Adult Congenital Heart Disease: What Are The Most Common Untreated Conditions That Should Not Be Missed?

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Disclosure

- None

Outline

- Shunt lesions
- Abnormal vascular connections
- Valvular lesions
- Aortic arch and coronary arteries

Atrial septal defect

- ASD can remain undiagnosed until adulthood
- Atrial level shunt is dependent on ventricular compliance
- Reduction in LV compliance: increases left to right shunt (hypertension, ischemic heart disease, cardiomyopathy, aortic and mitral valve disease)
- As a consequence, an ASD may become hemodynamically more important with age

2018 AHA/ACC guideline: Circulation. 2019;139: 698–800
2020 ESC Guidelines: *European Heart Journal*, 2021, Pages 563–645

Atrial septal defects

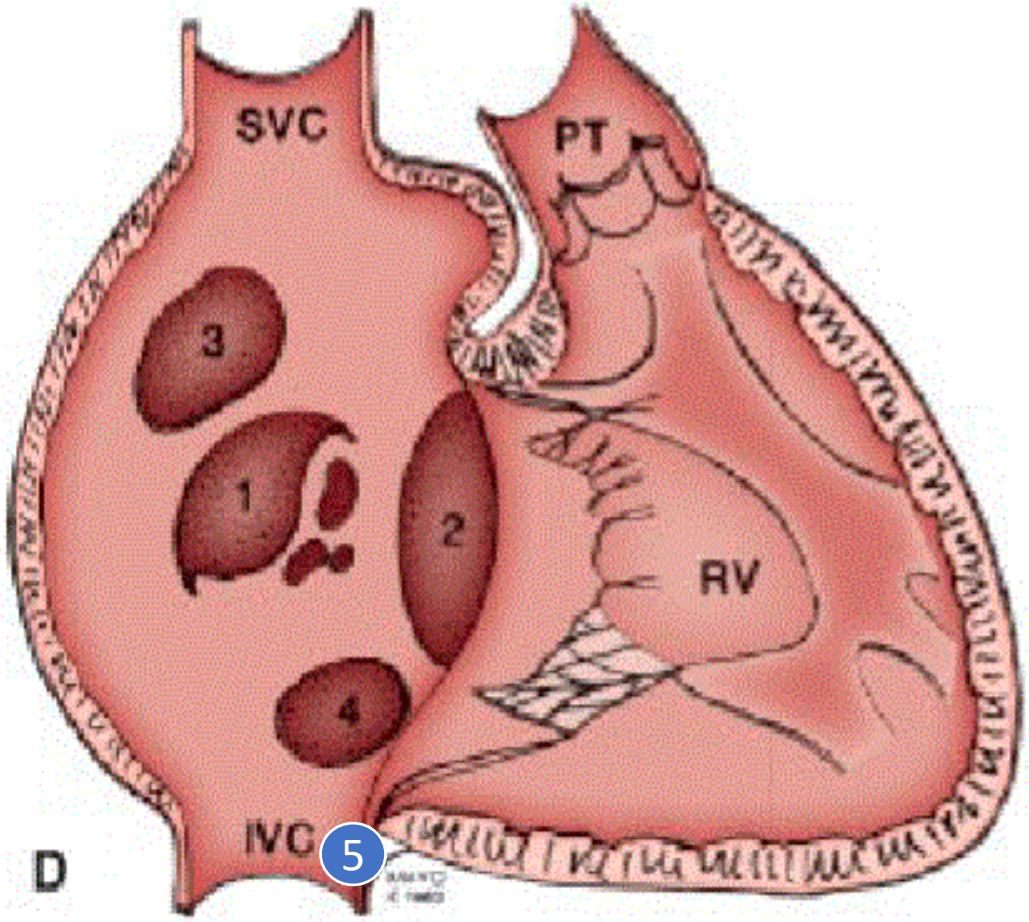
1. Secundum ASD: 80%

2. Primum ASD:15%

3. Superior sinus venosus defect: 5%

4.Unroofed coronary sinus: <1%

5.Inferior sinus venosus defect: <1%

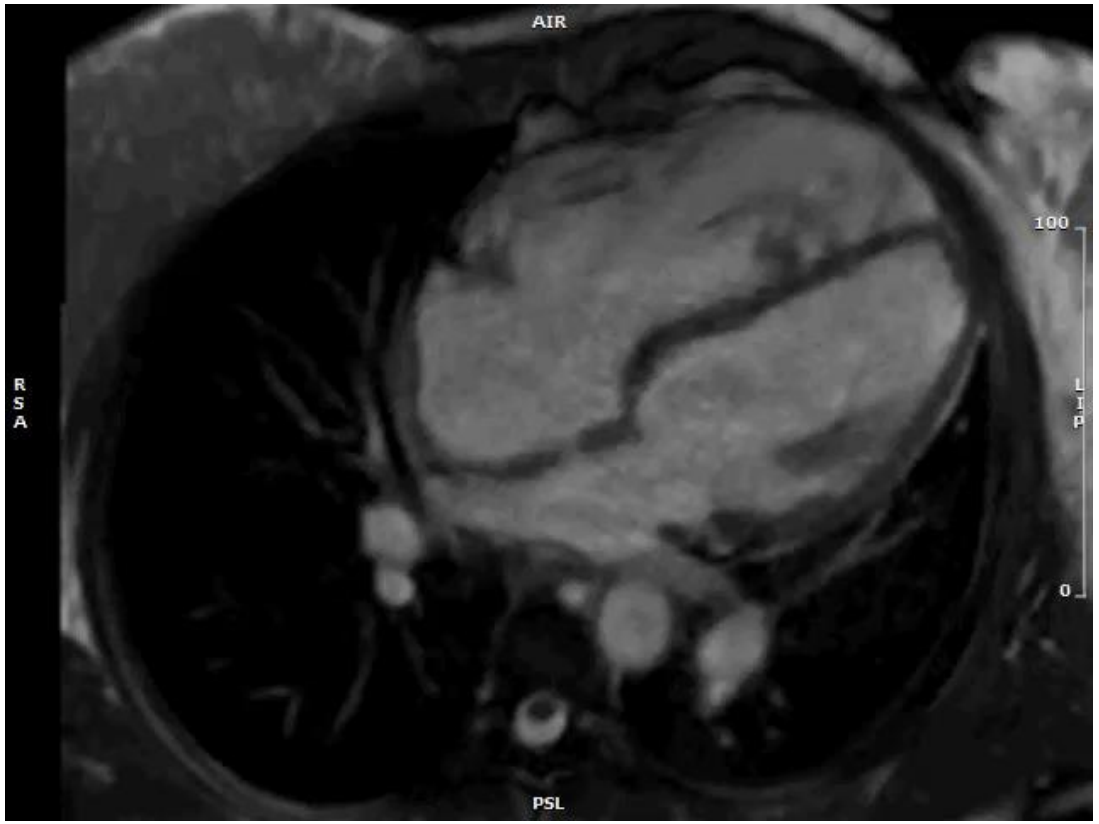


