AMSER Case of the Month
July 2020

Aortic Root Dilatation in Marfan Syndrome

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AMSER

Patient Presentation

- KM 19 y/o Female
- PMH: Marfan Syndrome, aortic root dilatation, mitral valve prolapse s/p repair, mild left ventricular systolic dysfunction (EF 50-54%)

- Presents to cardiologist to establish adult care
  - Intermittent, non-radiating chest pain for several years
  - Intermittent shortness of breath with exertion
  - Brief lightheadedness with postural changes (orthostatic)
  - Denies syncope, heart palpitations, edema, chest pressure, and PND

- PSH: Mitral valve repair with Carpentier annuloplasty ring (2011)
- Social Hx: occasional marijuana use, no tobacco or EtOH use

Pertinent Labs & Findings

- Chart review shows increase in aortic root dilatation measured via echocardiography
  - 2017 measured at 4.08cm
  - 2019 measured at 4.3cm

- Beta hCG negative

What Imaging Should We Order?

ACR Appropriateness Criteria

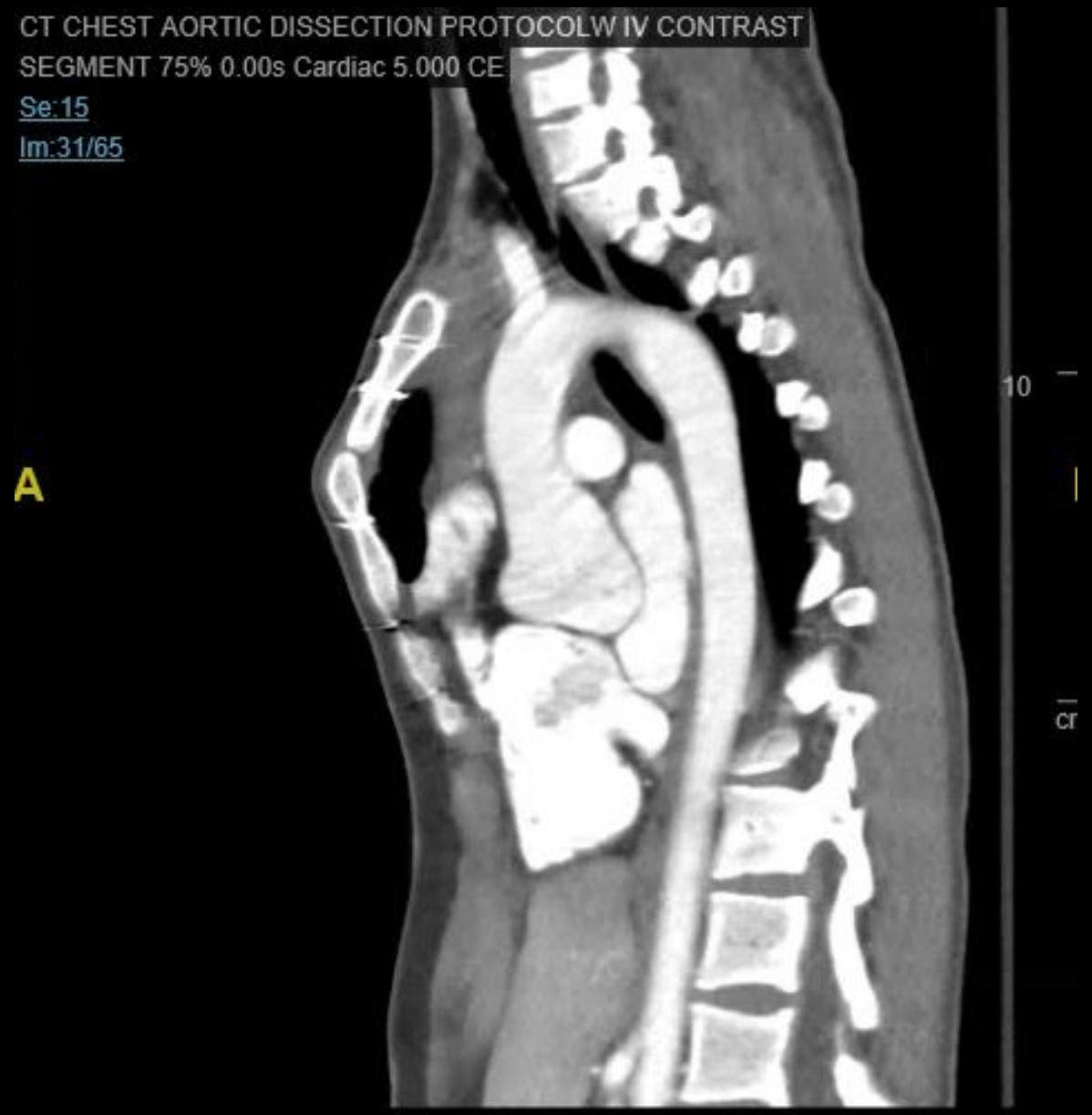
Ordered by cardiologist

American College of Radiology
ACR Appropriateness Criteria®
Suspected Thoracic Aortic Aneurysm

