AMSER Case of the Month December 2021

57-year-old male with shortness of breath, heart palpitations, and weight gain

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Patient Presentation

- HPI: 56 year-old male with shortness of breath (SOB), heart palpitations and weight gain
- PMH: obesity, hypertension, atrial fibrillation
- FH: premature coronary artery disease in maternal grandmother
- Medications: diltiazam, hydrochlorothiazide – lisinopril combination pill
- Physical Exam: cough, SOB, palpitations, JVD present, irregularly irregular rhythm, faint radial pulses bilaterally, mild bilateral peripheral edema

Pertinent Labs

- Pro-BNP: 2,207 (elevated)
- Creatinine: 1.4 (elevated; four days prior: 1.06 (normal))
- ALT: 20
- AST: 18
- Kappa QNT Free Light Chains: 20.4 (Reference Range: 3.3-19.4)

EKG

- Atrial fibrillation with rapid ventricular response, heart rate 109 BPM, left ventricular hypertrophy with repolarization abnormalities

What Imaging Should We Order?

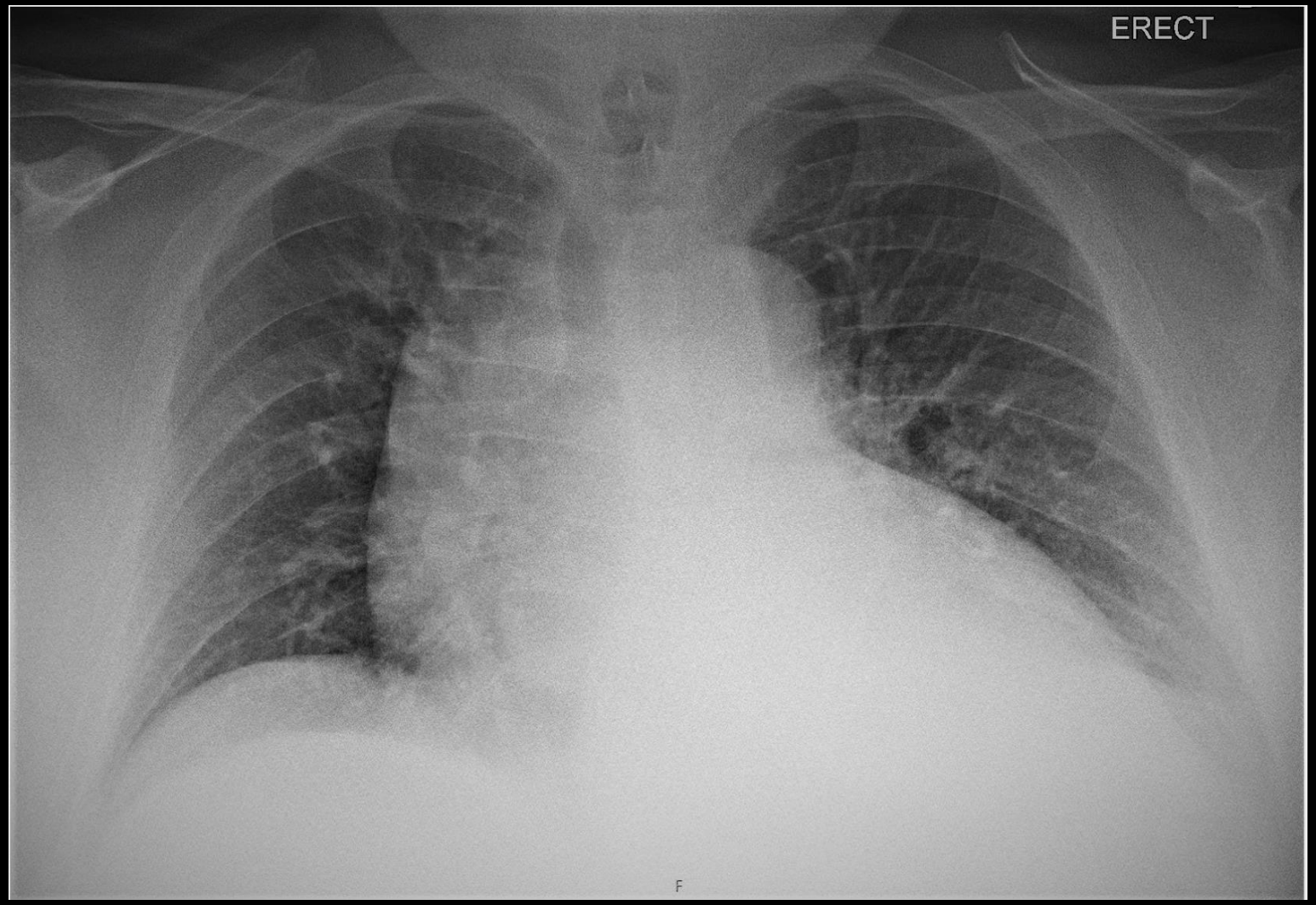
Select the applicable ACR Appropriateness Criteria

This imaging modality was ordered by the ER physician

Variant 1: Dyspnea due to heart failure. Ischemia not excluded.

