Gastrointestinal Best Case
Clinical information

51-year-old white man presents with a vague one year history of mild dysphagia, anemia, weight loss, and abdominal pain.
Well-differentiated liposarcoma/Atypical lipomatous tumor in a giant fibrovascular polyp

Kathleen Eddy
Victoria General Hospital
Halifax, Nova Scotia, Canada
Honorable mention
Clinical information

26-year-old man with six days of acute bilateral pitting edema with labs showing long standing hypoalbuminemia.
Foveolar glands – mucin producing

Oxyntic glands – acid producing

Normal
Menetrier disease

Nicholas DiGeorge
Naval Medical Center Portsmouth
Portsmouth, Virginia
Pulmonary and Mediastinal
Best Case
• 47 year old female with a history of chronic cough with recurrent episodes of bronchitis and sinus infections
Sinus disease
Situs inversus and lower lobe bronchiectasis
Situs inversus and lower lobe bronchiectasis
Situs inversus and lower lobe bronchiectasis
bronchiectasis
Primary ciliary dyskinesia

Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome

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Abstract: Primary ciliary dyskinesia is a genetically heterogeneous disorder of male cilia. Most of the disease-causing mutations identified to date include amino acid substitutions or deletions in the axonemal dynein arm. A diagnosis is indicated in cases with recurrent respiratory infections, abnormal sperm motility, and situs inversus. Clinical genetics testing for primary ciliary dyskinesia is available in most centers. The diagnosis is established through a combination of genetic testing, immunochemistry, and electron microscopy. Imaging studies often show a characteristic pattern of situs inversus, sinus abnormalities, and bronchial variation. The diagnosis is confirmed by genetic testing, which can be performed on DNA samples obtained from peripheral blood lymphocytes. The diagnosis is important because it can help to identify individuals at risk for certain congenital anomalies and to provide guidelines for parental counseling.

KEY WORDS: primary ciliary dyskinesia, Kartagener syndrome, situs inversus, cilia

OVERVIEW

Primary ciliary dyskinesia (PCD) is a genetically heterogeneous, sporadically inherited recessive disorder characterized by cyclic dysfunction and abnormal motility of the cilia, which results in an array of clinical manifestations. Although PCD is a rare disease, it is important to recognize because it can lead to significant morbidity and mortality. The diagnosis is typically made in individuals with recurrent respiratory infections, abnormal sperm motility, and situs inversus. The diagnosis can be confirmed by genetic testing, which can be performed on DNA samples obtained from peripheral blood lymphocytes. The diagnosis is important because it can help to identify individuals at risk for certain congenital anomalies and to provide guidelines for parental counseling.

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REVIEW

Ciliary structure and function

Normal ultrastructure of cilia

Cilia and flagella are evolutionarily ancient organelles whose structure and function have been tightly conserved throughout life. They are essential for the proper function of many epithelial cells. Cilia are responsible for the movement of fluids over mucosal surfaces, the transport of secretions, and the sensation of touch. They are also involved in the regulation of cell metabolism and transport of fluids over mucosal surfaces, and they have recently been recognized to have a sensory function that modulates elements of development and cell function. Both

Genetics in Medicine Volume 11, Number 7, July 2009

DOI: 10.1089/gim.2008.0164
Cardiovascular Best Case
48 year old female with four-month history of dyspnea and non-productive cough.

Initial workup revealed mild tachycardia, elevated D-dimer, and CTA with multiple intravascular filling defects in the pulmonary arteries.

She was discharged on Lovenox, but returned 10 days later with significant worsening of symptoms.
CTA: large broad-based filling defect, right side of main PA extending into right PA. (Shaggy margins, possible peripheral enhancement?)
Pulmonary angiogram: cut-off of right main PA
Autopsy performed after cardiac arrest. Heterogeneous-appearing soft tissue fills main PA (arrows). Adjacent aortic lumen (Ao) is clear.
Lobulated soft tissue mass with frond-like margins (arrows) also invades the pulmonary valve (pv).
H&E stained tissue confirms sheets of tumor (arrows) coating pulmonary valve leaflets (pv).
Gross right lung shows central intravascular soft tissue (arrow) which was also found extending into distal lobar vessels.
H&E stained tissue shows dense field of pleomorphic spindle cells (arrows).
Pulmonary Artery Intimal Sarcoma

Jessica Chan, MD
University of Utah
Salt Lake City, Utah
Pediatric Best Case
A 14-year-old female presents with progressive obstructive airway disease as well as a history of repeated choking episodes since 18 months old as well as poor weight gain.
Esophageal Leiomyomatosis associated with Alport Syndrome

Brian Pogatchnik
University of Minnesota Medical Center
Minneapolis, MN
Breast Best Case
32-year-old male with a palpable lump and serosanguinous discharge from his left nipple occurring 8 weeks prior to presentation.
Mammogram and US
Histology
DCIS in a Papilloma with adjacent microinvasion in a Male

Karen Tran-Harding
University of Kentucky
Lexington, KY

Nice educational article in RadioGraphics on male breast disease
Nguyen C. May 2013
Clinical Information

- 49 year old male with PSA 8.0. Prostate MRI performed.
Neuroradiology Best Case
25-year-old male with progressive left nasal obstruction, enlarging mass of hard palate, increasing left facial swelling and numbness
Chondroblastic Osteosarcoma

Louis Skidmore, M.D.
University of Pittsburgh
Pittsburgh, Pennsylvania
Musculoskeletal Best Case
13 month old otherwise healthy male presented initially with slowly growing firm mass over 4 months along the ulnar aspect of the right ring finger.
SOFT TISSUE CHONDROMA

Dr. Matthew Burgess
Naval Medical Center
San Diego, CA
Honorable mention
62 year old African American male mass in his right axilla causing decreased shoulder range of motion without associated pain
MYXOFIBROSARCOMA

Dr. Trilochan Hiremath
University of Pittsburgh Medical Center
Pittsburgh, PA