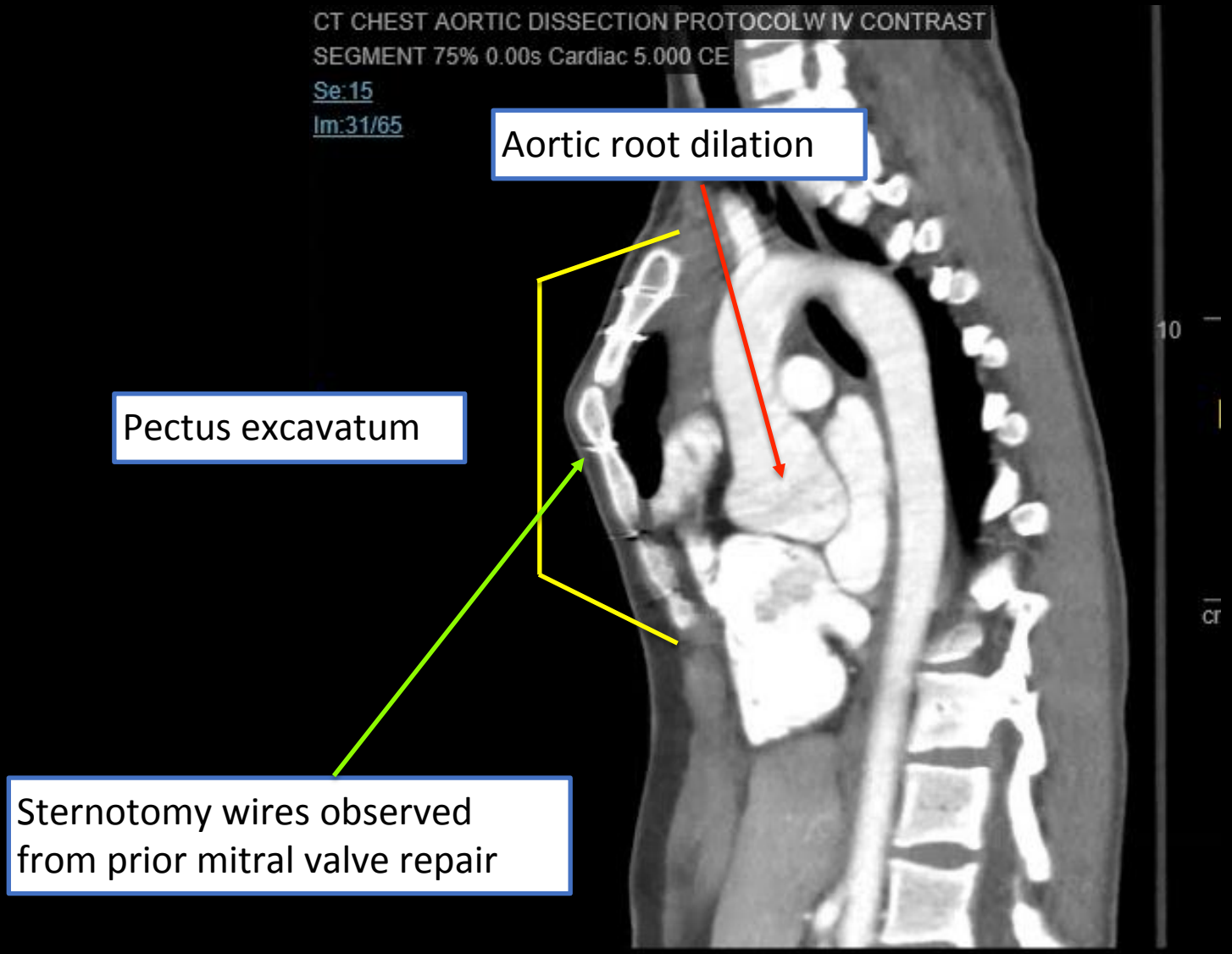
Variant 1: **Suspected thoracic aortic aneurysm. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
CTA chest with IV contrast	Usually Appropriate	☢☢☢
MRA chest with IV contrast	Usually Appropriate	O
MRA chest without IV contrast	Usually Appropriate	O
CT chest without IV contrast	May Be Appropriate	☢☢☢
US echocardiography transesophageal	May Be Appropriate	O
X-ray chest	May Be Appropriate	☢
CTA chest abdomen pelvis with IV contrast	May Be Appropriate (Disagreement)	☢☢☢☢☢
MRA chest abdomen pelvis with IV contrast	May Be Appropriate (Disagreement)	O
MRA chest abdomen pelvis without IV contrast	May Be Appropriate (Disagreement)	O
US echocardiography transthoracic resting	May Be Appropriate	O
CT chest abdomen pelvis without IV contrast	Usually Not Appropriate	☢☢☢☢
CT chest abdomen pelvis with IV contrast	Usually Not Appropriate	☢☢☢☢
CT chest abdomen pelvis without and with IV contrast	Usually Not Appropriate	☢☢☢☢
CT chest with IV contrast	Usually Not Appropriate	☢☢☢
CT chest without and with IV contrast	Usually Not Appropriate	☢☢☢
Aortography chest abdomen pelvis	Usually Not Appropriate	☢☢☢☢

