AMSER Case of the Month

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Malabsorption and Recurrent Respiratory Infections

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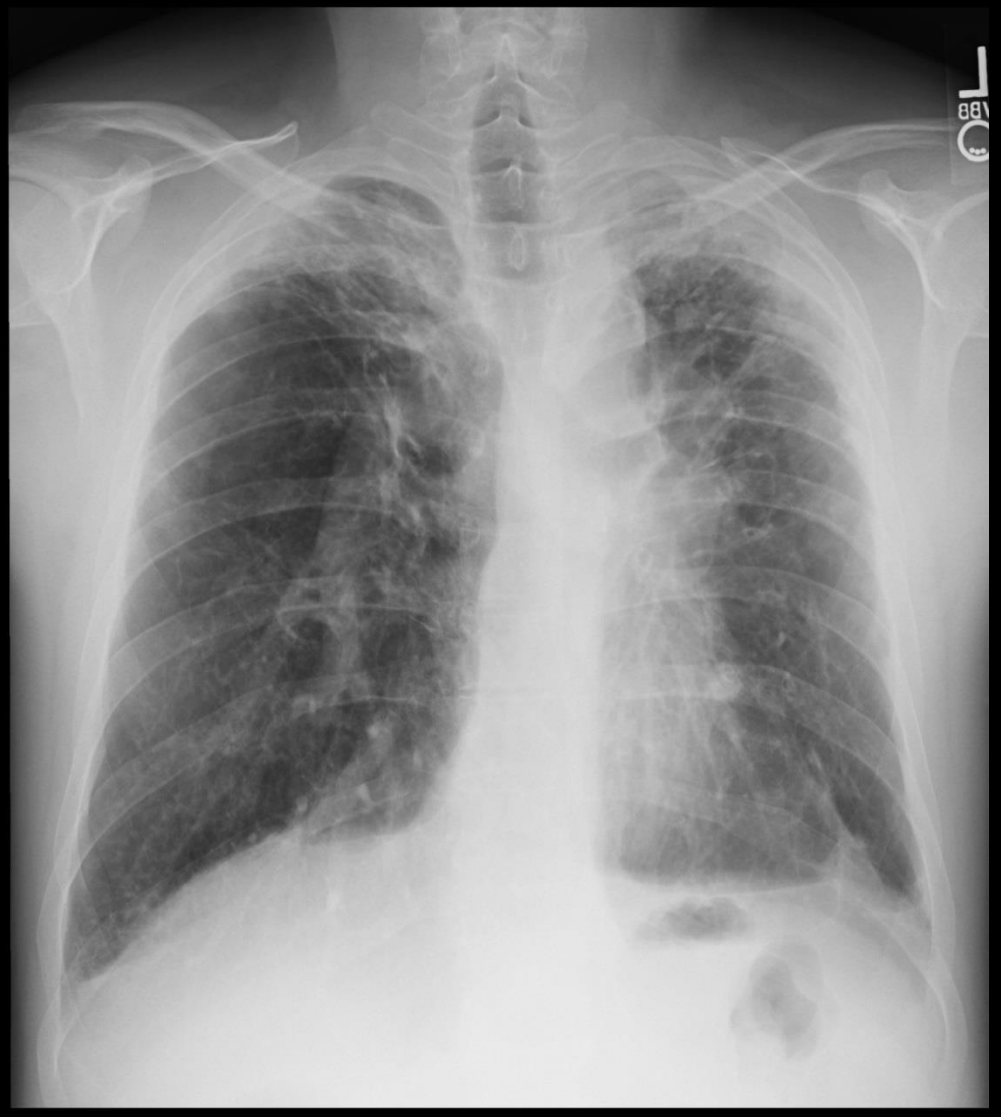
Clinical History