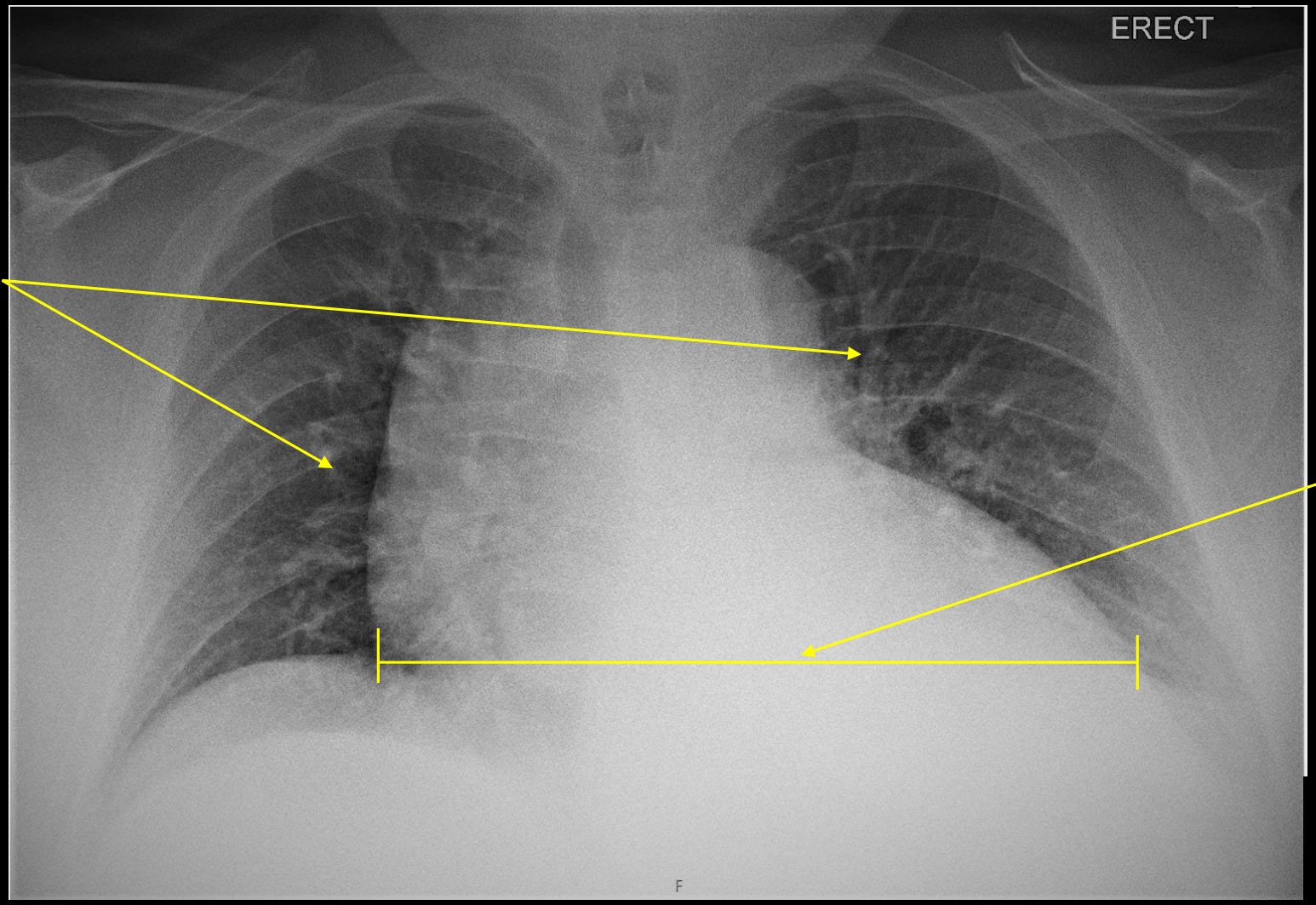
Procedure	Appropriateness Category	SOE	Adult RRL	Peds RRL	Rating	Median	Final Tabulations								
							1	2	3	4	5	6	7	8	9
US echocardiography transthoracic resting	Usually appropriate		O 0 mSv	O 0 mSv [ped]	9	n/a	0	0	0	0	0	0	0	0	0
US echocardiography transthoracic stress	Usually appropriate		O 0 mSv	O 0 mSv [ped]	9	n/a	0	0	0	0	0	0	0	0	0
Radiography chest	Usually appropriate		☢ <0.1 mSv	☢ <0.03 mSv [ped]	9	n/a	0	0	0	0	0	0	0	0	0
Arteriography coronary with ventriculography	Usually appropriate		☢☢☢ 1-10 mSv		8	n/a	0	0	0	0	0	0	0	0	0
MRI heart function and morphology without and with IV contrast	Usually appropriate		O 0 mSv	O 0 mSv [ped]	8	n/a	0	0	0	0	0	0	0	0	0
MRI heart with function and inotropic stress without and with IV contrast	Usually appropriate		O 0 mSv	O 0 mSv [ped]	7	n/a	0	0	0	0	0	0	0	0	0

