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# 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”

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- 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

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

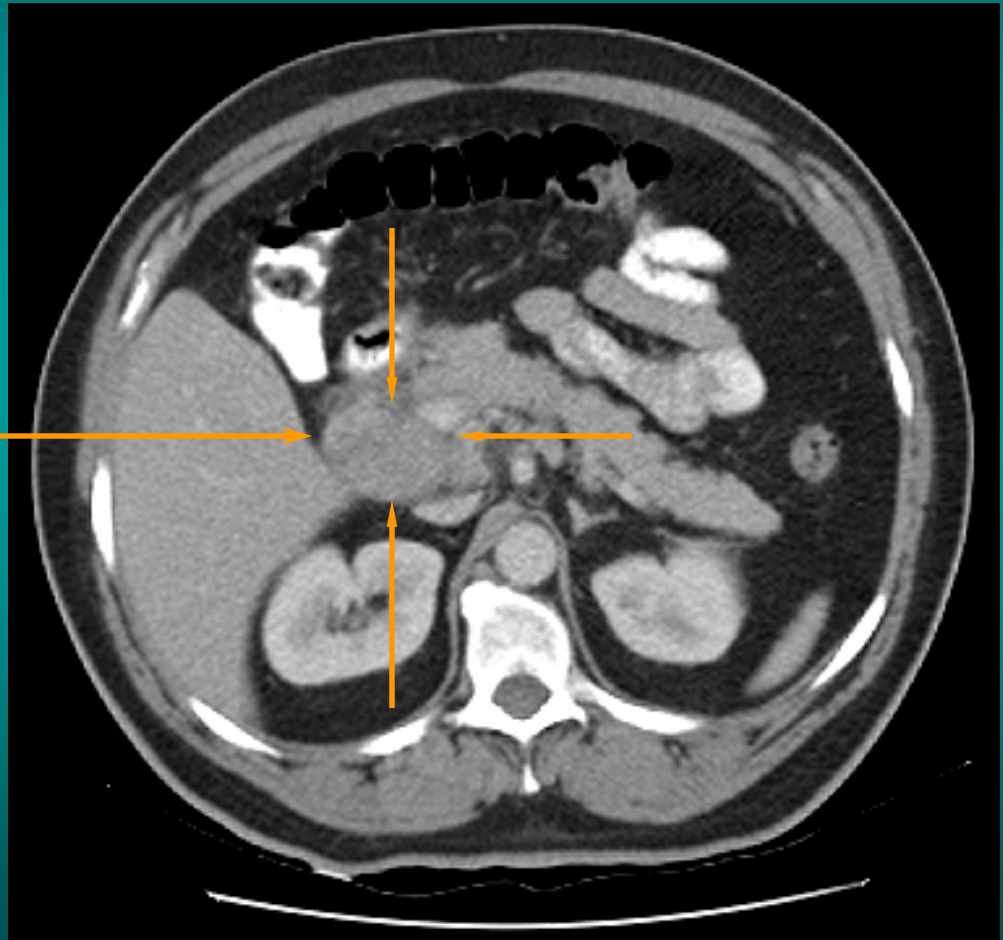
∴ Step 2 → Abdominal CT



# 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





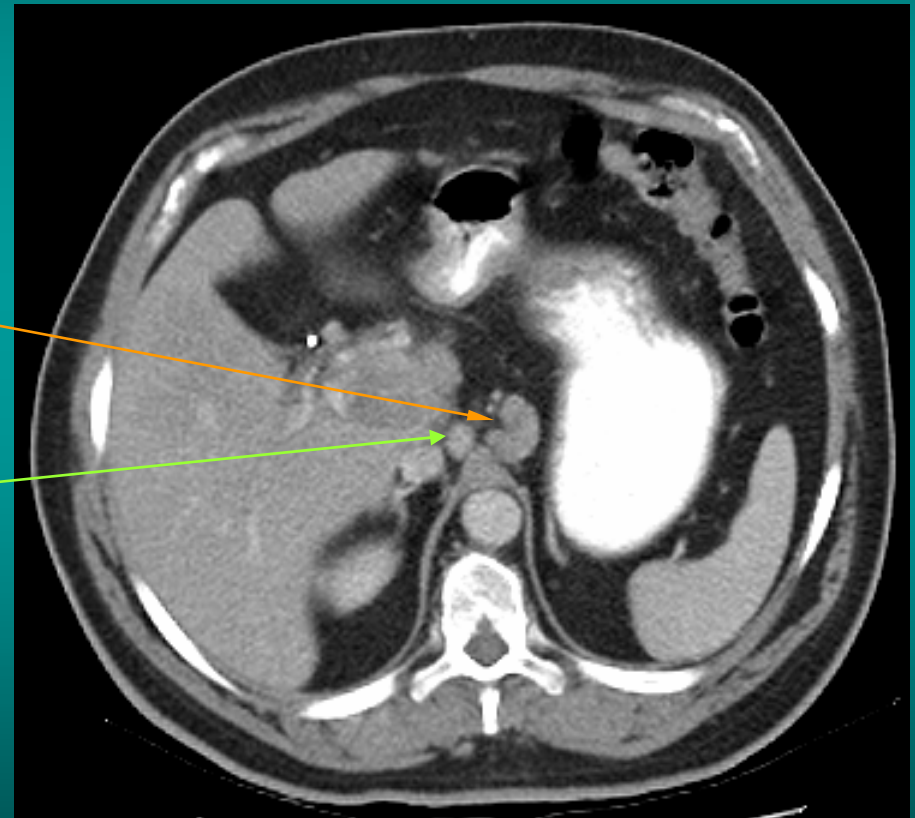
# Abdominal CT

...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



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# Abdominal CT