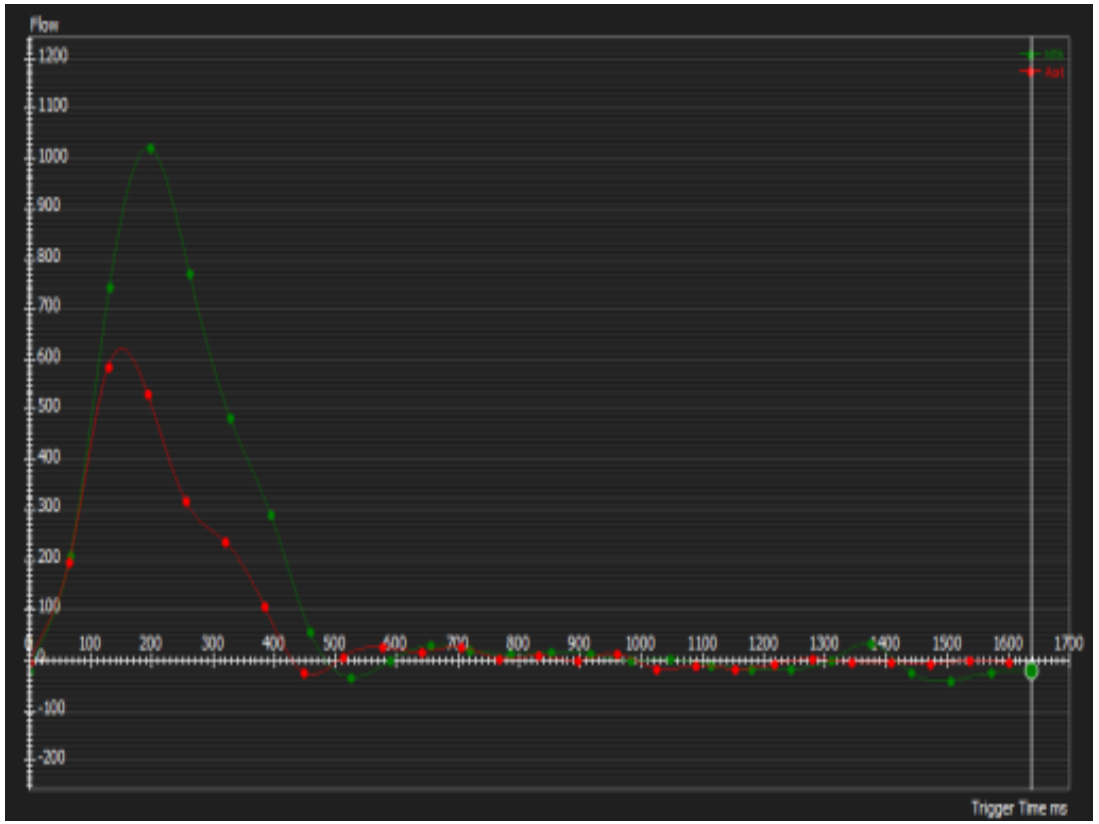
Moss and Adams: 2016 edition

A 39-year-old female with complete heart block diagnosed during pregnancy

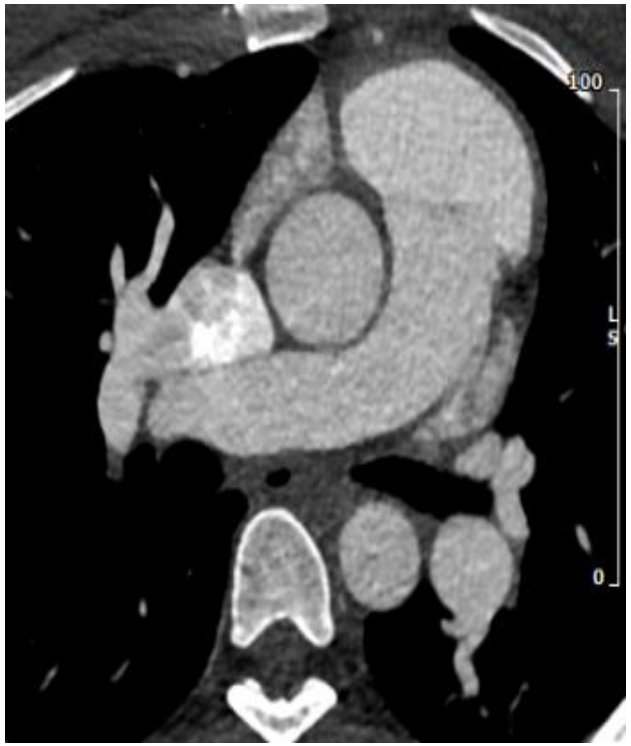
- Work up completed after delivery

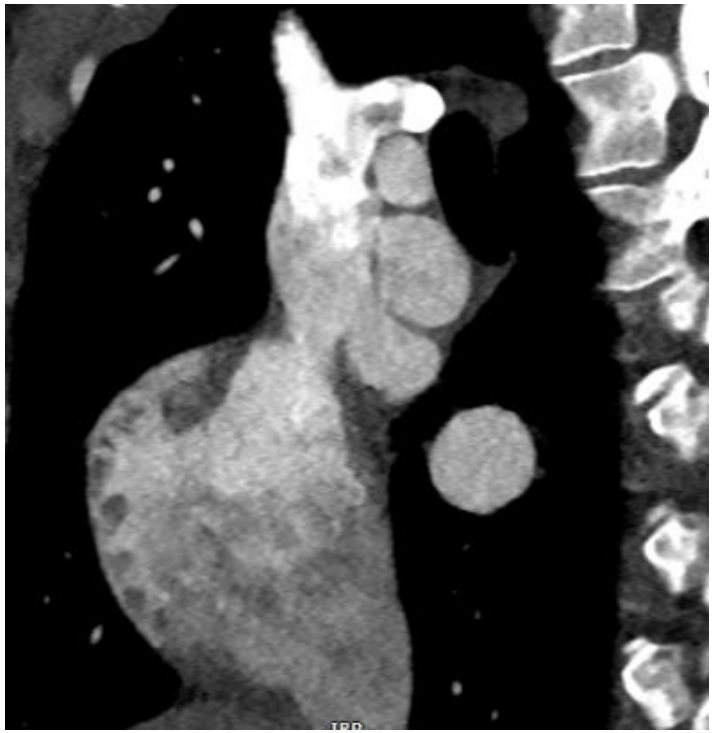
Cardiac MRI: RV Volume and QP:QS 2:1

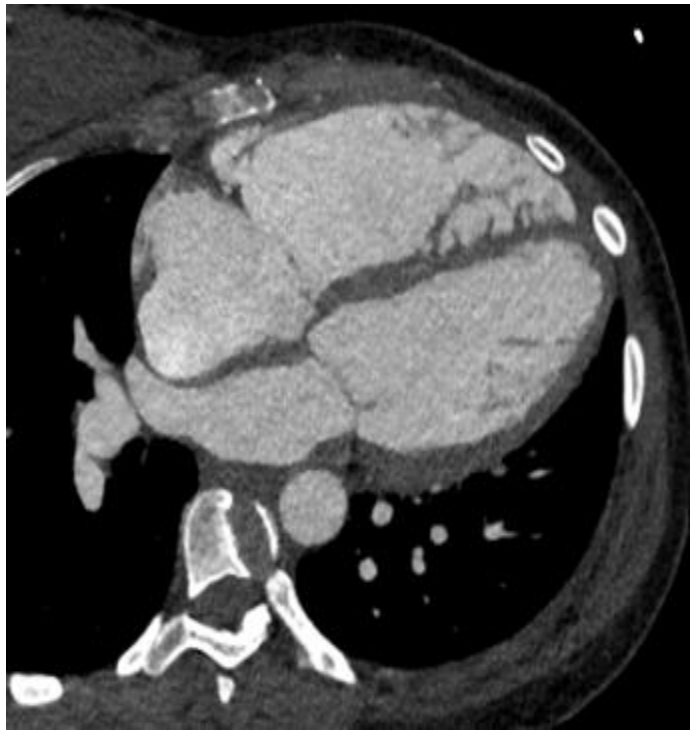




Cardiac CT to evaluate pulmonary veins







Final Diagnosis

- Complete heart block
- Sinus venosus ASD and PAPVR: QP:QS 2:1

- Underwent surgical repair and pacemaker placement

ASD: Clinical presentation and natural history

- Majority develop symptoms beyond the fourth decade
- With increasing age, tachyarrhythmias become more common
- Systemic embolism may be caused by paradoxical embolism or atrial flutter/fibrillation

ASD: Diagnostic workup

- Fixed splitting of the second heart sound and a systolic pulmonary flow murmur
- Echocardiography (TTE/TEE) is the first-line diagnostic technique
- CMR: RV volume, Qp:QS
