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Images in Hospital Medicine

Primary Ciliary Dyskinesia

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Abstract

Primary ciliary dyskinesia, also known as immotile-ciliary syndrome causes impaired mucociliary clearance most notably in the upper and lower airways. We describe a middle aged man with a history of primary ciliary dyskinesia who presented to the hospital following a witnessed seizure at home.

A 53-year-old male presented to the emergency department (ED) following a witnessed seizure at home. His medical history included primary ciliary dyskinesia, pulmonary mycobacterial avium complex infection, unhealthy alcohol use, seizure disorder, and atrial fibrillation. He had quit smoking following a 40-pack year history. While in the ED he developed non-bloody, non-bilious vomiting, and was admitted for further management. On examination, his vital signs were normal. Neither cognitive impairment nor focal neurological deficits were noted. Complete blood count and comprehensive metabolic panel were unremarkable. Non-contrasted computed tomography (CT) of the patient's head noted sinus mucosal disease involving the bilateral ethmoid air cells and maxillary sinuses, along with bilateral mastoid effusions (**Figure 1**). Chest radiography showed dextrocardia and bibasilar opacities without focal consolidation. Contrasted CT scanning of abdomen and pelvis showed bibasilar bronchiectasis, diffuse centrilobular opacities within the lung bases (**Figure 2**) and situs ambiguous (with reversed positioning of the abdominal aorta and inferior vena cava) (**Figure 3**). The patient's seizure was suspected to be related to alcohol use lowering seizure threshold. Nausea and vomiting resolved with supportive care and was attributed to alcohol induced gastritis. The patient was ultimately discharged home following supportive care.

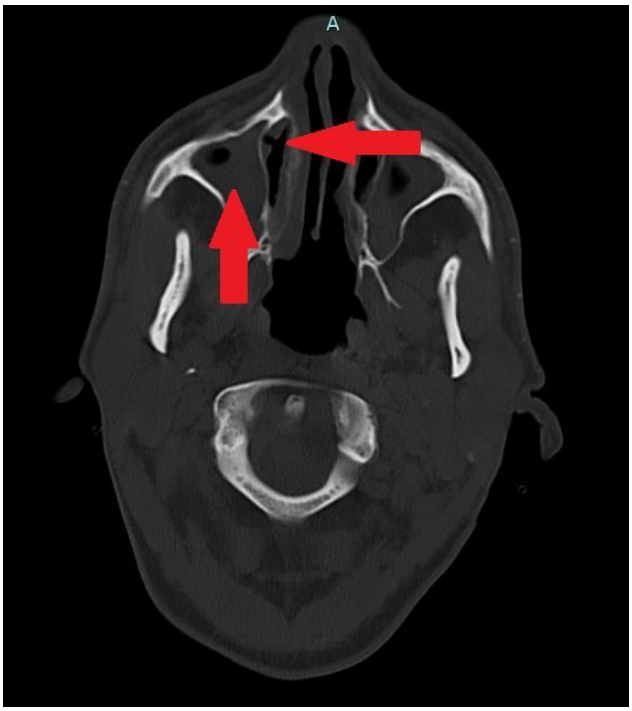

Figure 1. Bilateral ethmoid and maxillary sinus mucosal disease

Primary ciliary dyskinesia (PCD), also known as immotile-ciliary syndrome, is a genetically and clinically heterogenous disorder which follows an autosomal recessive transmission pattern.[1] Ciliary structure and/or function are affected, leading to impaired mucociliary clearance most notably in the upper (**Figure 1**) and lower airways (**Figure 2**). Resultant clinical manifestations in a cohort of subjects with confirmed PCD included chronic rhinitis/sinusitis (100%), recurrent otitis media (95%), neonatal respiratory symptoms (73%), and situs inversus (55%).[2] There is no single "gold standard test" for PCD. Confirmatory tests include visualization of cilia by video or electron microscopy, and genetic testing for common mutations.[3] Late diagnosis may be associated with decline in lung function and colonization with *Pseudomonas aeruginosa*.[4] Prognosis in patients with PCD likely relates to the specific genetic abnormality/structural defect underlying the condition.[5]

Patients with PCD are best managed at multi-specialty centers. However, internists play an important role in identifying patients for testing, early diagnosis, and ap-

