Kartagener’s Syndrome

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let’s meet our patient..

- 75 y/o woman presents to the ED complaining of chest pain over the anterior right chest wall

  ➢ The pain does not radiate anywhere. She denies nausea, vomiting, palpitations, dizziness or lightheadedness

- Symptoms of upper respiratory tract infection: nasal congestion, productive cough, SOB, fever & chills

- PE: stable vital signs; crackles over both lung fields posteriorly; heart sounds were irregularly irregular
PMH

• atrial fibrillation

• recurrent pneumonia as a child → up until the age of 14

• recurrent upper respiratory tract infections (two episodes of bronchitis/year)

• no history of tobacco

• no children
Chest pain

Workflow:

- DDx

- Diagnostic Tests
ECG to rule out MI

Atrial fibrillation. Extreme right axis deviation. Poor R wave progression across the precordium → left-sided leads in patient with dextrocardia.
Chest x-ray to evaluate for infiltrate
Chest x-ray to evaluate for infiltrate

- Situs inversus:
  - Dextrocardia with cardiomegaly
  - right sided gastric bubble
  - right sided aorta
  - left sided liver

- Bronchiectasis within opacity in left middle lobe (arrows)

- no effusions, no pneumothorax

- patchy consolidation in left lower lobe (best seen on lat. view, prev. page)
  → left lower lobe pneumonia
CTA to rule out pulmonary embolism

Chronic air space disease with bilateral bronchiectasis and chronic scar tissue in the peribronchial region. Worse in the left middle lobe. Situs inversus totalis with dextrocardia. No evidence of pulmonary embolism.
Exercise MIBI to evaluate myocardial perfusion

- Tc-99m sestamibi
- Exercise protocol: Modified Gervino (14 min) %max of predicted HR: 101%
- Anatomical left ventricular cavity size is normal (situs inversus)
- Uniform tracer uptake throughout LV myocardium
- Normal myocardial perfusion
Kartagener’s Syndrome

Kartagener’s Syndrome:

- primary ciliary dyskinesia
- situs inversus

Clinical features:

- recurrent respiratory infections
- bronchiectasis
- sinusitis
- chronic otitis
- dextrocardia - situs inversus
- infertility
Kartagener’s Syndrome

• autosomal recessive inheritance (incidence 1:30,000)

• first described by Manes Kartagener, an internist in Zürich in 1936

• mutations in several genes, including chromosome 9, 5 and 7 → cause morphologic or functional abnormality of cilia

• much heterogeneity → wide range of defects in ciliary ultrastructure and motility, which impairs mucociliary clearance

• usually diagnosed during childhood

→ index patient was diagnosed at age 75 due to moderate course of disease
Primary ciliary dyskinesia

In Primary ciliary dyskinesia cilia are either unable to move, or they beat with a uncoordinated and inefficient motion.

- 50% of pt with PCD have situs inversus, which then is called Kartagener’s Syndrome
Cilia

- Function: to move fluid or mucous over their surface

- two central single tubules surrounded by 9 microtubule doublets

- from each doublet a pair of dynein arms extends to join neighboring microtubules

- radial spokes and nexin links help to stabilize the cilia’s structure
Defects

• Ultrastructural defects:
  - absence of proteins (dynein arms, radial spokes, central tubules)
  - abnormal structures (centriole with giant roots and double feet, cilia lacking all internal microtubular structures, cilia with twice the normal length)

• Functional defects:
  - hypo/hypomotility
  - disorientated arrangement and beat direction
Kartagener’s Syndrome - Radiologic findings

- Nose/Sinuses
  - swollen nasal mucosa, nasal polyps
  - mucosal thickening in sinuses, opacified sinus cavities, hypoplastic frontal sinuses
  - (impaired sense of smell)
Kartagener’s Syndrome - Radiologic findings

• Ears
  ‣ middle-ear effusion with retracted tympanic membrane
  ‣ recurrent otitis media → often require ventilation tubes
  ‣ (bilateral conductive hearing loss)

Transverse CT section through the epitympanum:
  • the mastoid air cells are opacified
    → thickened sclerotic septae
    → chronic otitis
  • suspicious cholesteatoma with erosion of the short process of the incus (arrow)
Kartagener’s Syndrome - Radiologic findings

- Lower respiratory tract
  - bronchiectasis (lower lobes!)
  - bronchial wall thickening
  - hyperinflation
  - chronic infection
  - atelectasis
  - obstructive lung disease
  - situs inversus
  - (chronic bronchitis, recurrent pneumonia)
Situs inversus

Laterality defects

Situs solitus (normal)

Lung
Heart
Liver
Spleen
Stomach

Situs inversus totalis

Situs inversus thoracalis

Situs inversus abdominalis

Flieg auf
Situs inversus - chest

PA chest x-rays → Patients with Primary ciliary diskinesia.

Situs inversus totalis: Dextrocardia, right sided aorta, right sided gastric bubble and spleen, left sided liver.
Situs inversus - abdomen

Situs inversus totalis with stomach and spleen on the right, liver on the left side.
## Diagnostic Tests

### Radiologic

- **Chest X-ray**
  
  usually the first imaging examination
  
  → findings are often nonspecific

- **CT lung**
  
  to evaluate for infiltrates, bronchiectasis

- **X-ray / CT paranasal sinuses**
  
  pansinusitis, opacification of maxillary, ethmoid and frontal sinuses

- **US/X-ray/CT abdomen**
  
  to evaluate for situs inversus totalis

### Histologic

- **Mucosal biopsy**
  
  from ciliated epithelium (nasal, tracheal, adenoid tissue) > histopathologic and electron microscope examination

- **Semen analysis**
  
  to evaluate sperm motility and ultrastructure

### Special Tests

- **Saccharine test**
  
  Saccharine placed in the nose - measurement of speed of transport into nasopharynx

- **Audiologic testing**

- **Pulmonary function studies**
Treatment

• The cause of Kartagener’s Syndrome is genetic → no curative treatment

• Infection control:
  - Prophylactic antibiotics (long-term low-dose)
  - Immunizations/vaccines
  - Antibiotics: continuous or intermittent to treat airway infections

• Symptomatic:
  - Mucolytics
  - Bronchodilators, inhaled corticosteroids and pulmonary toilet

• Surgical:
  - Lung transplantation
Summary

• Kartagener’s Syndrome should always be considered in patients with recurrent respiratory infections and hx of situs inversus

• Symptoms result from impaired mucociliary clearance

• Clinical features include:
  • recurrent respiratory infections
  • bronchiectasis
  • sinusitis
  • chronic otitis
  • dextrocardia - situs inversus
  • infertility
References


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