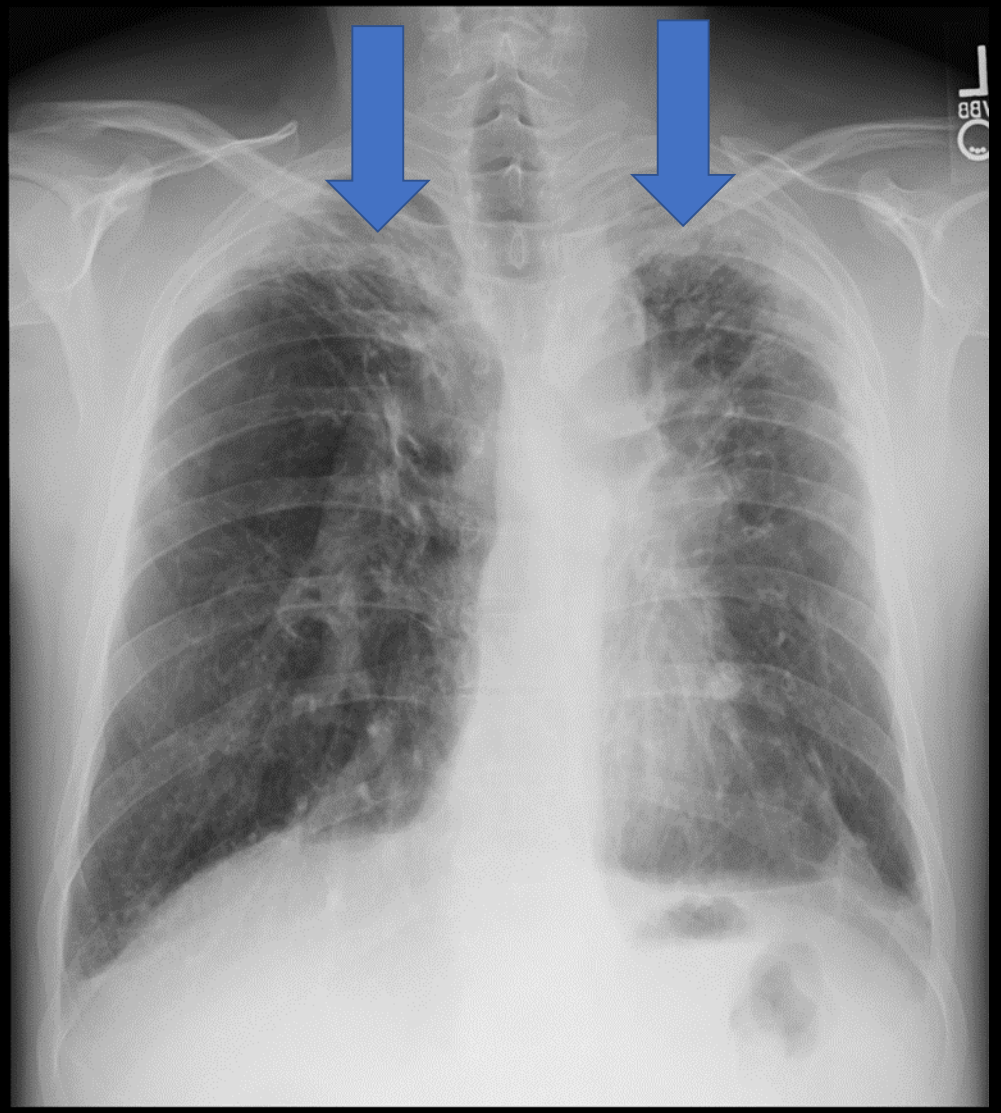
- This is a 44-year-old male presenting for a routine follow up to monitor chronic airway disease.
- This patient has a long-standing history of recurrent respiratory infections, malabsorption, and shortness of breath.

