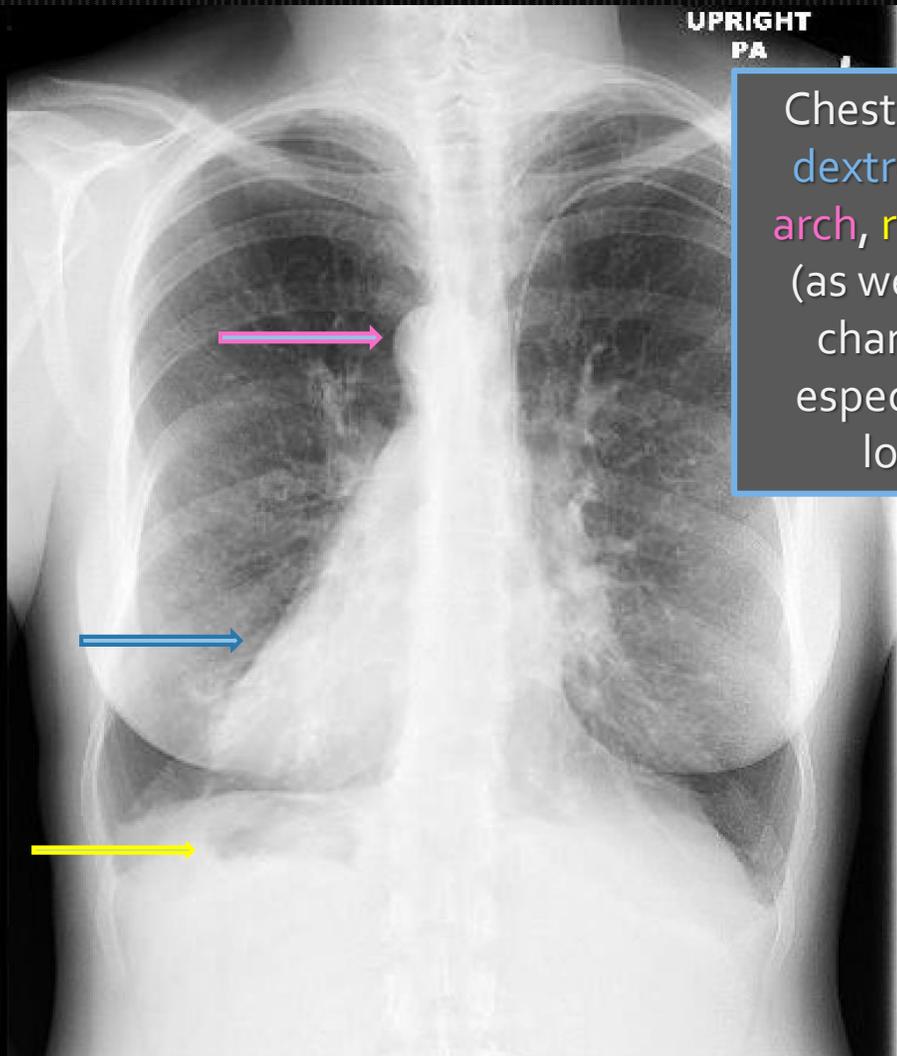
# Discussion: Primary Ciliary Dyskinesia

- Characterized by congenital impairment of mucociliary clearance due to defective ciliary structure and function in the airway epithelia
- Inherited as an autosomal recessive disease
- Highly heterogeneous disorder with a variety of clinical manifestations, including:
  - Neonatal respiratory distress
  - Chronic, daily productive cough from birth
  - Recurrent otitis media with frequent hearing loss
  - Recurrent sinusitis
  - Infertility/subfertility (immotile spermatozoa in men; impaired ciliary function in the fallopian tubes in women)<sup>1,4</sup>