## ...Liver Window Reveals Finding #3

### Finding #3

- Diffuse, heterogeneous, low attenuation of liver







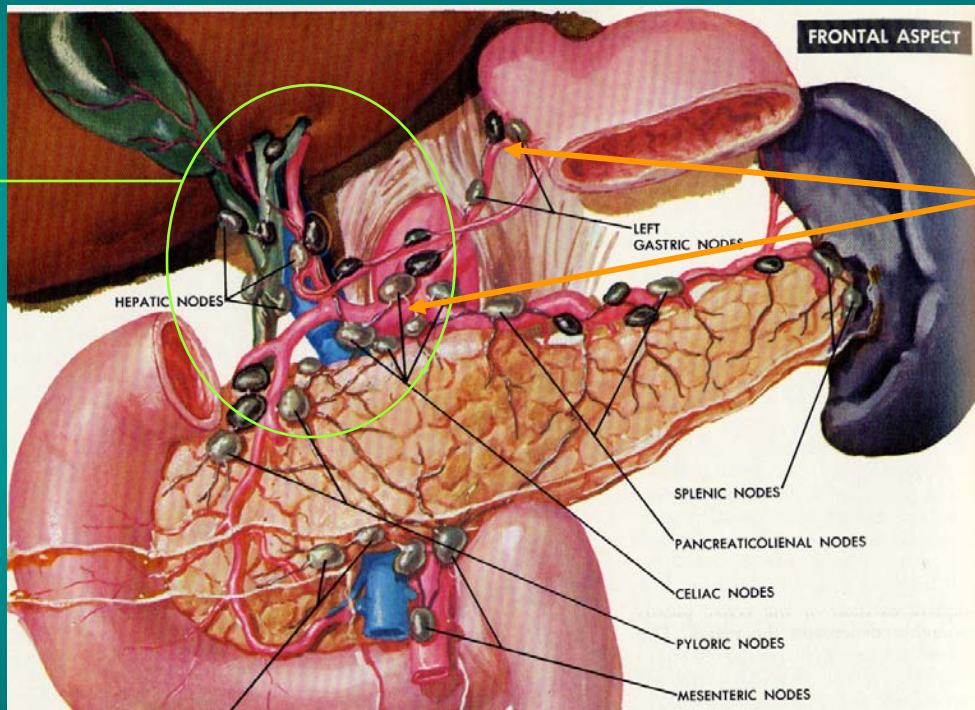
- An anatomy review will help to place Mr. M's findings in context...



# 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

•From Netter, FH. *The Ciba Collection of Medical Illustrations*/prepared by Frank H. Netter. West Caldwell, NJ: Ciba, 1983.



- What are the differential diagnoses for Mr. M's findings?



# Differential Diagnoses for Mr. M's Findings

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## DDx for Mass in the Region of the Pancreas on CT