Chest X-Ray (Unlabeled)



Chest X-Ray (Labeled)

Increased bronchovascular markings and cephalization of the pulmonary vessels suspicious for mild pulmonary edema



Enlarged cardiac silhouette

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MRI heart function and morphology without and with IV contrast	Usually appropriate		O 0 mSv	O 0 mSv [ped]	8	n/a	0	0	0	0	0	0	0	0	0
MRI heart with function and inotropic stress without and with IV contrast	Usually appropriate		O 0 mSv	O 0 mSv [ped]	7	n/a	0	0	0	0	0	0	0	0	0

Transthoracic Echocardiogram

- Ejection fraction: 55 to 60%
- Severe concentric hypertrophy
- No wall motion abnormalities
- Biatrial enlargement

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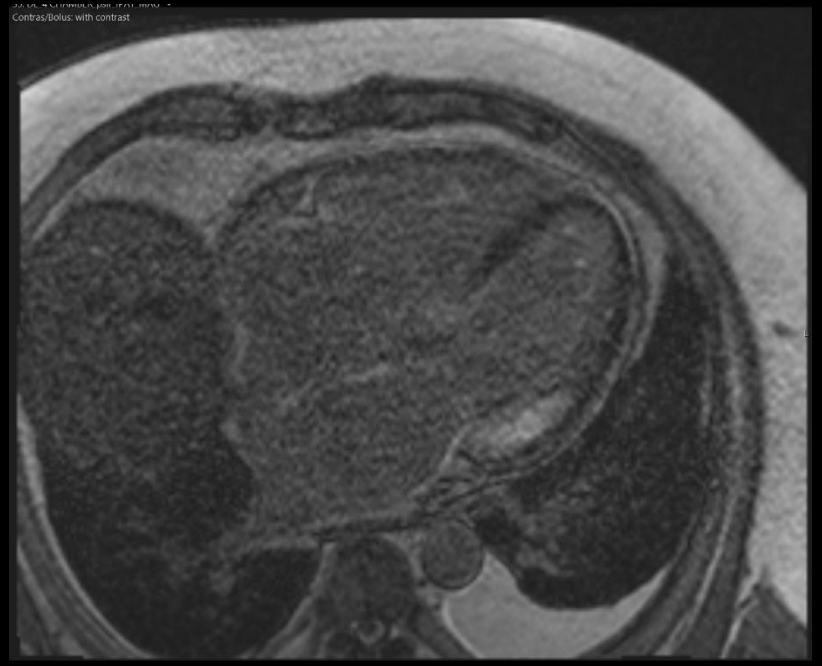
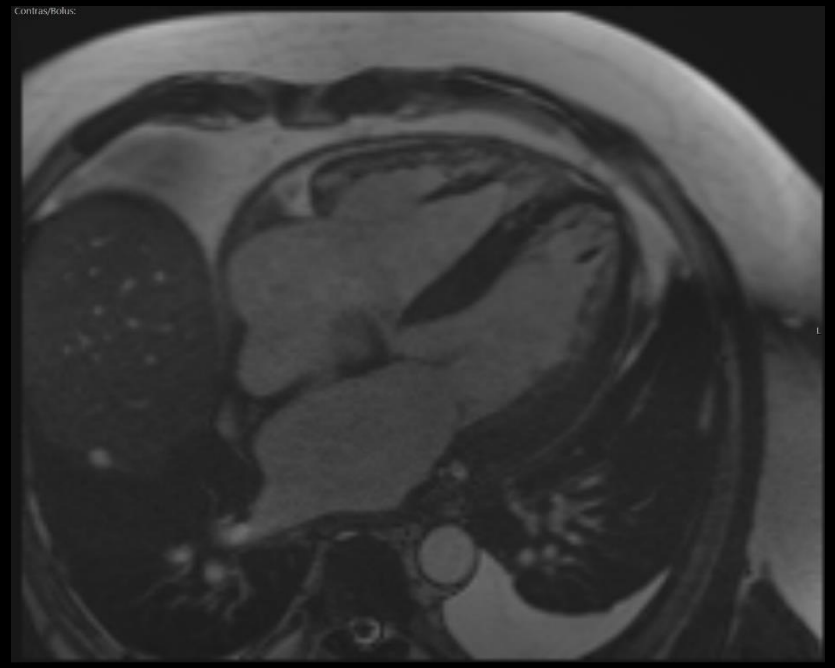
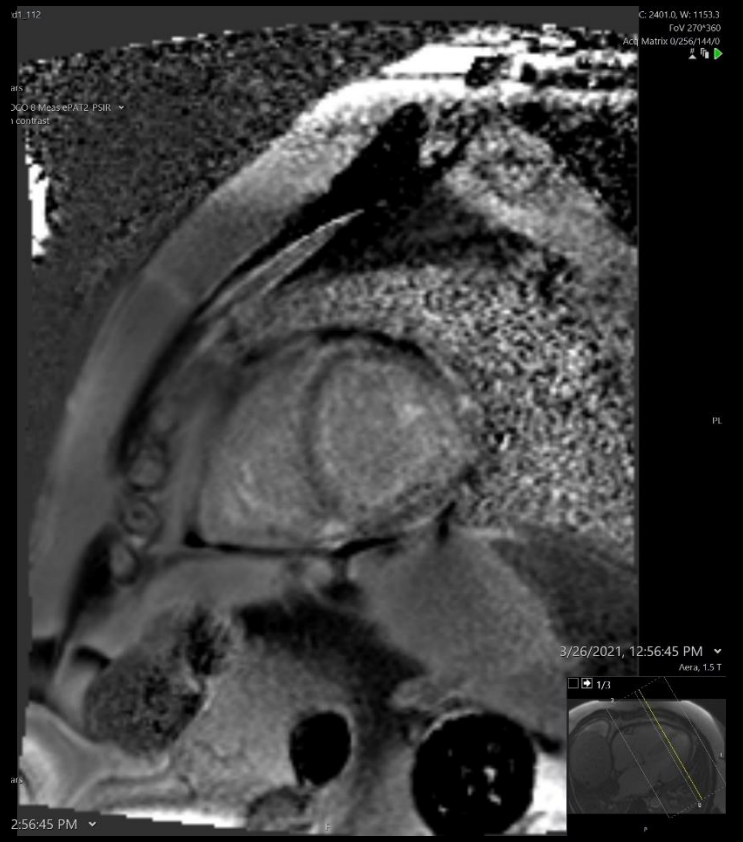
Cardiac MRI (Unlabeled)