PA Chest Radiograph

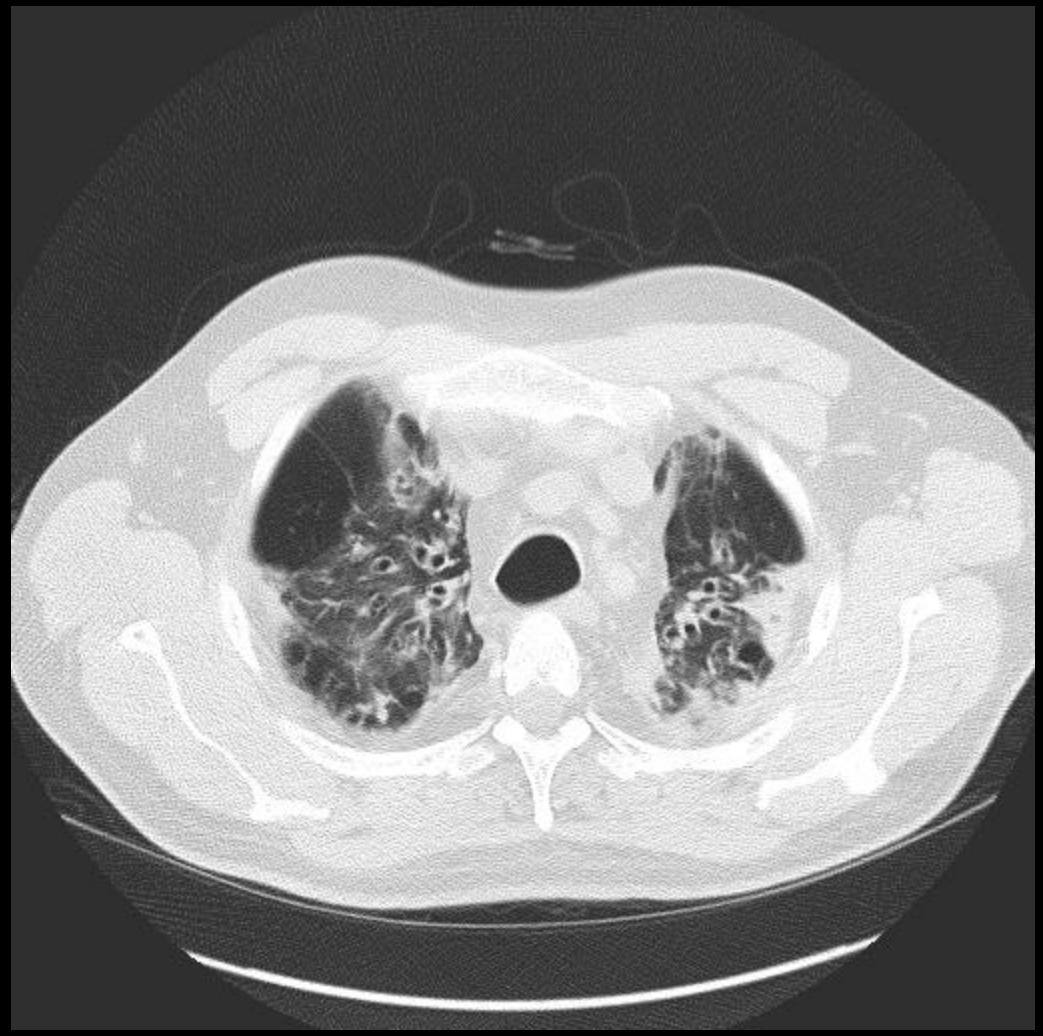


PA Chest Radiograph

Apical and perihilar predominant bronchiectasis, note the superior displacement of the transverse fissure and tenting of the diaphragmatic pleura

