



Adult Congenital Heart Disease: What the Internist Needs to Know

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Epidemiology

- ▶ 1% of babies are born with congenital heart disease
- ▶ >90% of babies with CHD now survive to adulthood
- ▶ Now more adults than children living with CHD
- ▶ Patients with CHD require LIFELONG CARE
 - ▶ Palliative vs corrective surgeries
 - ▶ Long-term complications/sequelae of repairs

Objectives for Today

- ▶ Learn to identify ACHD commonly diagnosed for the first time in adults
- ▶ Identify important follow up requirements for previously diagnosed ACHD
- ▶ Discuss pregnancy planning in your patients with ACHD

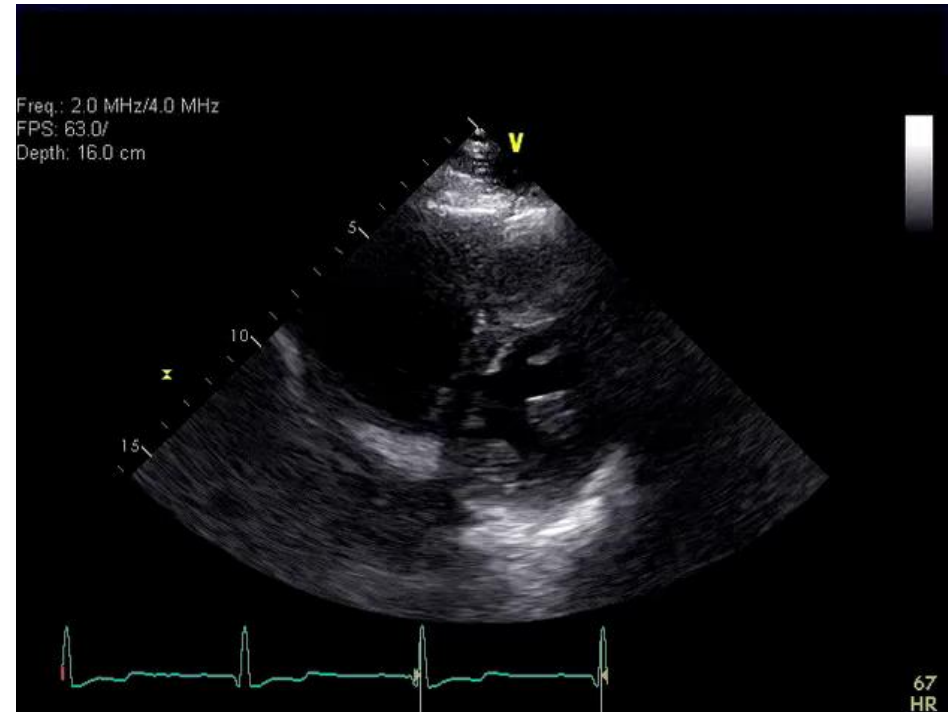
Making a New Diagnosis of ACHD



- ▶ Some congenital disorders may not be diagnosed until adulthood
- ▶ Know which congenital heart defects may go undetected
- ▶ Have high index of suspicion
- ▶ Order the right tests
- ▶ Refer to the right people