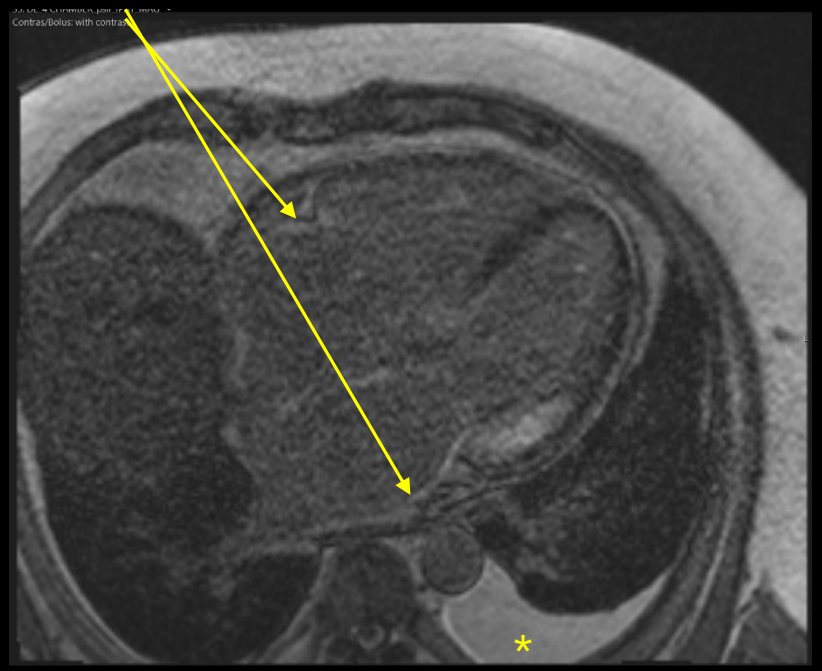






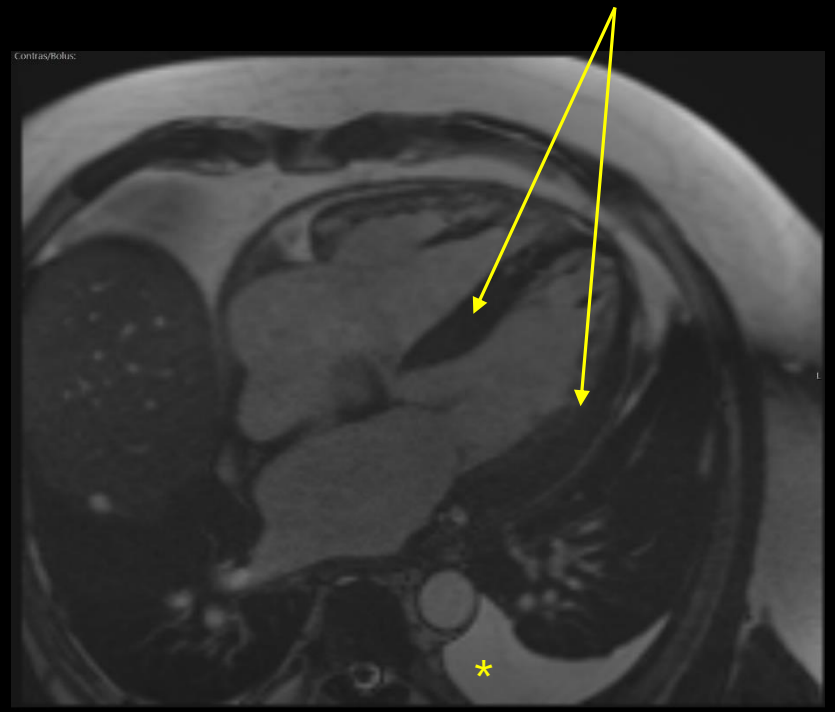
Cardiac MRI (Labeled)

Delayed myocardial hyperenhancement of all four chambers (arrows highlight bilateral atria)



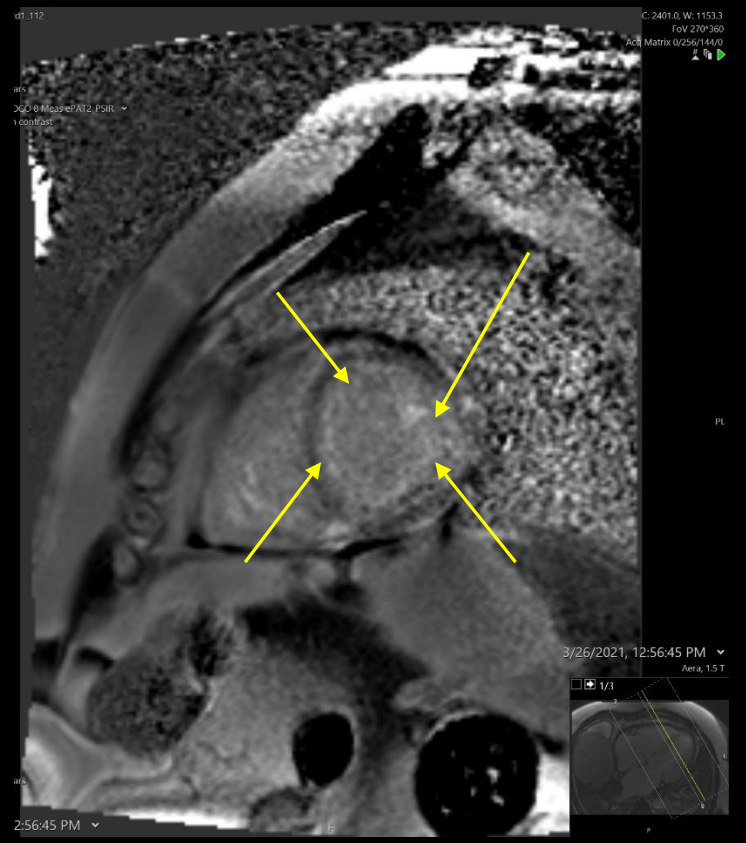
Inversion Recovery Sequence

LV wall thickening with septum measuring 15mm



Balanced Steady State Precession (bSSFP)