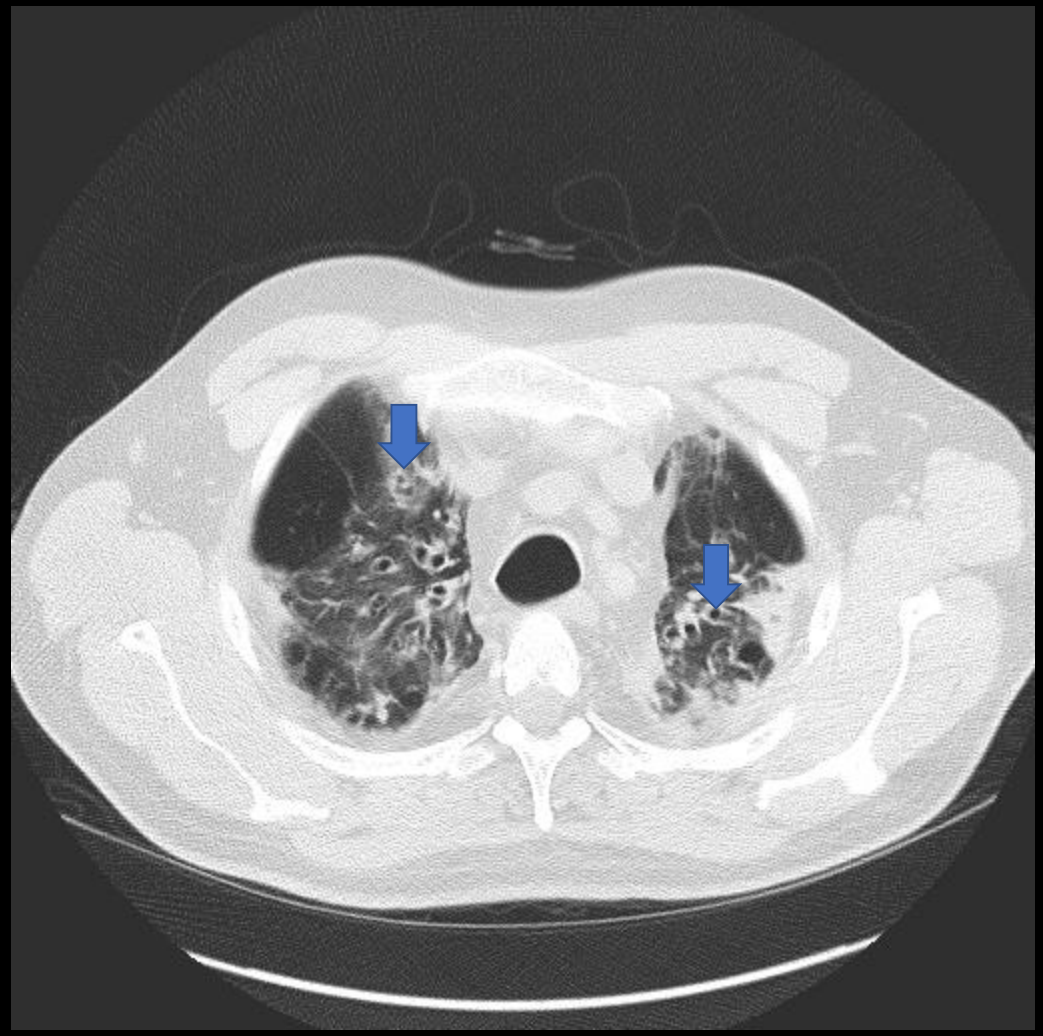


Axial Non-Contrast Chest CT

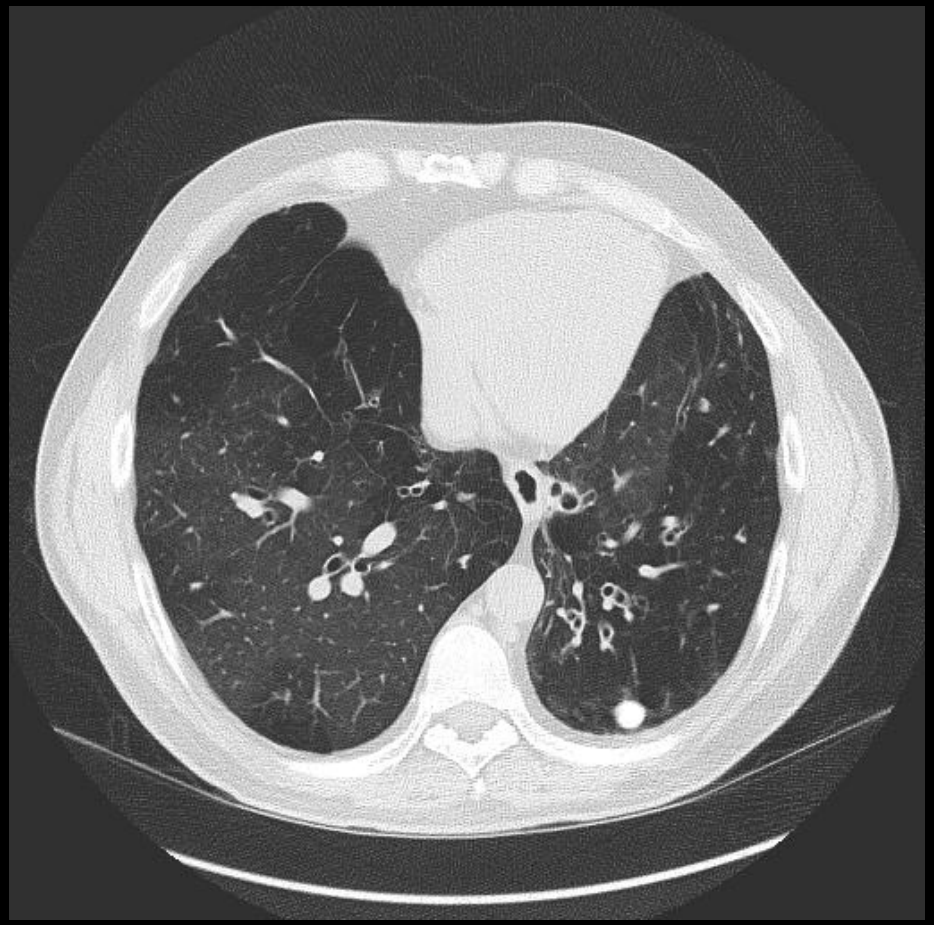


Axial Non-Contrast Chest CT

Diffuse bronchiectasis and bronchial wall thickening, circumferential pleural thickening, apical and perihilar fibroreticular scarring, bullous emphysematous changes