- Cardiac CT: evaluation of pulmonary venous connection
- Cardiac catheterization: to determine PVR
- Exercise testing: desaturation in the setting of pulmonary arterial hypertension

ASD: Management

- Surgical
- Device closure in Cath lab
- No closure: Left ventricular disease, severe pulmonary hypertension
- Operative risk must be weighed against the potential benefit of intervention

ASD: Regular follow up after repair

- Residual shunt, elevated PAP, or arrhythmias

- Repaired at adult age (particularly >40 years)

ASD: Other considerations

- Exercise/sports: no restrictions in asymptomatic patient with no pulmonary hypertension, no significant arrhythmias and normal RV function

- Pregnancy: low risk in patients without PHTN, increased risk of paradoxical embolism.

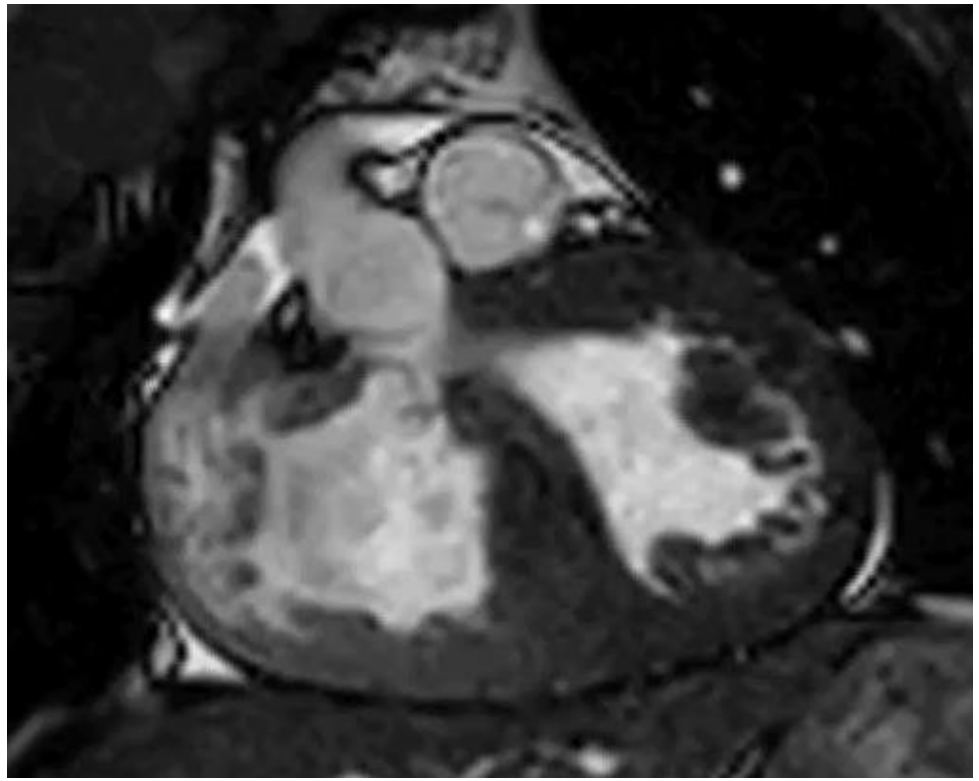
- IE prophylaxis: recommended for 6 months after device closure

A 34-year-old male with new onset syncopal episodes

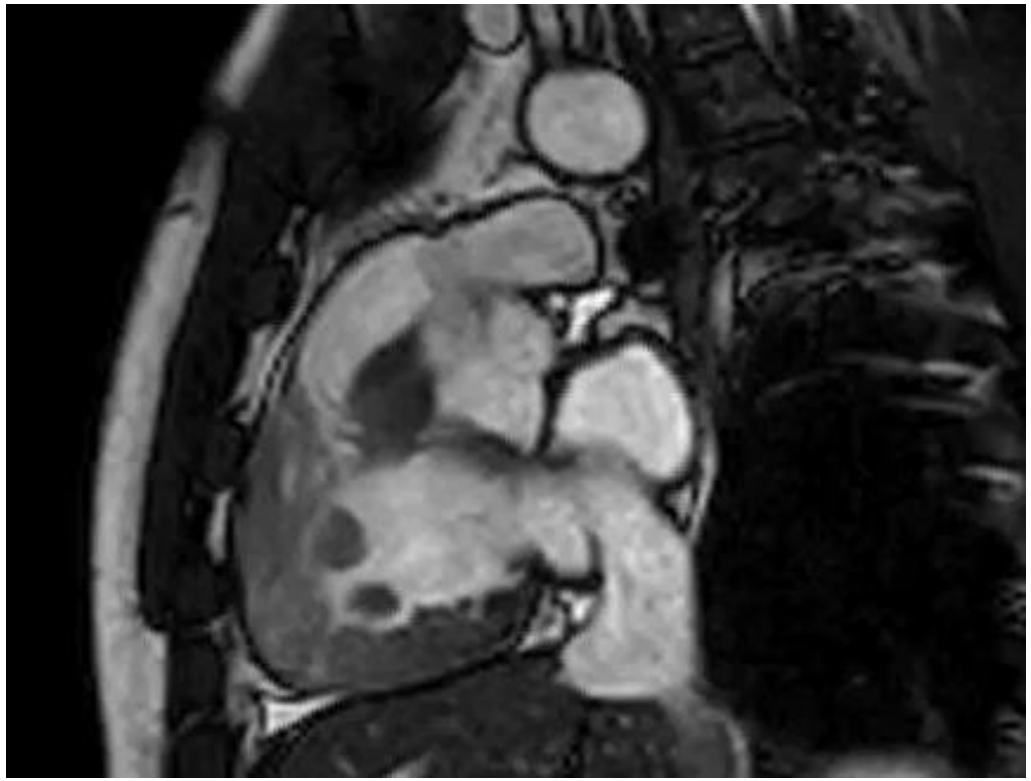
- Differential diagnosis?