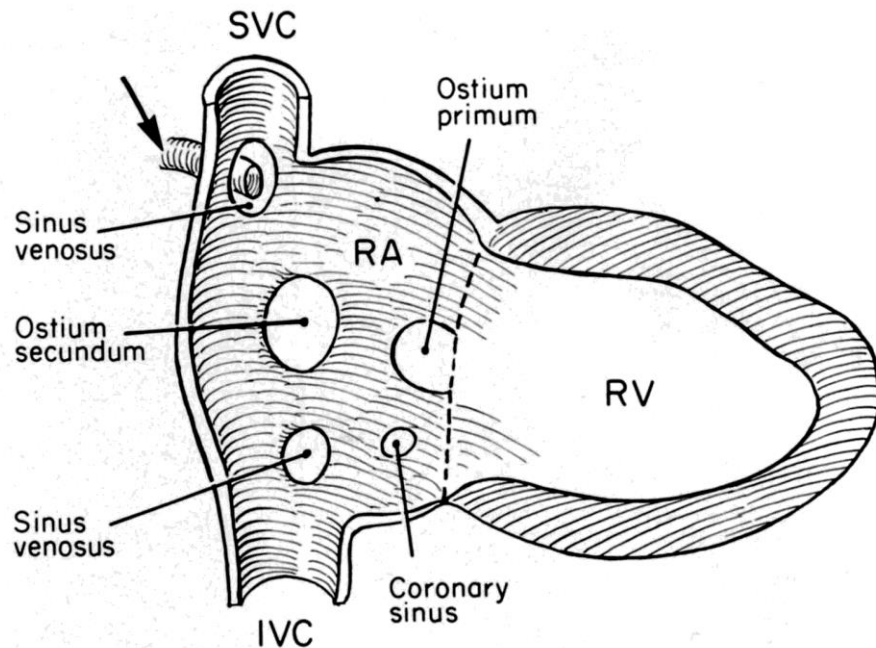
Case #1

- ▶ 42 year old woman presents to your office complaining of 6 months of progressive dyspnea on exertion and intermittent palpitations. A transthoracic echocardiogram reveals the following:
