Bronchiectasis Case Presentation

- Presents in September, 2017 with progressive dyspnea on exertion
  Currently walks 2 blocks. Could walk 1 mile a year earlier

- Cough, chronic sputum production.

- PMH: GERD, Hiatal hernia, Gastroparesis, LV diastolic dysfunction

- PMH: MAC Rx
  a) 1996 x 18 months
  b) 2002 x 13 weeks
  c) 2004 x 6 months
  d) 2015 x 8 months
  e) 2017 6 months to current (fevers, weight loss, cough and dyspnea)

- Pancreatic Insufficient. Failure to thrive as an infant.

- Chronic Sinusitis. Recurrent nasal polypectomy
Bronchiectasis Case Presentation

- **Physical Exam**
  - Thin. BMI = 18.5
  - Mild maxillary Tenderness
  - Chest: Decreased breath sounds with scattered rhonchi. Rare exp wheezing
  - No clubbing

- **Laboratory Data**
  - CBC: Normal
  - ESR: 110
  - CRP: 5.2
  - Chem20: Normal
  - Ig and IgG subclasses: Normal
  - IgE: 4

- **Microbiology Data**
  - AFB: MAC/MAI
  - Fungal: Aspergillus intermittently
  - Routine: *Pseudomonas aeruginosa*
    - *Stenotrophomonas maltophilia*
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- Relevant CF data
  - Sweat Chloride: 25meq/ml
  - Borderline nasal potential difference
  - CFTR genetics
    a) M470V/M470V
    b) IVS 5T allele
  - Fecal pancreatic elastase was 52 ug/g (nl > 200ug/g)
  - Vitamins E and A were normal
  - Vitamin D was low

- Relevant PCD data
  - EM cilia showed normal outer and inner dynein arms and questionable central apparatus.
  - Single DNAH5 variation
    p.F4392C: uncertain pathogenicity
  - Single DNAH11 variation
    p.D892V: uncertain pathogenicity
Bronchiectasis Case Presentation

Leading considerations for etiology of bronchiectasis

- Atypical Cystic Fibrosis
  Strong clinical phenotype but lacks convincing laboratory confirmation
- Chronic NTM (MAC/MAI) airway infection
- Chronic aspiration
- Atypical PCD
  PCD is recessive.
  Unclear if the carrier state is clearly normal

- “Multiple hit” concept