A 34-year-old male with new onset syncopal episodes: Cardiac MRI

MRI LVOT view



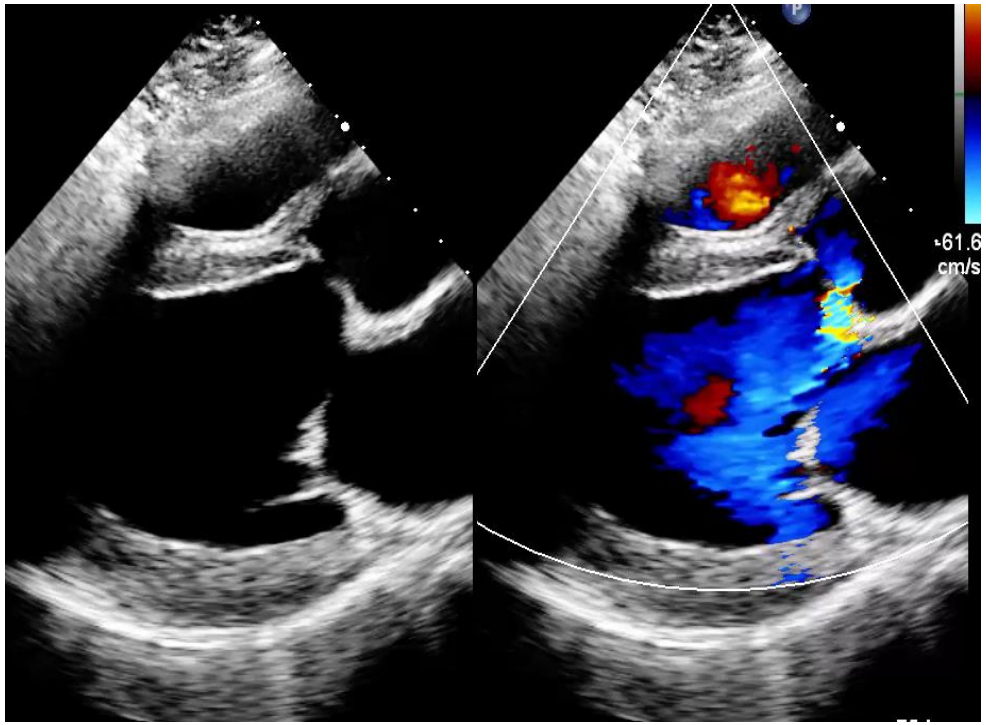
MRI: RVOT view

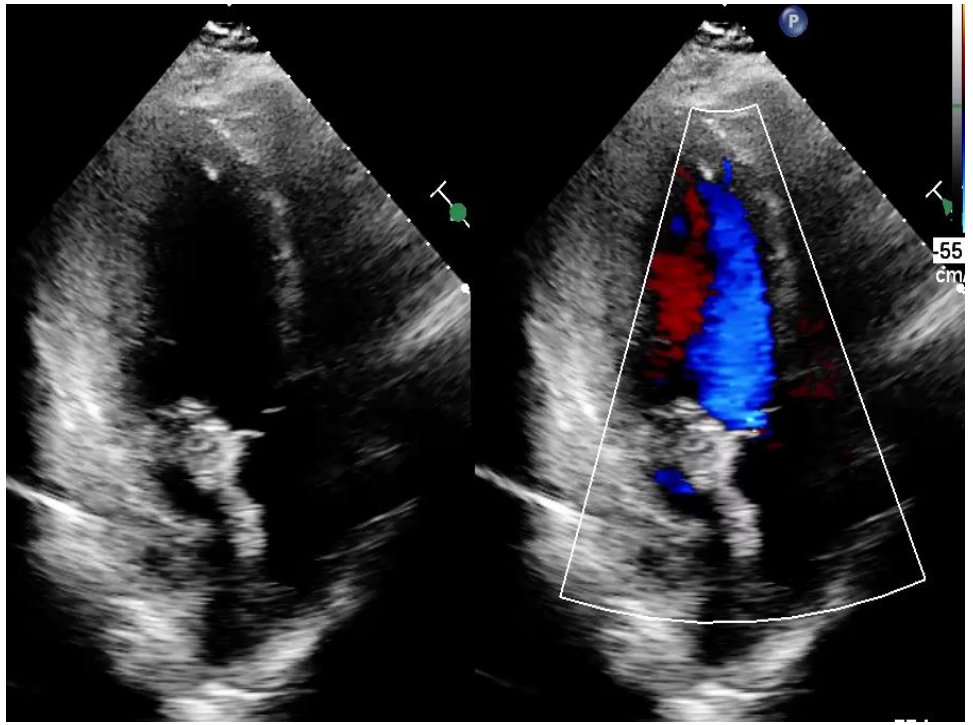


Ventricular septal defect with advancing age

- A double-chambered RV (DCRV) can develop over time, mostly in perimembranous defects

A 54-year-old female with aortic valve regurgitation





Ventricular septal defect…..

- Risk of aortic valve prolapse and progressive aortic valve regurgitation

- Subarterial (supracristal) VSD or less commonly perimembranous

Ventricular septal defect….

- Ventricular septal defect is one of the most common congenital heart defects
- Most small VSDs close spontaneously
- Most patients with small unrepaired, asymptomatic, uncomplicated VSDs require no intervention
- But at risk of development of double-chambered right ventricle or aortic regurgitation
- Require periodic follow-up

Patent ductus arteriosus

- In adults, it is usually an isolated finding
- Adult patients with a large PDA have, in general, developed Eisenmenger physiology
- Differential hypoxemia and differential cyanosis (lower extremities cyanotic, sometimes left arm too)
- Aneurysm formation of the duct is a very rare complication

PDA: Management in adults

- In adults, calcification of the PDA may cause a problem for surgical closure.