Findings (unlabeled)

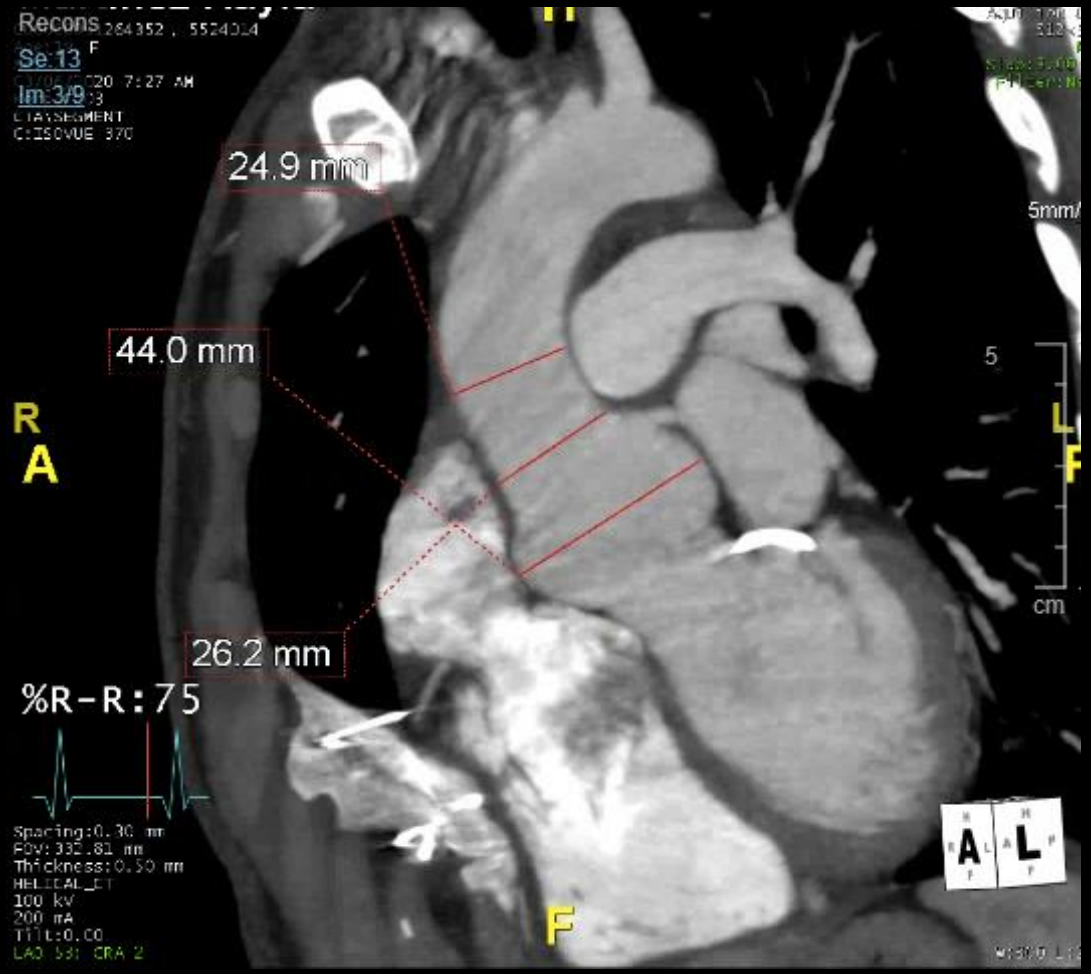


Findings: (labeled)