- ▶ What undiagnosed ACHD should be considered, and how can we distinguish between the two?



Isolated RV enlargement: DDx

Atrial Septal Defect



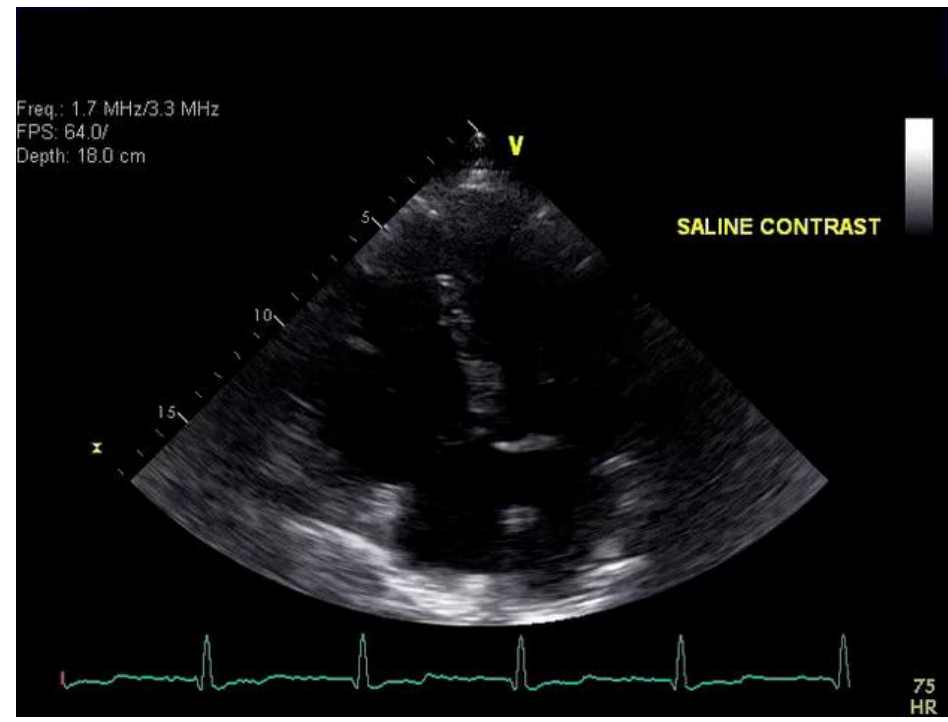
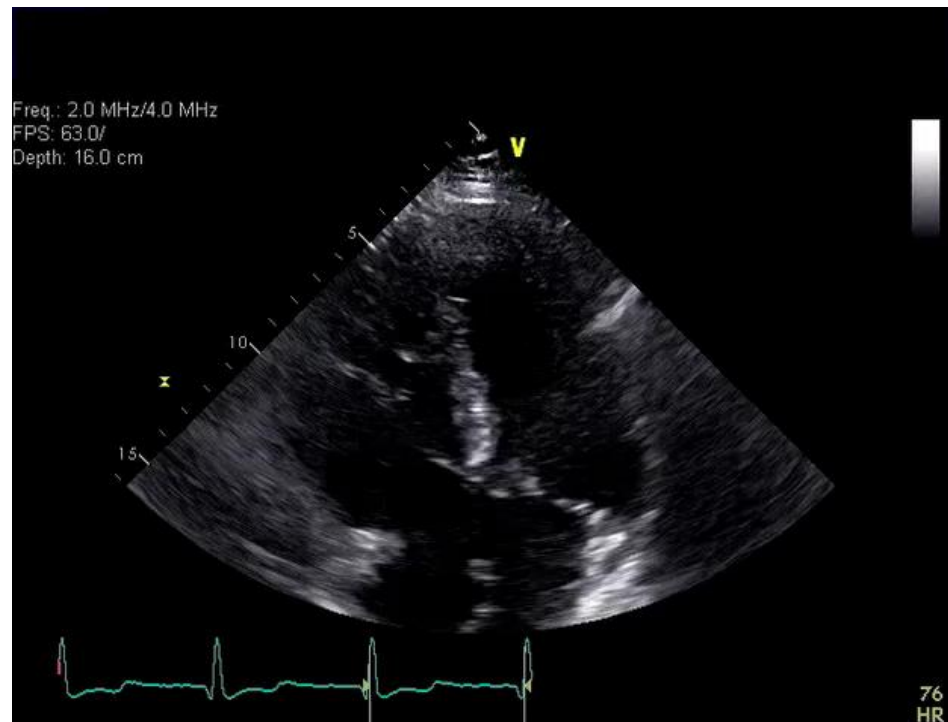
Partial Anomalous Pulmonary Venous Return