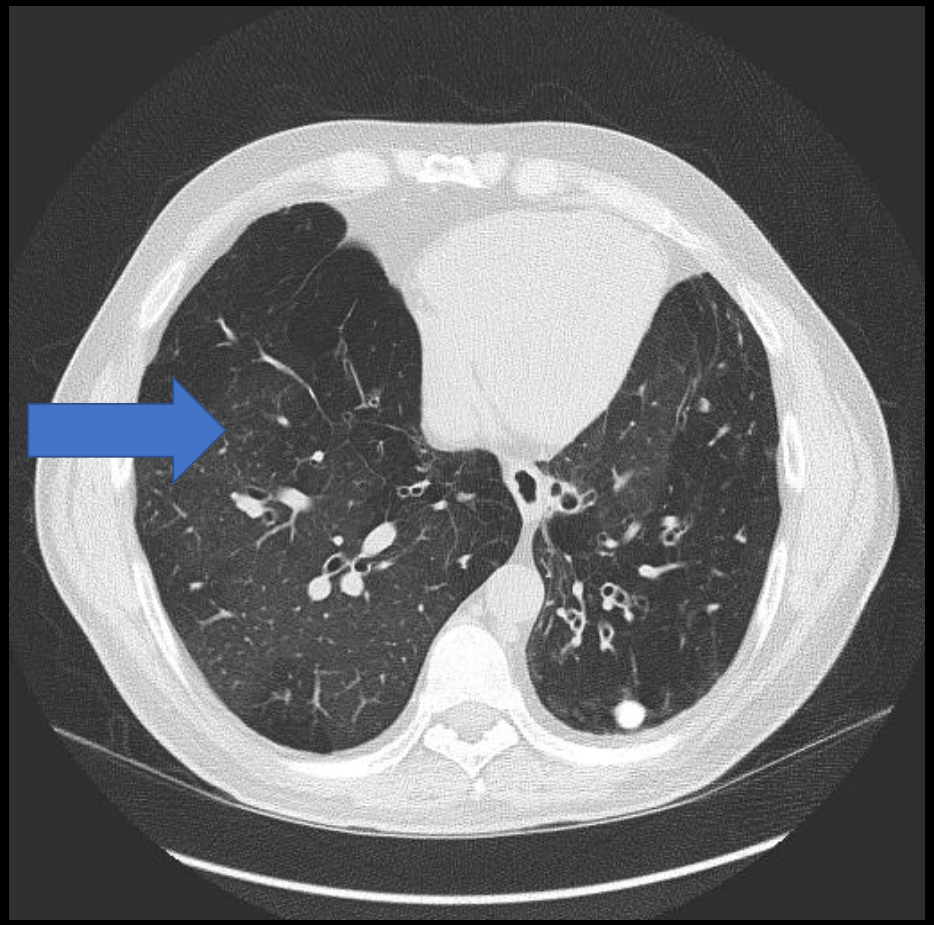


Axial Non-Contrast Chest CT

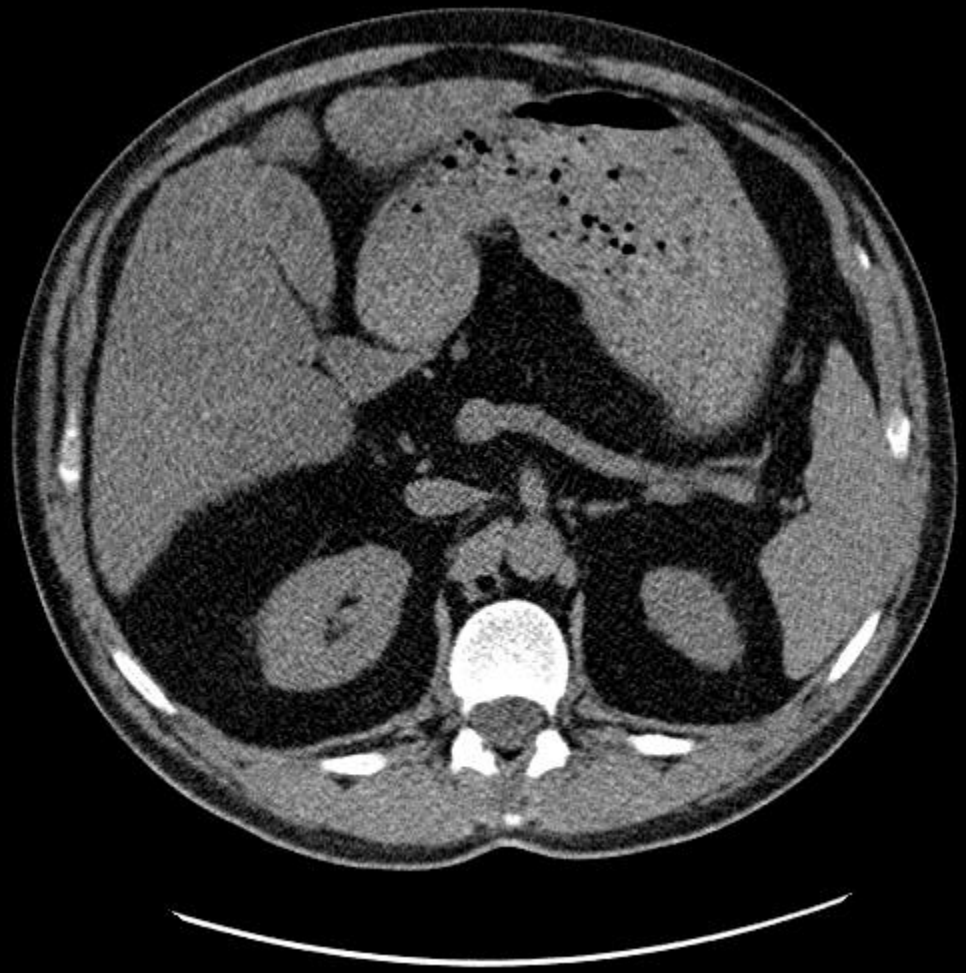


Axial Contrast Enhanced Chest CT

Mosaic attenuation pattern

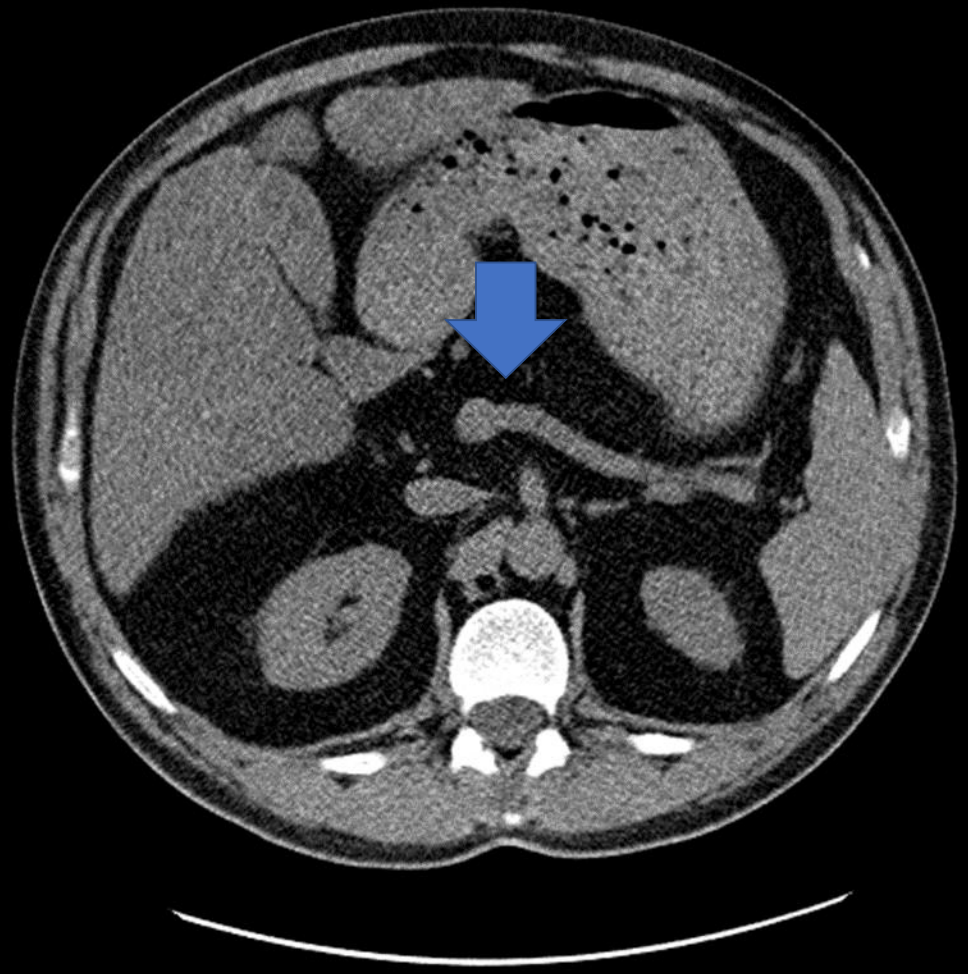


Axial Non-Contrast Abdominal CT



Axial Non-Contrast Abdominal CT

Fatty replacement of the pancreas, the splenic vein can clearly be seen in place of the pancreas



Differential Diagnosis

- Cystic Fibrosis
  - Recurrent sinopulmonary infections, pancreatic insufficiency
- Severe Combined Immunodeficiency
  - Recurrent sinopulmonary infections
- Primary Ciliary Dyskinesia
  - Recurrent sinopulmonary infections, situs inversus
- Scwachman-Diamond Syndrome
  - Pancreatic insufficiency, skeletal abnormalities, bone marrow dysfunction

Review of Cystic Fibrosis

- Cystic fibrosis (CF) is the most common lethal autosomal recessive disease in the white population, affecting 28,000 persons in the United States.