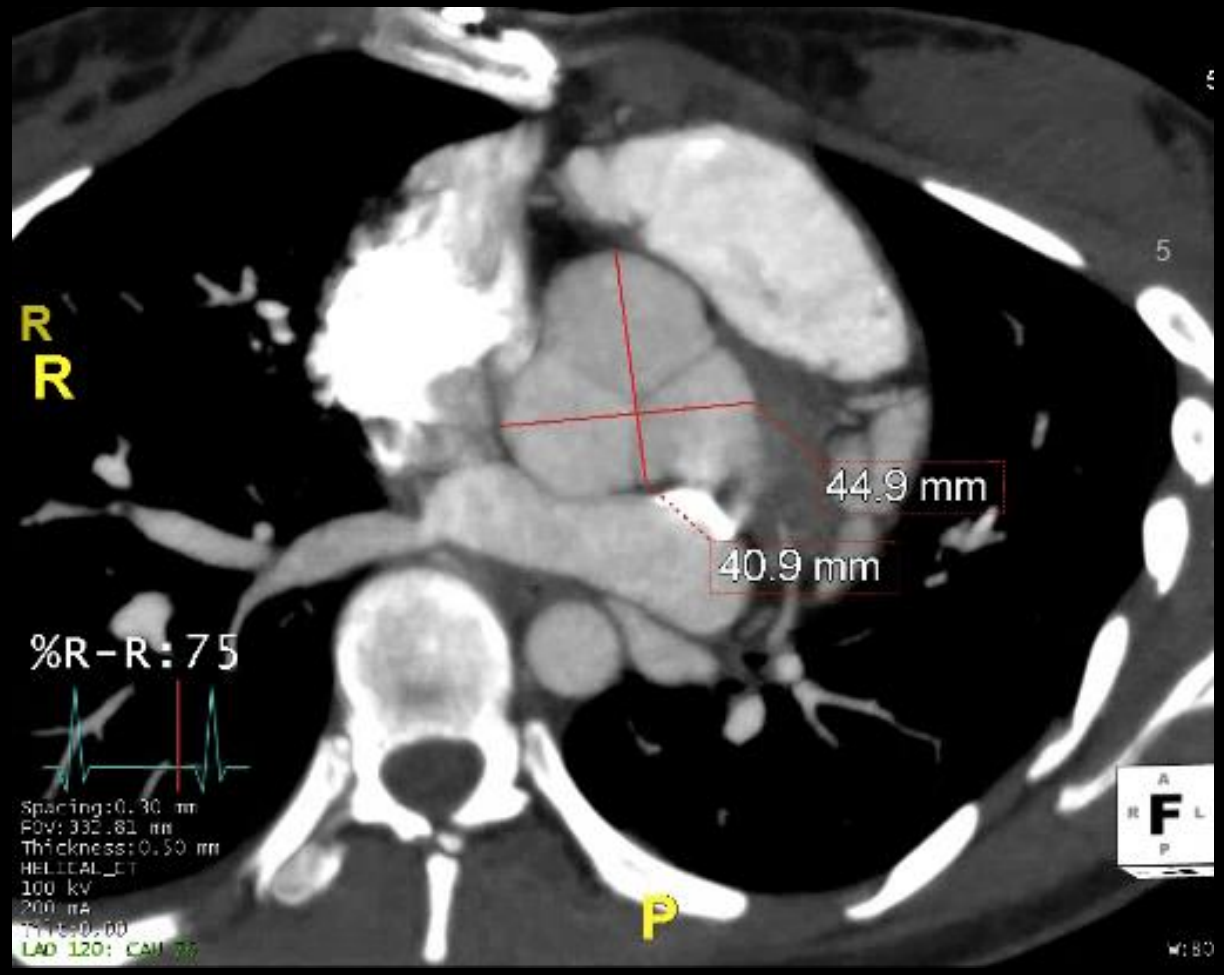


Findings: (labeled)

Aortic root is > 3.8cm (normal value) [1]



Aortic valve > 3.0 to 4.0 cm2 (normal value) [1]



Final Dx:
aortic root aneurysm

Discussion: Thoracic Aorta Dilation

- The normal thoracic aorta decreases in diameter as it courses distally
- normal values: [1]
  - 3.5-4.0 cm at aortic root
  - 2.4-2.7 cm at the diaphragm (distal thoracic aorta)

  - dilation <50% over normal= ***aortic ectasia*** [2]
  - dilation >50% over normal or >2 standard deviations above mean for age & sex= ***aortic aneurysm*** [2]

- Diagnosis of thoracic aortic root dilation is best made with CTA of the chest with IV contrast and best surveilled with CTA of the chest, abdomen, and pelvis with IV contrast
  - Provides the highest sensitivity for identifying a developing dissection
    - (82% sensitivity and 100% specificity)[3]

Discussion: Treatment of Aortic Root Dilation [1]

- >4.0cm diameter
  - β-blocker therapy
  - maintain strict blood pressure control (<120/80)
  - moderate physical activity restriction
  - perform yearly follow-up by TTE and/or CT/MRI

- In those WITHOUT connective tissue disorders, perform surgical repair at >4.5cm diameter

- In those WITH connective tissue disorders, perform surgical repair >5.0 cm diameter or progressive aortic growth >0.5 cm/y