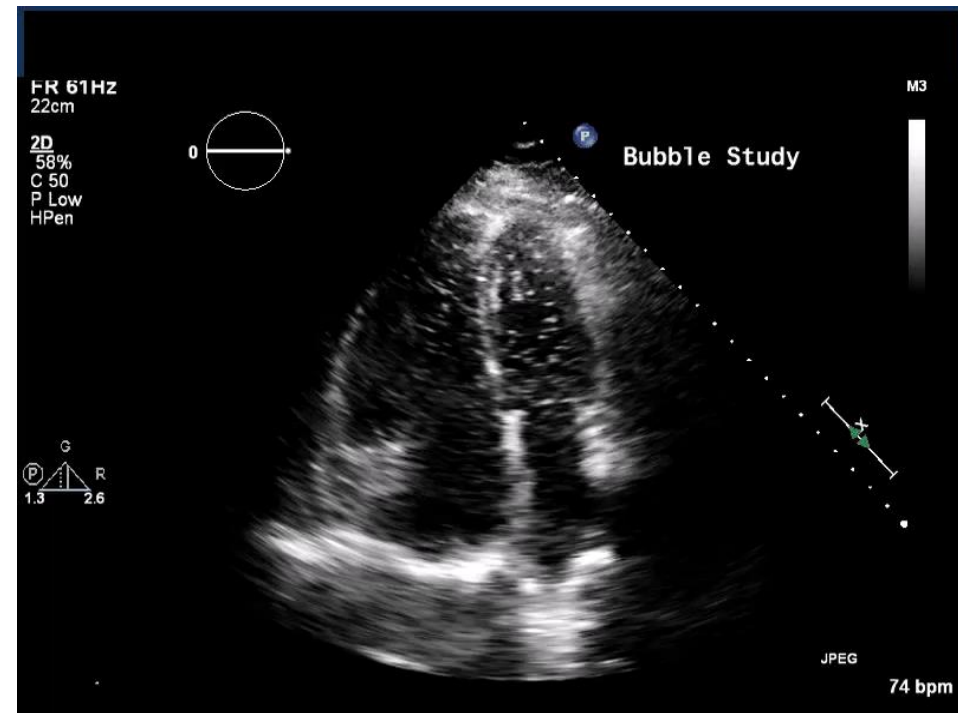
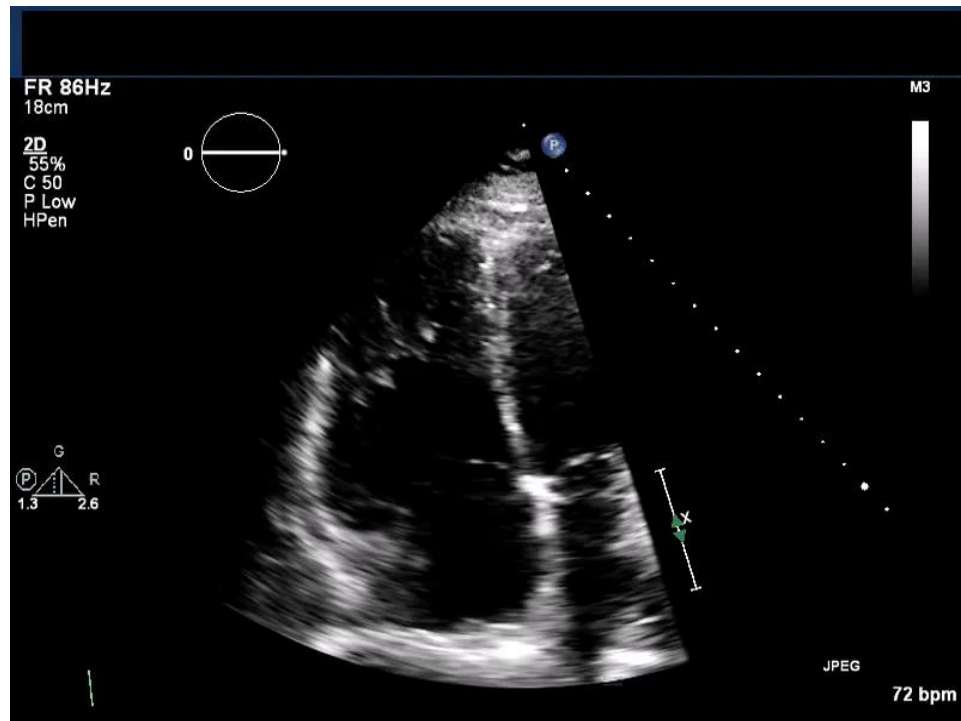
Atrial Septal Defect/PAPVR

