

Learning Objectives

- Recognize the key signs, symptoms, and medical history that can distinguish primary ciliary dyskinesia from other respiratory illnesses
- Identify the criteria necessary for a diagnosis of asthma

Case Presentation

This is a 26-year-old woman, never smoker

- CC: wheeze, cough and dyspnea
- PMH: chronic sinusitis, asthma, **recurrent pneumonia, sinusitis, and otitis media** throughout childhood
- PSH: sinus surgery, **tympanostomy**
- Medications: albuterol (without relief)

Her tests revealed an obstructive lung process, with an FEV1/FVC ratio of **less than 70%** and no response to bronchodilators.

CT imaging showed bronchiectasis.

To test for PCD, nasal nitric oxide was measured, which was **less than 5 ppb**.

PCD and asthma present with similar symptoms. However, the crucial distinction is that asthma demonstrates reversible obstructive pathophysiology, whereas PCD does not.

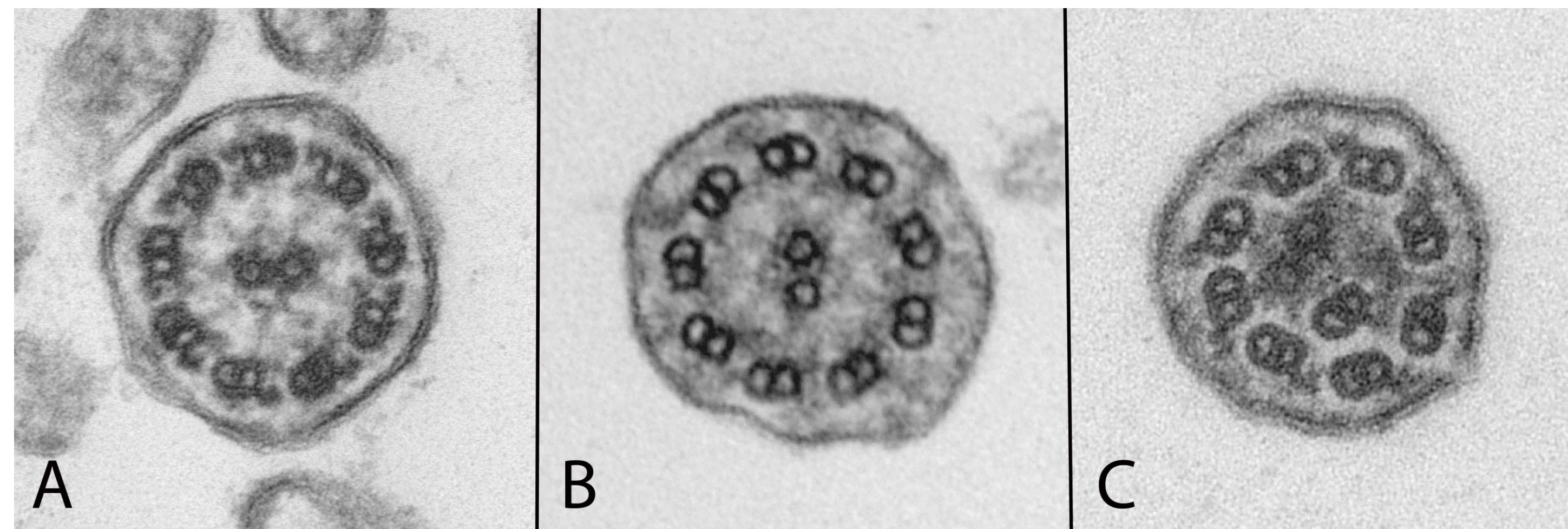


Figure 1. Electron microscopy (EM) of cilia comparing normal cilia structure with two PCD phenotypes. **A)** Normal; expected cilia in a healthy patient. **B)** Dynein deficiency; a genetic mutation resulting in an absence of cytoskeletal protein that confer cilia motility. **C)** Microtubular disarray; abnormal formation of microtubules in the cilia.

References

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Discussion

- Our patient's history of recurrent infections raised the possibility of PCD, which was supported with nasal nitric oxide testing.
- PCD should be considered in patients who do not respond to asthma therapies, those who have irreversible airflow limitations, and those with a history consistent with recurrent sinus or respiratory infections.

Take-Away Points

- Many respiratory diseases mimic asthma symptoms. Additional investigation is warranted in individuals who do not respond to traditional asthma therapies.
- Combined histories of recurrent pneumonia, otitis media, and/or sinusitis should raise concern for PCD.