- Device closure is the method of choice

- Surgery is reserved for the rare patient with a duct too large for device closure or with unsuitable anatomy such as aneurysm formation.

Anomalous pulmonary venous connections

- Usually associated with superior sinus venosus ASD
- Can be an isolated lesion
- Physiological effect similar to that of an ASD
- No potential for right to left shunting

Turner syndrome

- Partial anomalous pulmonary venous return (PAPVR): 25% of the cases
- Our experience

-Retrospective review of 21 young adults with Turners syndrome who had cardiac CTA or CMR at our center between 2017- 2022

-PAPVR was present in 6 of 21 patients (29%)

-New diagnosis of PAPVR was made in 4 subjects on retrospective review (which was missed at initial evaluation)

Learning point:

- Partial anomalous pulmonary venous return is common in adults with Turners syndrome

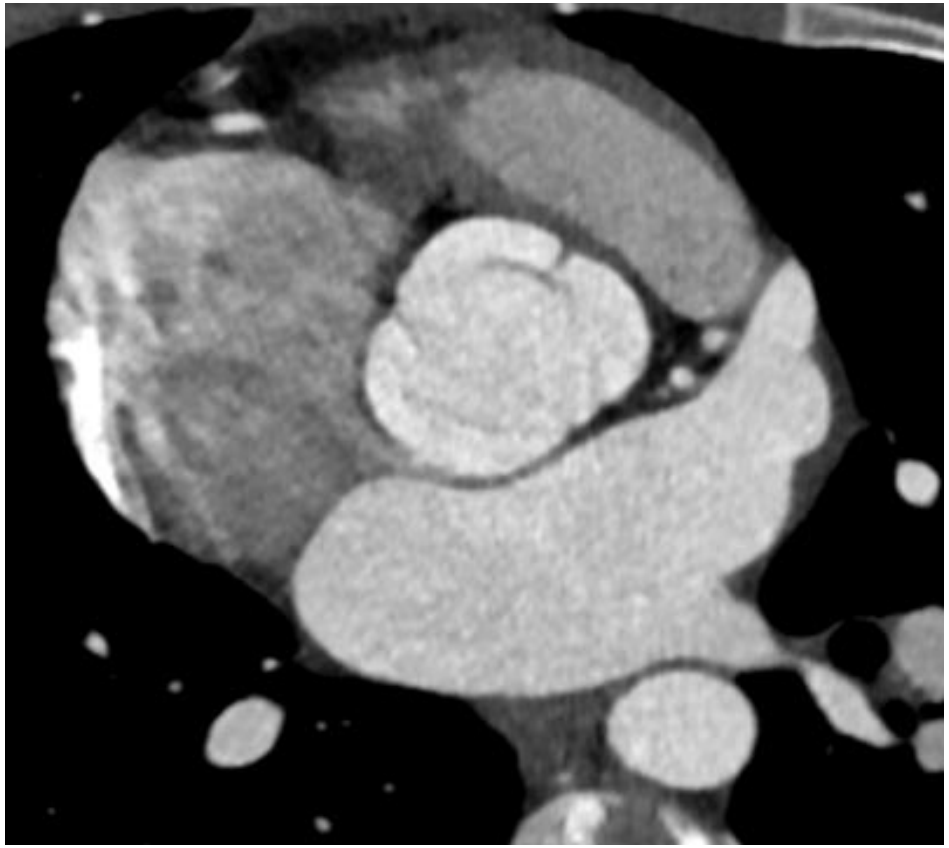
Coarctation of the aorta (CoA)

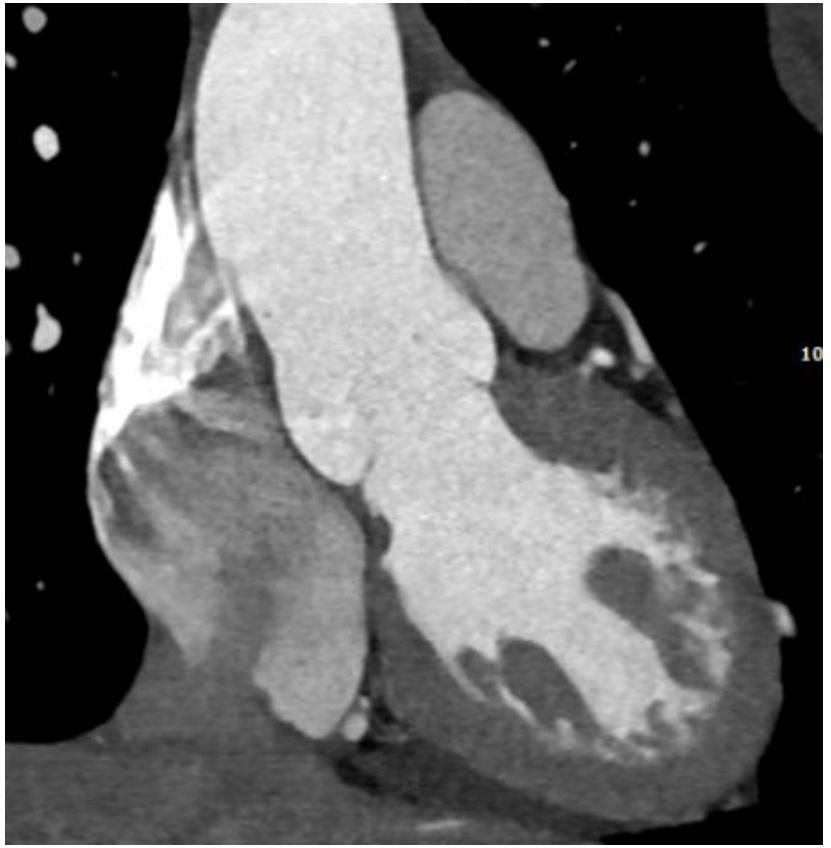
- Mild coarctation may not become evident until adulthood, where CoA is detected in the work-up of arterial hypertension
- Office blood pressure measurement in the upper and lower extremities are the primary studies required in all coarctation patients
- A blood pressure gradient between upper and lower extremities (systolic ≥20 mmHg) indicates significant CoA

DX and Management: CoA

- Echocardiography: first line
- CMR and Cardiac CT: preferred non-invasive techniques
- Preferred pediatric surgical techniques: resection and end-to-end anastomosis, or extended end-to-end anastomosis
- Generally feasible techniques in adults: interposition of a tube graft or bypass tube (jump) grafts.
- Ascending-to-descending aorta conduits (in the setting of difficult anatomy)

Bicuspid aortic valve





Bicuspid aortic valve (BAV)

- About 1% of general population
- Familial occurrence: 5–10% in first-degree relatives
- 20–84% of patients with a BAV develop ascending aortic dilatation
- Much lower risk of dissection than in Marfan syndrome but higher than general population (x8)
- CoA is associated with an increased risk for dissection

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Symptomatic 45 year old male