- ▶ Left to Right Shunt
 - ▶ RV volume overload → dilated right atrium and right ventricle
 - ▶ Symptoms related to RV volume overload
 - ▶ Dyspnea
 - ▶ Atrial arrhythmias
 - ▶ Fatigue
 - ▶ Can contribute to pulmonary arterial hypertension
 - ▶ Indications for closure:
 - ▶ Evidence of right sided enlargement
 - ▶ $Q_p:Q_s > 1.5:1$
 - ▶ CONTRAINDICATED in setting of irreversible pulmonary hypertension

How to distinguish between ASD and PAPVR?



How to distinguish between ASD and PAPVR?



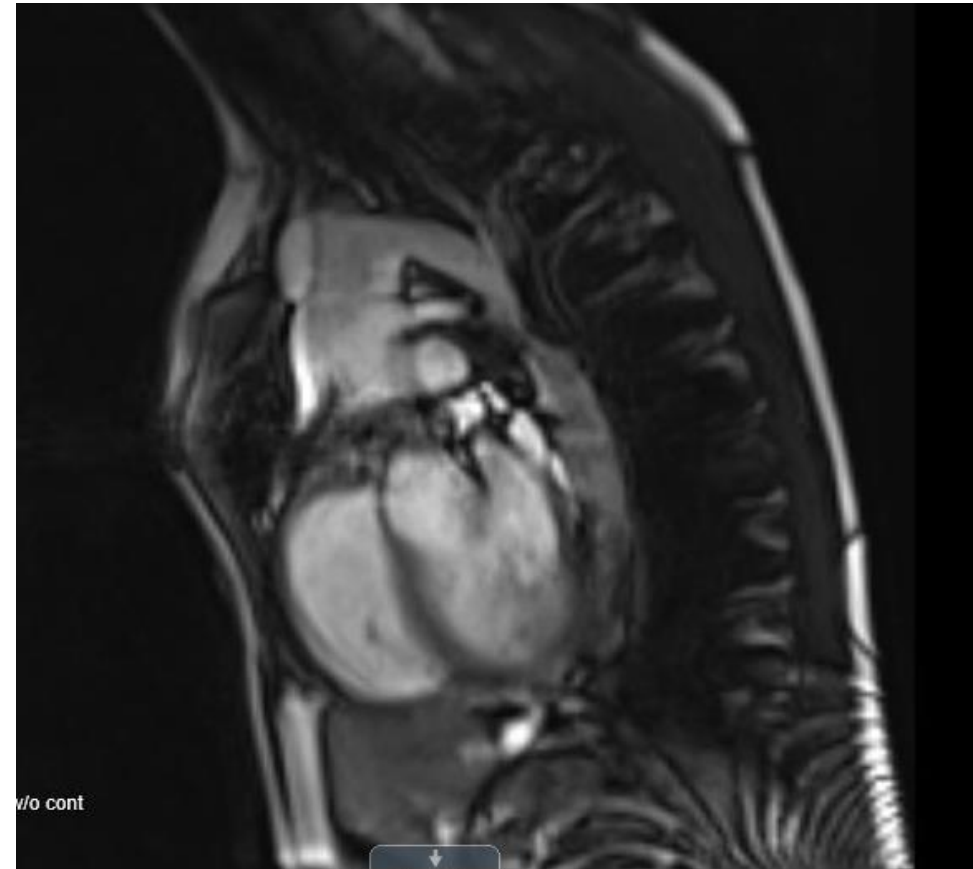
Case #2

- ▶ 38 year old man presents with difficult to manage hypertension. His blood pressure remains above goal despite treatment with 3 agents. The patient reports medication and dietary compliance. He reports he was first diagnosed with hypertension in his 20s.
- ▶ What ACHD diagnosis should be considered?



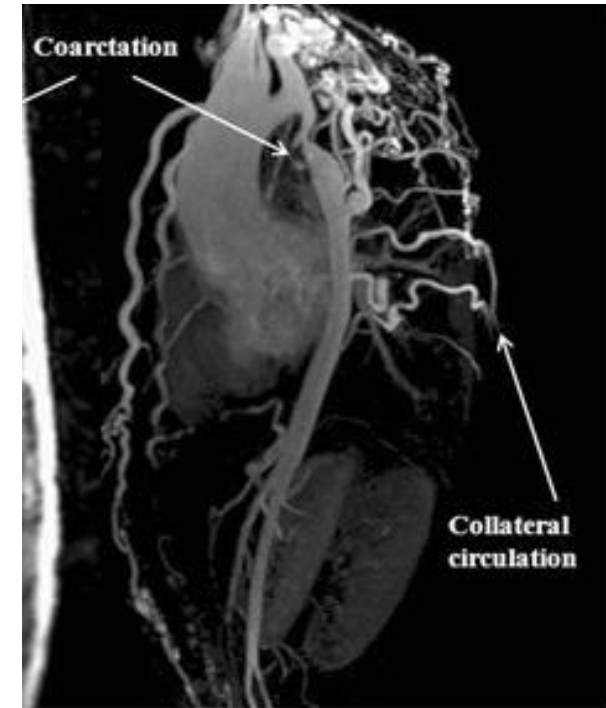
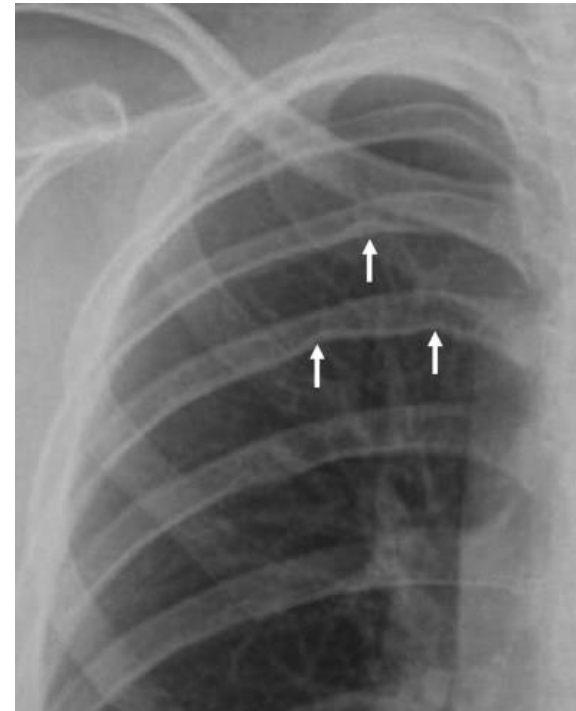
Coarctation of the Aorta