- Abscess
- 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

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

∴ Dx → AMYLOIDOSIS

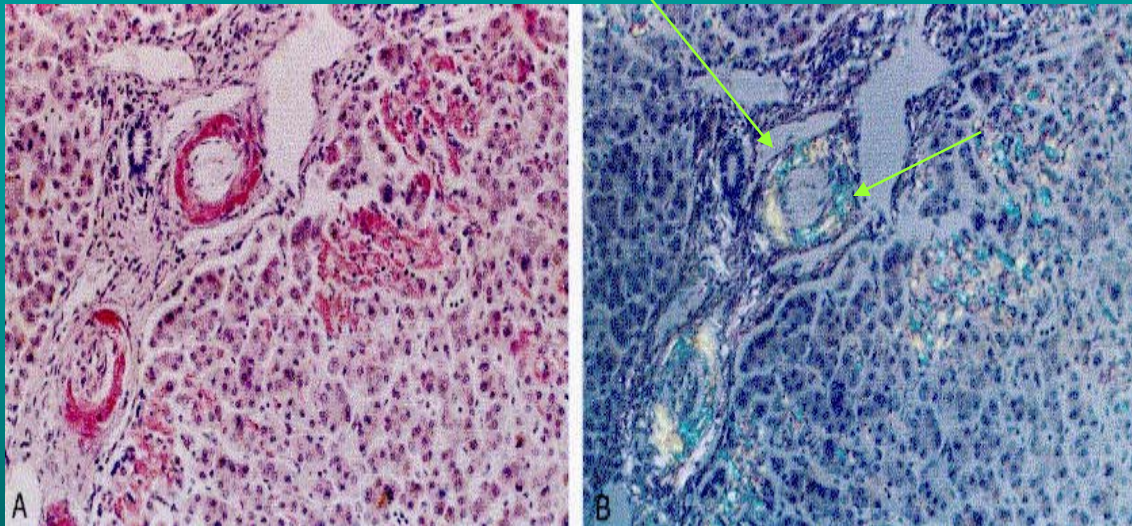


- The pathological diagnosis is amyloidosis.
- Here is an example of the histology of amyloidosis...



# 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

From *Robbins Pathologic Basis of Disease, 6th ed*, by Ramzi S Cotran, Vinay Kumar, and Tucker Collins, eds, 1425 pp. Philadelphia, Pa: WB Saunders & Co, 1998.





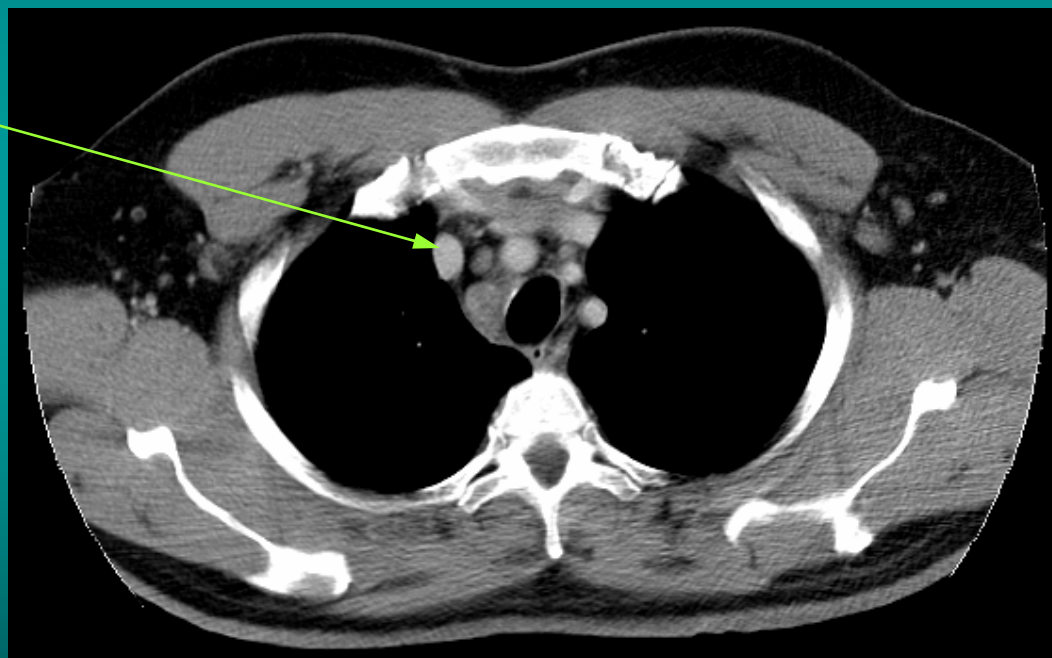
- Are there any other findings in Mr. M that support the diagnosis of amyloidosis?
- Thoracic CT reveals supportive findings...



# Support of Diagnosis in Mr. M: CT and Pathology Findings in Thorax

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- Mediastinal and hilar lymphadenopathy
- Ultrasound-guided biopsy of lymph nodes → amyloid deposits



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- Mr. M has amyloidosis.
- What is amyloidosis?



# Amyloidosis: Definition

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- 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

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- Heart → CHF
- Kidneys → Nephrotic syndrome
- Liver → Hepatomegaly



# Amyloidosis:

## Incidence and Classification

- Incidence: 1275-3200 patients/year in USA

Subtype	Underlying Disease Process	Prognosis (Median survival after diagnosis)
Primary amyloidosis	Plasma cell dyscrasias	1-2 yrs
Secondary amyloidosis	Chronic inflammation	Variable
Familial amyloidosis	Mutated proteins	Up to 15 yrs



- Let's look at another patient's presentation of amyloidosis...