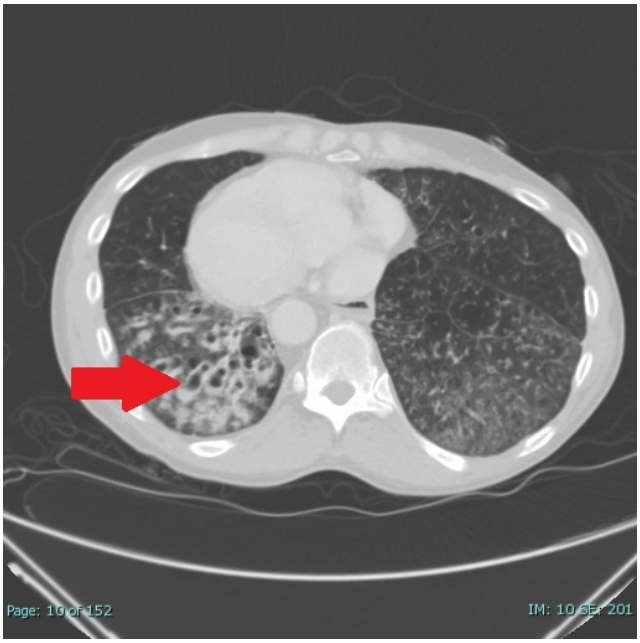

Figure 2. Right lower lobe bronchiectasis

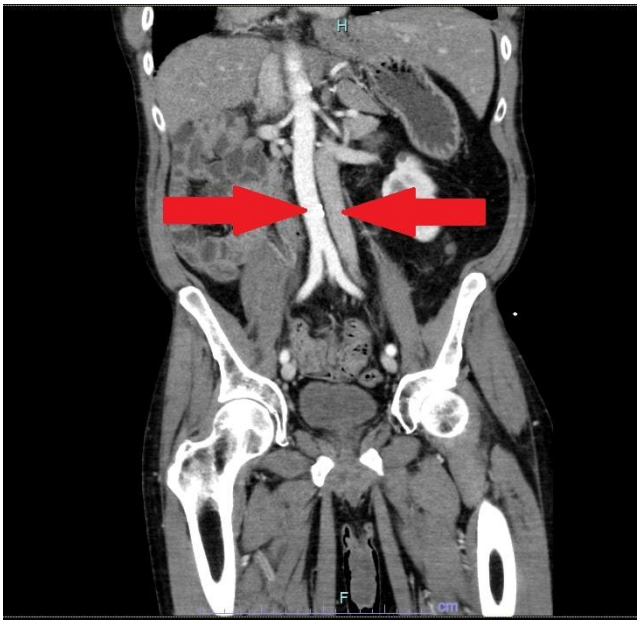

Figure 3. Situs ambiguous

propriate referrals. Smoking cessation is even more imperative in those with PCD so as to avoid acceleration of lung disease. Internists are also critical in the stewardship of appropriate antimicrobial use in PCD. Diagnosis of sinusitis and pneumonia rely on identifying a compatible clinical syndrome in addition to radiographic findings to appropriately initiate antimicrobial treatment. Upper and lower respiratory tract anatomical changes are hallmarks of PCD and are likely to be seen chronically -and thus the history and physical exam become critical. When patients with PCD develop lower respiratory tract infections early and prolonged antimicrobial therapy may be indicated.[6] PCD is a rare and interesting diagnosis, and when diagnosed early and treated appropriately, patient outcomes may be improved.

Author Contributions

All authors have reviewed the final manuscript prior to submission. All the authors have contributed significantly to the manuscript, per the International Committee of Medical Journal Editors criteria of authorship.

- Substantial contributions to the conception or design of the work; or the acquisition, analysis, or interpretation of data for the work; AND
- Drafting the work or revising it critically for important intellectual content; AND
- Final approval of the version to be published; AND
- Agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Disclosures/Conflicts of interest

The authors have no conflicts of interest to disclose.

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REFERENCES

1. Ciancio N, de Santi MM, Campisi R, Amato L, Di Martino G, Di Maria G. Kartagener's syndrome: review of a case series. *Multidiscip Respir Med*. 2015;10(1):18. doi:10.1186/s40248-015-0015-2

2. Noone PG, Leigh MW, Sannuti A, et al. Primary ciliary dyskinesia: diagnostic and phenotypic features. *Am J Respir Crit Care Med*. 2004;169(4):459-467. doi:10.1164/rccm.200303-365oc

3. Lucas JS, Burgess A, Mitchison HM, et al. Diagnosis and management of primary ciliary dyskinesia. *Archives of Disease in Childhood*. 2014;99(9):850-856. doi:10.1136/archdischild-2013-304831

4. Shah A, Shoemark A, MacNeill SJ, et al. A longitudinal study characterising a large adult primary ciliary dyskinesia population. *Eur Respir J*. 2016;48(2):441-450. doi:10.1183/13993003.00209-2016

5. Lucas JS, Barbato A, Collins SA, et al. European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia. *Eur Respir J*. 2017;49(1):1601090. doi:10.1183/13993003.01090-2016

6. Davis SD, Rosenfeld M, Lee HS, et al. Primary Ciliary Dyskinesia: Longitudinal Study of Lung Disease by Ultrastructure Defect and Genotype. *Am J Respir Crit Care Med*. 2019;199(2):190-198. doi:10.1164/rccm.201803-0548oc