Discussion: Marfan Syndrome

Marfan Syndrome [4]

- Autosomal dominant connective tissue disorder caused by mutation of the *FBN1* gene resulting tissue instability due to decreased function of Fibrillin-1

- Frequency: 1 in 5,000

- Common findings and sequelae include…
  - Tall body habitus with arm span that exceeds height
  - Arachnodactyly
  - Pectus excavitum or pectus carinatum
  - Mitral value prolapse
  - Aortic root dilation → can result in aortic dissection or rupture
  - Ectopia lentis
  - Berry aneurysms → subarachnoid hemorrhage
  - Spontaneous pneumothorax

References:

1. Cozijnsen, L., Braam, R. L., Waalewijn, R. A., Schepens, M. A., Loeys, B. L., Oosterhout, M. F. V., … Mulder, B. J. (2011). What Is New in Dilatation of the Ascending Aorta? Circulation, 123(8), 924–928. doi: 10.1161/circulationaha.110.949131
2. Nataf, P. (2006). Dilation of the thoracic aorta: medical and surgical management. Heart, 92(9), 1345–1352. doi: 10.1136/hrt.2005.074781
3. ACR Appropriateness Criteria® Thoracic Aorta Interventional Planning and Follow-Up. (2017). Retrieved March 11, 2020, from https://acsearch.acr.org/docs/3099659/Narrative/
4. Marfan syndrome - Genetics Home Reference - NIH. (2020, March 3). Retrieved March 11, 2020, from https://ghr.nlm.nih.gov/condition/marfan-syndrome#statistics