Amyloidosis: Imaging a Zebra

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Goals of this Presentation

• Outline a patient work-up that leads to a diagnosis of amyloidosis
• Define amyloidosis
• Outline basic facts about the epidemiology, classification, etiology, and prognosis of amyloidosis
• Illustrate briefly two other presentations of amyloidosis
Initial Presentation of Our Patient: “Mr. M”

- Mr. M → 52-year-old man
- History → HTN, hypercholesterolemia, s/p cholecystectomy
- Presentation → abdominal pain x 4 hours radiating to his back
- DDx:
  - Pancreatitis
  - Abdominal aortic aneurysm
  - Perforation
• The work-up of Mr. M’s presentation…
Step 1 in Work Up: Radiographs and Labs

- Abdominal supine and erect radiographs → within normal limits
- Labs → within normal limits, incl. normal amylase/lipase
- Step 1 tests are inconclusive

∴ Step 2 → Abdominal CT
Step 2 in Work Up: Abdominal CT

…Reveals Finding #1

Finding #1

- 3.5 x 4.9 cm rounded soft tissue mass located in the inferior portion of the porta hepatis
- Extends into caudate lobe of liver
Abdominal CT

...Slice at Level of Paragastric and Paracaval Lymph Nodes Reveals Finding #2

Finding #2

• Paragastric lymphadenopathy
• Paracaval lymphadenopathy

...other slices reveal lymphadenopathy in the celiac nodes as well
Abdominal CT

...Liver Window Reveals Finding #3

Finding #3

- Diffuse, heterogeneous, low attenuation of liver
• An anatomy review will help to place Mr. M’s findings in context…
Anatomy of Lymph Nodes in the Region of the Porta Hepatis and the Pancreas

Finding #1
Soft tissue mass near porta hepatis/ head of pancreas

Finding #2
Lymphadenopathy of paragastric, paracaval, and celiac nodes

Finding #3: Diffuse, heterogeneous low attenuation of liver

• What are the differential diagnoses for Mr. M’s findings?
Differential Diagnoses for Mr. M’s Findings

DDx for Mass in the Region of the Pancreas on CT

- Abcess
- Carcinoma of liver, bile duct, pancreas
- Metastasis
- Lymphadenopathy

DDx for Generalized Low Attenuation Lesions on Non-contrast CT

- Diffuse malignancy
- Fatty infiltration
- Amyloidosis (uncommon)

…No clear diagnosis → CT-guided biopsies
• As imaging studies yield no clear diagnosis for Mr. M, the work-up requires a biopsy of Mr. M’s lesions.

• The biopsy is performed under CT guidance…
Step 3 in Work Up: Biopsy of Lesions

- CT-guided biopsies
  1. mass near porta hepatis
  2. liver tissue in area of low attenuation
- Congo red stain → Green bi-refringence

∴ Dx → AMYLOIDOSIS
• The pathological diagnosis is amyloidosis.

• Here is an example of the histology of amyloidosis…
Step 3 in Work Up: Biopsy of Lesions

- Histology of amyloidosis in liver tissue
- Note amyloid deposits in blood vessel walls

Liver tissue: H&E stain  
Congo Red stain

• Are there any other findings in Mr. M that support the diagnosis of amyloidosis?

• Thoracic CT reveals supportive findings…
Support of Diagnosis in Mr. M: CT and Pathology Findings in Thorax

- Mediastinal and hilar lymphadenopathy
- Ultrasound-guided biopsy of lymph nodes → amyloid deposits
• Mr. M has amyloidosis.

• What is amyloidosis?
Amyloidosis: Definition

- Not a single disease entity

- Term for diseases that lead to extracellular deposition of insoluble fibrillar proteins in tissues

- Fibrills are all beta-pleated sheets
Amyloidosis: Most Common Presentations

- Heart → CHF
- Kidneys → Nephrotic syndrome
- Liver → Hepatomegaly
Amyloidosis: Incidence and Classification

- Incidence: 1275-3200 patients/year in USA

<table>
<thead>
<tr>
<th>Subtype</th>
<th>Underlying Disease Process</th>
<th>Prognosis (Median survival after diagnosis)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Primary amyloidosis</td>
<td>Plasma cell dyscrasias</td>
<td>1-2 yrs</td>
</tr>
<tr>
<td>Secondary amyloidosis</td>
<td>Chronic inflammation</td>
<td>Variable</td>
</tr>
<tr>
<td>Familial amyloidosis</td>
<td>Mutated proteins</td>
<td>Up to 15 yrs</td>
</tr>
</tbody>
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• Let’s look at another patient’s presentation of amyloidosis…
Another Presentation of Amyloidosis: Patient #2

• Patient #2 → 19-year-old man with a 9 year history of progressive wheezing

• Chest radiographs were obtained
Patient #2 → PA Chest Radiograph

- Region of increased radio-opacity in tracheal lumen suggestive of a mass
- Left tracheal deviation

Note increased radio-opacity in tracheal lumen

Courtesy of Dr. Phillip Boiselle, BIDMC
• What is the differential diagnosis for an intratracheal mass as seen in Patient #2?
Patient #2 → Differential Diagnosis

• **DDx of Intratracheal Mass**
  - Adenoma
  - Neoplasm, primary or secondary
  - Amyloidosis
  - Hamartochondroma
  - Lymphoma
  - Many more…

• Next step in work up → MRI
• An MRI is needed to characterize further the intratracheal mass…
MRI of Tracheobronchial Amyloidosis

- Coronal view of trachea
- Exophytic mass
- Biopsy → amyloid

Courtesy of Dr. Phillip Boiselle, BIDMC
• Biopsy of the intratracheal mass revealed amyloid deposits, solidifying a diagnosis of amyloidosis for Patient #2.

• Let’s look briefly at one more way that amyloidosis can present…
Yet Another Presentation of Amyloidosis: Small Bowel Amyloidosis

• Barium study of small bowel

• Nodular, irregular thickening of bowel folds

• Biopsy → amyloid

Courtesy of Dr. Jonathan Kruskal, BIDMC
Take-Home Message

• Amyloidosis has a protean presentation

• Can affect almost any organ

• Uncommon disease that commonly appears on DDx for imaging findings…Even though it is a “zebra” diagnosis, it should be considered!

• If high clinical suspicion → biopsy to confirm
References

- Pear, BL. Other Organs and Other Amyloids. *Seminars in Roentgenology* 1986; XXI: 150-164.
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