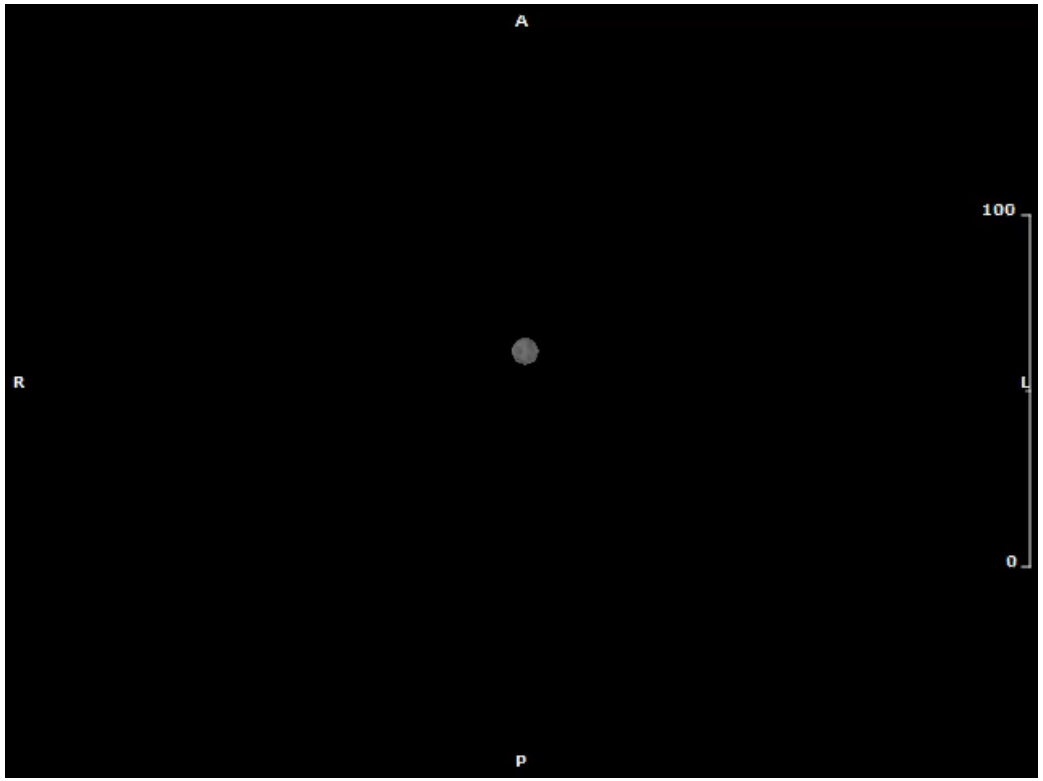
- Known history of diabetes mellitus
- Systemic hypertension
- Chest pain
- Shortness of breath
- Leg edema

Differentials???

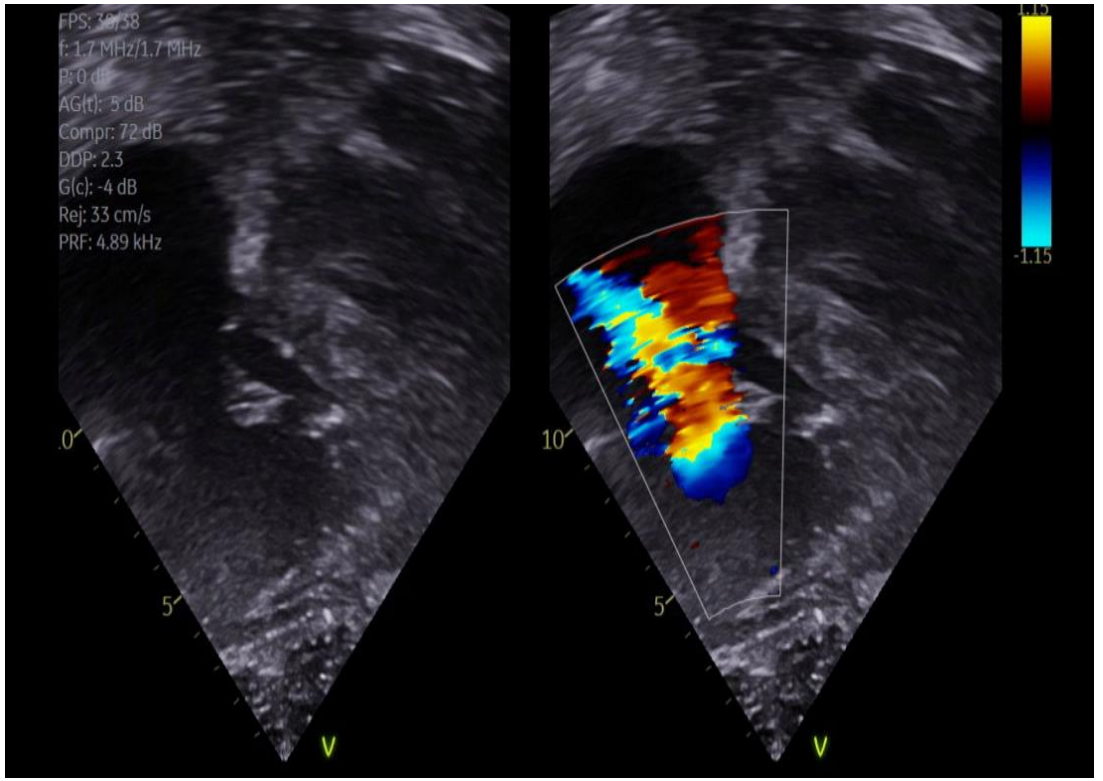
- Cardiac CT to evaluate coronary arteries

