AMSER Case of the Month
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35-Year-Old Female Presenting with Dyspnea

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Patient Presentation

- **History of Present Illness:** A 35-year-old female presents to clinic with dyspnea, productive cough, and wheezing.

- **Medical History:** Patient recalls several bouts of pneumonia as a child.

- **Surgical History:** Patient mentions two sinus surgeries, adding that she has residual congestion and pressure.

- **Social History:** Patient formerly smoked ½ pack-per-day.

- **Physical Exam:** Chest auscultation reveals bilateral wheezing.
Pertinent Labs

• **Basic Metabolic Panel:** Results are within normal limits.

• **Complete Blood Count:** Results are within normal limits.

• **Microbiology Sputum Culture:** Final yeast and bacterial cultures are still pending.
What imaging should we order?
### ACR Appropriateness Criteria

**Imaging modality ordered by outpatient pulmonologist.**

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Appropriateness Category</th>
<th>Relative Radiation Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>Radiography chest</td>
<td>Usually Appropriate</td>
<td>★</td>
</tr>
<tr>
<td>CT chest without IV contrast</td>
<td>May Be Appropriate (Disagreement)</td>
<td>★★★★</td>
</tr>
<tr>
<td>CT chest with IV contrast</td>
<td>May Be Appropriate</td>
<td>★★★</td>
</tr>
<tr>
<td>CT chest without and with IV contrast</td>
<td>Usually Not Appropriate</td>
<td>★★★★</td>
</tr>
<tr>
<td>FDG-PET/CT skull base to mid-thigh</td>
<td>Usually Not Appropriate</td>
<td>★★★★</td>
</tr>
<tr>
<td>MRI chest without and with IV contrast</td>
<td>Usually Not Appropriate</td>
<td>★★★★</td>
</tr>
<tr>
<td>MRI chest without IV contrast</td>
<td>Usually Not Appropriate</td>
<td>O</td>
</tr>
<tr>
<td>US chest</td>
<td>Usually Not Appropriate</td>
<td>O</td>
</tr>
</tbody>
</table>
Findings

- Embolization Coils
- Dextrocardia
- Situs Inversus
- Correct Orientation
Final Diagnosis

Kartagener’s Syndrome
Case Discussion: Kartagener’s Syndrome

- Kartagener’s syndrome is a subtype of primary ciliary dyskinesia
- It presents as a characteristic triad
  A. Bronchiectasis
  B. Chronic sinusitis
  C. Situs inversus

Case Discussion: Pathophysiology of Primary Ciliary Dyskinesia (PCD)

• Caused by abnormal ciliary beating
  • Most often caused by structural defects, especially dynein arms

• Motile cilia are also present in the lungs, middle ear, paranasal sinuses, reproductive organs, and brain ependyma
  • Therefore, most patients will also have non-chest symptoms, including subfertility

• Motile cilia in the embryonic node help create left-right body symmetry
  • Thus, ~50% of PCD patients will also have situs inversus

Arch Dis Child; 2014 Sep;99(9):850-6. DOI: 10.1136/archdischild-2013-304831.
Case Discussion: Diagnosis and Treatment

**Diagnosis**

- Current diagnostic guidelines are limited
- A low index of suspicion is recommended for Kartagener’s syndrome given risk of irreversible lung damage
- Nasal nitric oxide is often used as a screening tool; diagnosis is then confirmed with higher specificity test, like electron microscopy of nasal or bronchial biopsy
- Other available tests include high-speed videomicroscopy analysis, cell culture of ciliated cells, sperm motility, genetic testing, inhalation of colloid albumin tagged with 99Tc, and saccharin testing

**Treatment**

- There is a lack of evidence-based medicine in the management of PCD
- Most often, patients are managed empirically
- Consider chest physiotherapy for bronchiectasis and antibiotic treatment for exacerbations
- Chronic sinusitis and otitis media can be managed medically or surgically
- Patients experiencing subfertility can be referred for in vitro fertilization
- Patients generally have normal life expectancy when connected to multidisciplinary care
References