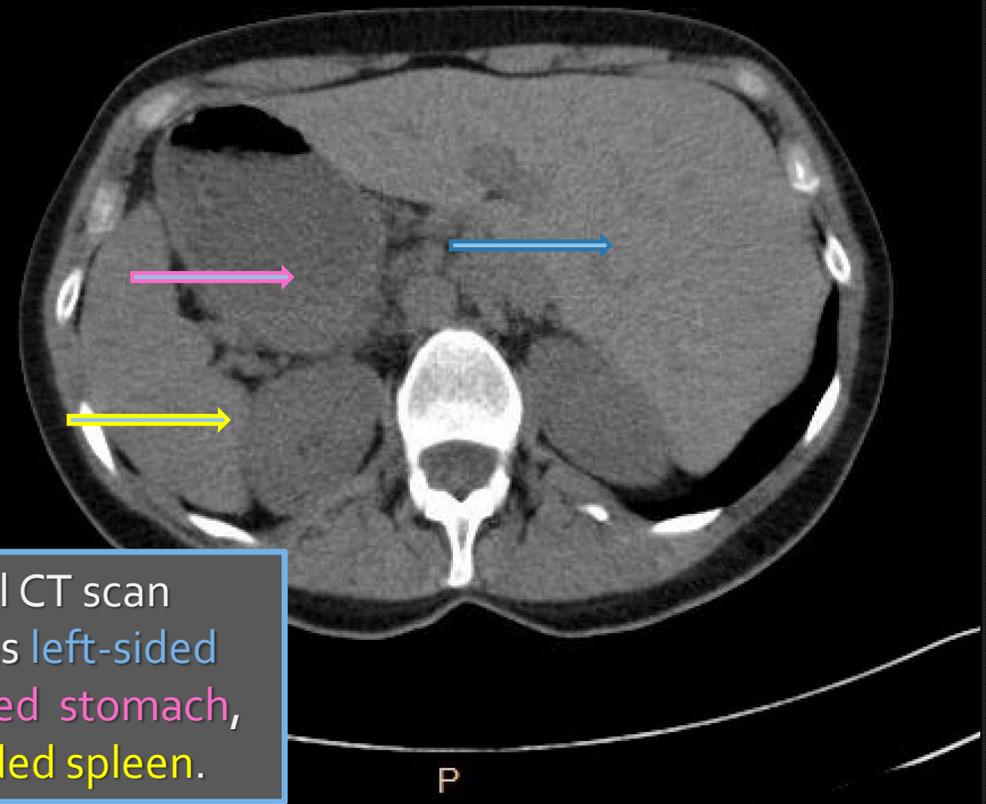
# Discussion: Primary Ciliary Dyskinesia

- Because the embryonic nodal cilia are also defective, approximately 50% of affected patients have situs inversus
- **Kartagener's syndrome** refers to the combination of situs inversus, chronic sinusitis, and bronchiectasis<sup>2,3,4</sup>

# Companion Case: Primary Ciliary Dyskinesia *with* Situs Inversus



Chest x-ray demonstrates dextrocardia, right aortic arch, right stomach bubble (as well as bronchiectatic changes in both lungs, especially in the mid and lower lung fields).



Abdominal CT scan demonstrates left-sided liver, right-sided stomach, and right-sided spleen.

# Imaging Pearls: Primary Ciliary Dyskinesia

- Chest x-ray is fairly nonspecific and may be normal, but can show **linear atelectasis, dilated and thickened airways, and irregular peripheral opacities**
- CT / HRCT is the preferred imaging modality and shows the **classic features of bronchiectasis** (bronchial dilation with increased bronchoarterial ratio, lack of airway tapering, bronchial wall thickening, mucus plugging) with a **middle/lower lobe predominance**
- **Situs inversus** is commonly present and can be a helpful clue in making the diagnosis
- Chest x-ray can be used in **follow-up** once a diagnosis has been established, especially given that this is a chronic illness requiring lifelong surveillance<sup>2,3</sup>

# References

- 1: Knowles MR, Daniels LA, Davis SD, et al. Primary ciliary dyskinesia. Recent advances in diagnostics, genetics, and characterization of clinical disease. *Am J Respir Crit Care Med* 2013; 188: 913.
- 2: Miller WT Jr, Panosian JS. Causes and imaging patterns of tree-in-bud opacities. *Chest* 2013; 144:1883.
- 3: Milliron B, Henry TS, Veeraraghavan S, Little BP. Bronchiectasis: Mechanisms and Imaging Clues of Associated Common and Uncommon Diseases. *Radiographics* 2015; 35:1011.
- 4: Noone PG, Leigh MW, Sannuti A, et al. Primary ciliary dyskinesia: diagnostic and phenotypic features. *Am J Respir Crit Care Med* 2004; 169:459.
- 5: Acsearch.acr.org. . (2018). Appropriateness Criteria. [online] Available at: <https://acsearch.acr.org/list> [Accessed 15 June 2018].

*Created in collaboration with Dr. Peadar Noone Professor of Medicine UNC School of Medicine*