- Mutation of the CF trans-membrane conductance regulator gene (CFTR) on chromosome 7 results in production of abnormally viscous mucus and secretions in the lungs, gastrointestinal tract, pancreas, and hepatobiliary system.
- Inspissated mucus in each of these systems leads to the luminal obstruction, and thus the radiologic and clinical manifestations of this disease.
- Early detection and follow up of lung disease in CF is crucial to allow prompt treatment adaptation.

Workup

- North American Cystic Fibrosis Guidelines recommend yearly follow-up chest radiographic exam in addition to pulmonary function testing to track the progression of lung damage.
- The decision to perform routine imaging should be tailored to patient age and disease severity.
- CT has been shown to be more accurate than FEV1 and chest radiography in the early detection of clinically relevant pathologic changes, detecting complications, and monitoring treatment effects
  - Bronchiectasis, peri-bronchial thickening, mucus plugging, emphysema
  - Can then use a scoring system, such as the Bhalla scoring system

Treatment Options

- Pancreatic enzyme replacement
- CFTR modulators
- Airway clearance therapies
  - Inhaled DNase, inhaled hypertonic saline, chest physiotherapy
- Infection prevention
  - Vaccines, palivizumab, infection control measures
- Bronchodilators
  - Inhaled beta-2 agonists
- Anti-inflammatory therapy
  - Oral azithromycin, high dose ibuprofen, inhaled glucocorticoids

Summary

- Cystic Fibrosis is the most common lethal autosomal recessive disease in the white population.
- Annual monitoring with PFT's and chest imaging is recommended to track the progression of pulmonary disease and response to treatment in certain patients.
- There are many treatment options available for the disease that can be tailored based on radiologic and clinical manifestations.

References

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