A 45 year old male with diabetes mellitus

Axial CT

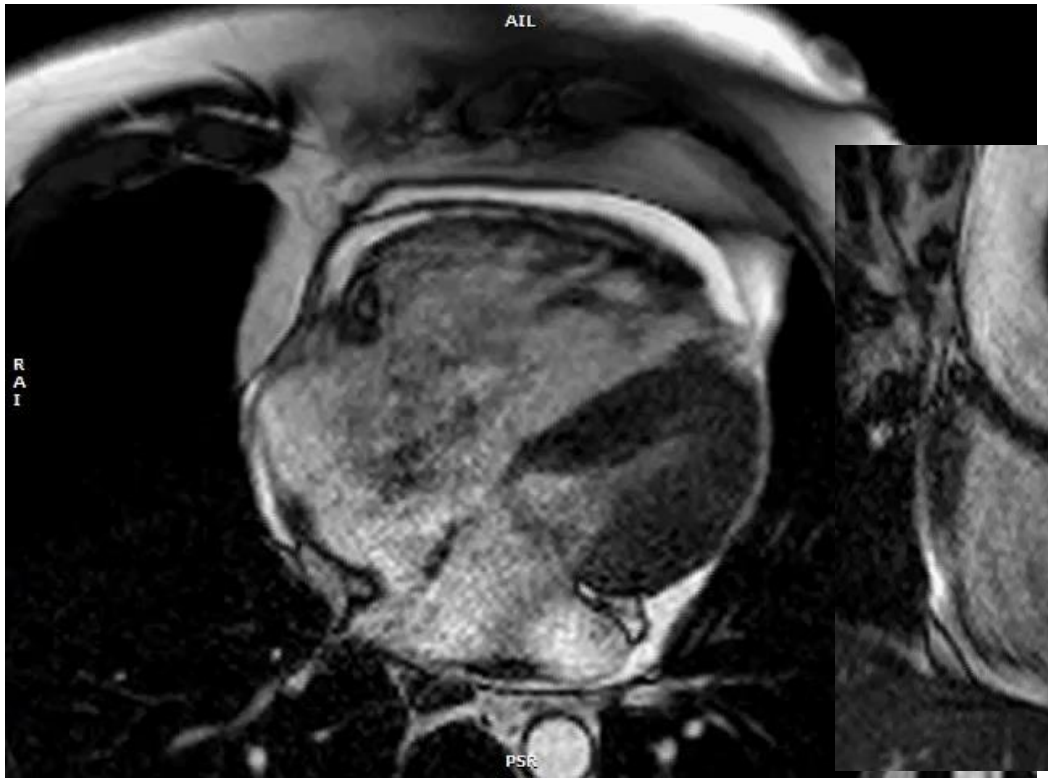


Apical view echo: Poor acoustic window

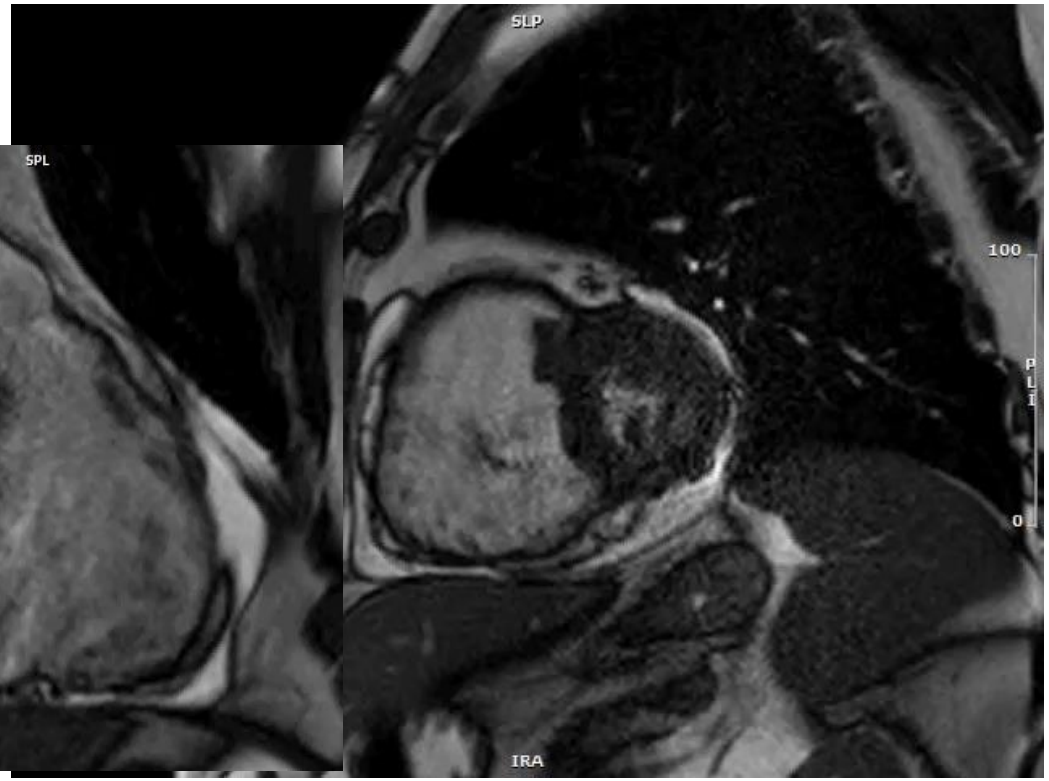


A 45 year old male with shortness of breath…

Cardiac MRU 4CH



Cardiac MRI RV in-out and SA



Ebstein anomaly

- Ebstein anomaly is characterized by abnormally formed and apically displaced leaflets of the tricuspid valve (TV)

- Septal and posterior leaflets are displaced towards the RV apex and often tethered to the endocardium
- Hemodynamic changes depend on the severity of the TV dysfunction

Ebstein anomaly: Clinical presentation and natural history

- Patients with mild forms can be asymptomatic over decades until they are diagnosed
- Significant TR: RV failure, liver cirrhosis, cerebral abscesses, paradoxical embolism, pulmonary embolism, tachyarrhythmias or SCD
- Atrioventricular reentrant tachycardia: catheter ablation before surgery

Management of Ebstein anomaly

- Tricuspid valve repair, if feasible, is preferred over TV replacement
- If the RV is too small for repair or RV dysfunction has developed, an additional bidirectional cavopulmonary (Glenn) anastomosis may be considered in adults with preserved LV function when LA pressure and LV end diastolic pressure are not elevated
- In patients with failed repair, or in severe biventricular dysfunction, heart transplantation may be the only option

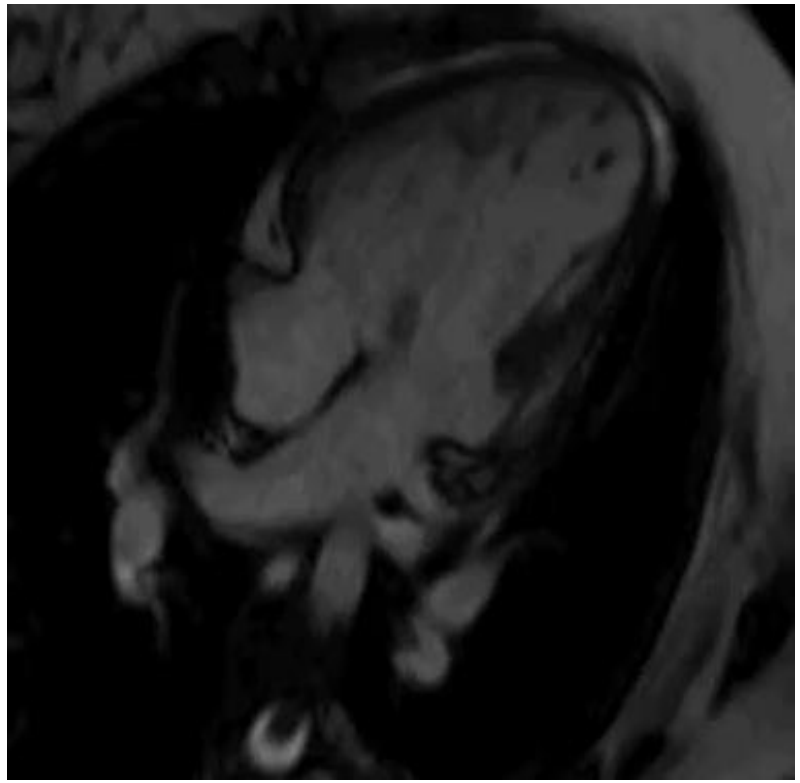
Congenitally corrected transposition of the great arteries (ccTGA)