Diffuse subendocardial delayed myocardial hyperenhancement



Inversion Recovery Sequence

* Left pleural effusion

Final Dx:

Cardiac Amyloidosis

Case Discussion

Etiology: A disorder caused deposition of amyloid fibrils (proteins that self assemble) which cause functional and structural organ damage, such as restrictive cardiomyopathy. There are two main forms of cardiac amyloidosis: amyloid light chain (AL; most common) and transthyretin-related (TTR).

Clinical Presentation:

- Right ventricular failure (lower extremity edema, elevated jugular venous pressure, hepatic congestion, ascites and dyspnea)
- Arrhythmias or atrioventricular block (syncope)

Differential Diagnosis:

- Other causes of restrictive cardiomyopathy (hypertrophic cardiomyopathy, cardiac sarcoidosis, cardiac lymphoma

Cardiac Amyloidosis - Diagnosis

Echocardiography: Reduction in global longitudinal strain with relative apical sparing; hypertrophy with non-dilated ventricles; pericardial and pleural effusions common findings

Cardiovascular MRI: Can detect early cardiac amyloidosis before LVH, but cannot distinguish between types of amyloidosis; late gadolinium enhancement (LGE)

Serum Protein Immunofixation, Protein immunofixation or Serum Free Light Chain Ratio Analysis: Identification of monoclonal protein

Endomyocardial biopsy: Needed for diagnosis and determining type of amyloid

Cardiac Amyloidosis - Treatment

Treatment for heart failure:

- Loop diuretics
- Beta blockers, ACE inhibitors and calcium channel blockers no benefit or contraindicated in this population
- Few are eligible for heart transplant

Treatment for underlying disease:

- Chemotherapy and/or autologous stem cell transplantation
- Liver transplantation

Cardiac Amyloidosis - Prognosis

With early diagnosis and treatment, survival can be significantly prolonged

Prognostic model	Risk groups		Survival in patients not undergoing stem cell transplantation		Survival in patients undergoing stem cell transplantation	
			Median in months	5-year survival rate (%)*	Median in months	4-year survival rate (%)*
Mayo Stage[1,2]	Stage I	Cardiac troponin <0.035 mcg/L and NT-proBNP <332 ng/L	26	28	Not reached at 40 months	85¶
	Stage II	Any 1 factor high	11	12	Not reached at 40 months	75¶
	Stage III	Cardiac troponin ≥0.035 mcg/L and NT-proBNP ≥332 ng/L	4	8	8	25¶
Revised Mayo Stage[3]	Stage I	NT-proBNP <1800 ng/L, cardiac troponin T <0.025 mcg/L, and difference between involved and uninvolved serum free light chains <18 mg/dL	55	50	Not reached	87
	Stage II	Any 1 factor high	19	35	97	72
	Stage III	Any 2 factors high	12	20	58	56
	Stage IV	NT-proBNP ≥1800 ng/L, cardiac troponin T ≥0.025 mcg/L, and difference between involved and uninvolved serum free light chains ≥18 mg/dL	5	15	22	46

References:

- Georgiades, Christos S., et al. ”Amyloidosis: Review and CT Manifestations.” *RadioGraphics*, 1 Mar. 2004, https://pubs.rsna.org/doi/10.1148/rg.242035114
- Ronald M. Witteles and Michaela Liedtke Ronald M. Witteles Division of Cardiovascular Medicine, et al. “Al Amyloidosis for the Cardiologist and Oncologist: Epidemiology, Diagnosis, and Management.” *JACC*, 1 Sept. 2019, https://www.jacc.org/doi/full/10.1016/j.jaccao.2019.08.002
- “Cardiac Amyloidosis Clinical Manifestations and Diagnosis.” *UpToDate*, https://www.uptodate.com/contents/cardiac-amyloidosis-clinical-manifestations-and-diagnosis
- Czeyda-Pommersheim, Ferenc, et al. “Amyloidosis: Modern Cross-Sectional Imaging.” *RadioGraphics*, 31 July 2015, https://pubs.rsna.org/doi/full/10.1148/rg.2015140179.

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