# Another Presentation of Amyloidosis: Patient #2

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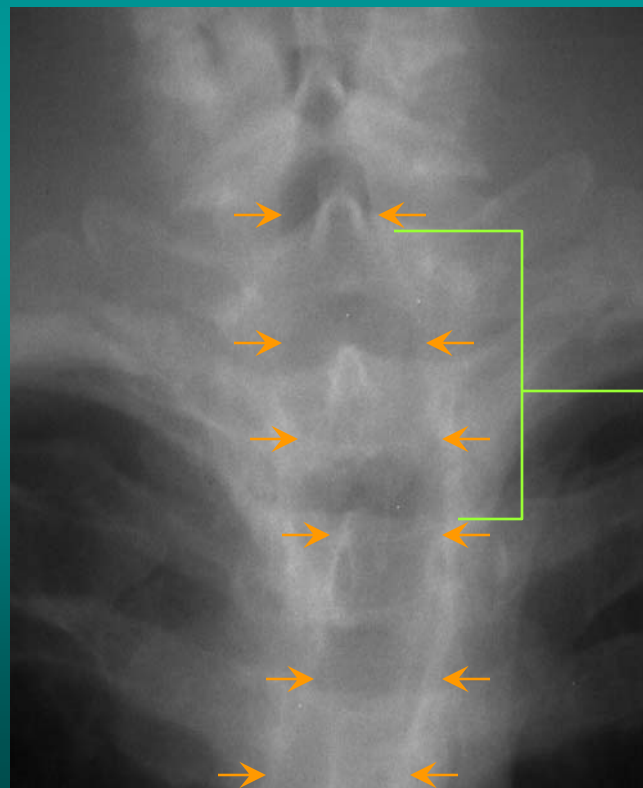
- Patient #2 → 19-year-old man with a 9 year history of progressive wheezing
- Chest radiographs were obtained





# 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



- What is the differential diagnosis for an intratracheal mass as seen in Patient #2?



# Patient #2 → Differential Diagnosis

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- DDx of Intratracheal Mass
  - Adenoma
  - Neoplasm, primary or secondary
  - Amyloidosis
  - Hamartochondroma
  - Lymphoma
  - Many more...
- Next step in work up → MRI

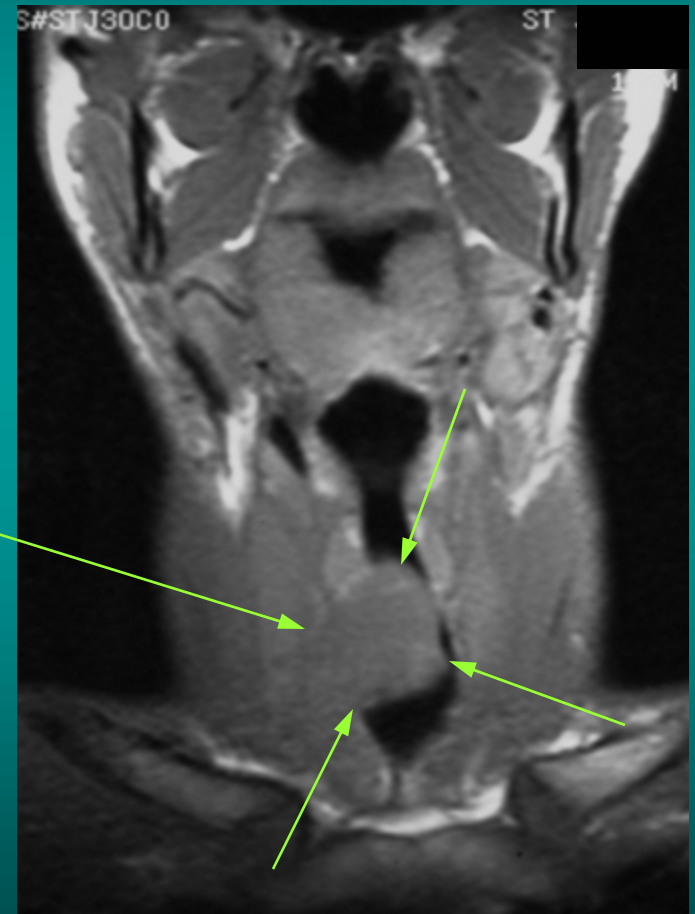


- An MRI is needed to characterize further the intratracheal mass...



# MRI of Tracheobronchial Amyloidosis

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





- 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

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- 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





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