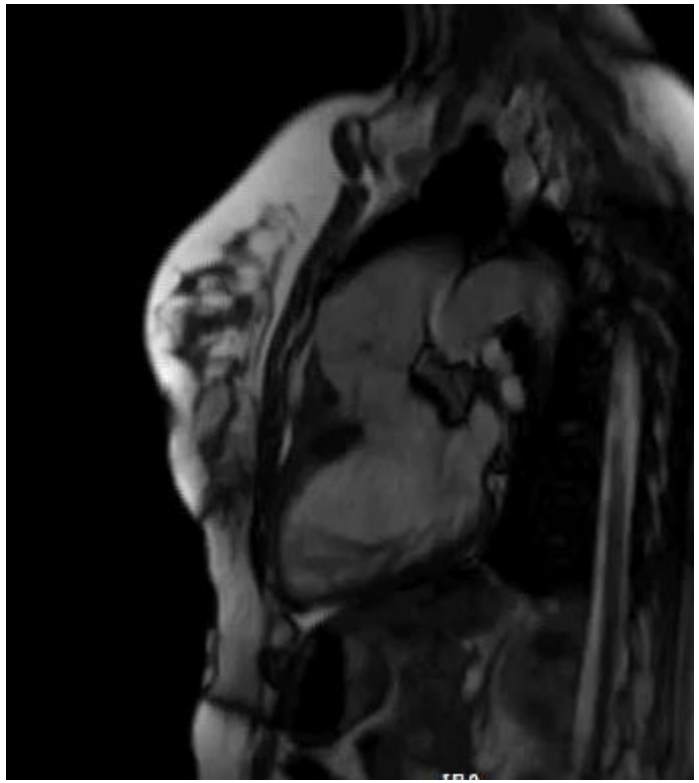
- Double discordance may be present in hearts with usual (95%) or mirror-image atrial arrangement (5%)
- Patients with isolated ccTGA rarely develop complications before adulthood
- Late complications: RV failure, Systemic TR, complete AV block (2% loss of AV conduction per year
- Without associated lesions, ~50% of patients were alive at the age of 60 years

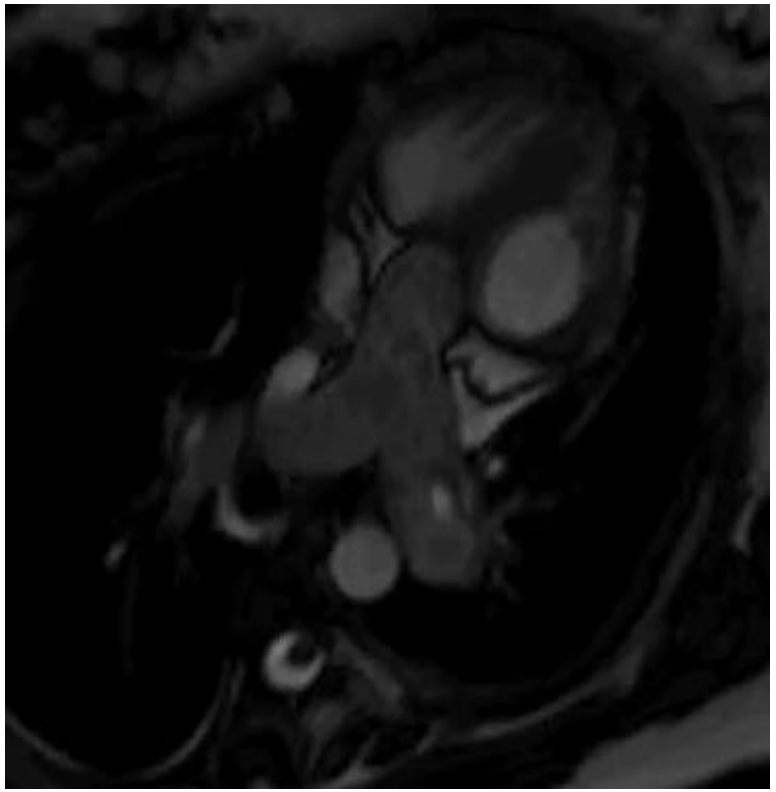
An 18-year-old female recently immigrated to United States

- Unremarkable past medical history
- New onset shortness of breath for 1 month
- Oxygen saturation 85% on room air

Case example: Cardiac MRI for further evaluation Qp:QS is 1.2:1. What would you do?







Univentricular heart with balanced circulation

- Double inlet left ventricle with pulmonary stenosis resulting adequate restriction
- The balanced circulation allows survival into adulthood without having surgery
- In rare cases, with a well-balanced hemodynamic situation, survival until the fifth, sixth, and even seventh decade has been reported

Coronary anomalies

- Anomalous aortic origin of a coronary artery (AAOCA)
- Anomalous coronary artery from the PA (ACAPA)
- Coronary fistula

Anomalous aortic origin of a coronary artery

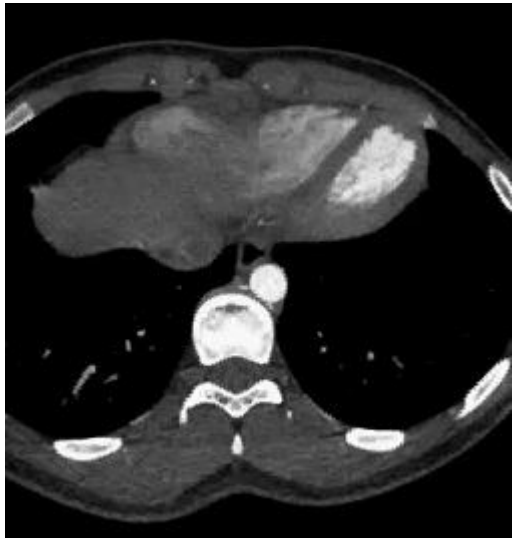
- Most patients are young (<35 years) and die during, or shortly after, exercise

- Left coronary artery arising from the opposite (right) sinus is less common, but more malignant than the right coronary artery from the left sinus.

- Ostial stenosis, slit-like/fish-mouth-shaped orifice, inter-arterial and intramural course and hypoplasia of the proximal coronary artery: are high risk factors

DX and management: Coronary anomalies

- Cardiac CT
- Stress MRI
- Exercise stress test
- Surgical reimplantation: symptomatic/myocardial ischemia or high risk anatomy



Thank you for your attention!

- Any question?