- ▶ Physical exam:
 - ▶ Upper extremity hypertension
 - ▶ Brachial-femoral delay
 - ▶ Reduced lower extremity pulses
- ▶ Associated Symptoms:
 - ▶ Exertional headaches
 - ▶ Leg claudication
- ▶ Associated Lesions:
 - ▶ Bicuspid AV
 - ▶ Aortic aneurysm
 - ▶ BERRY ANEURYSM



Treating Coarctation: When and How?

- ▶ When?
 - ▶ Peak to peak gradient >20 mmHg
 - ▶ Peak to peak gradient <20 mmHg if significant imaging evidence of collateral blood flow
- ▶ How?
 - ▶ Discrete stenoses often treated via stenting
 - ▶ Long segments or associated arch hypoplasia \rightarrow surgical repair
 - ▶ Should be performed in consultation with an ACHD center with experienced interventionalists and congenital surgeons

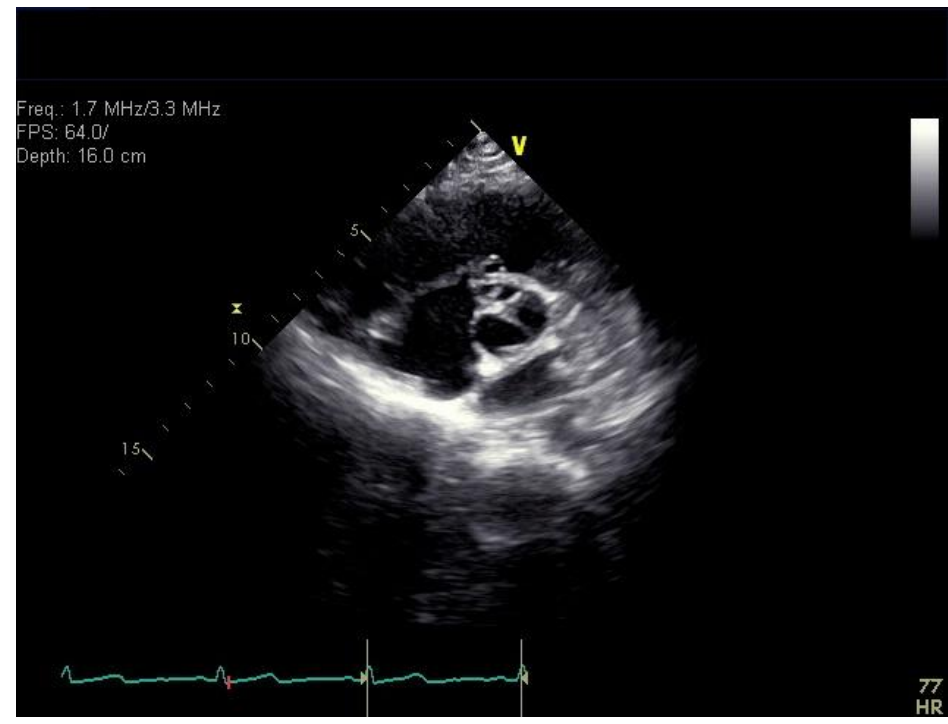
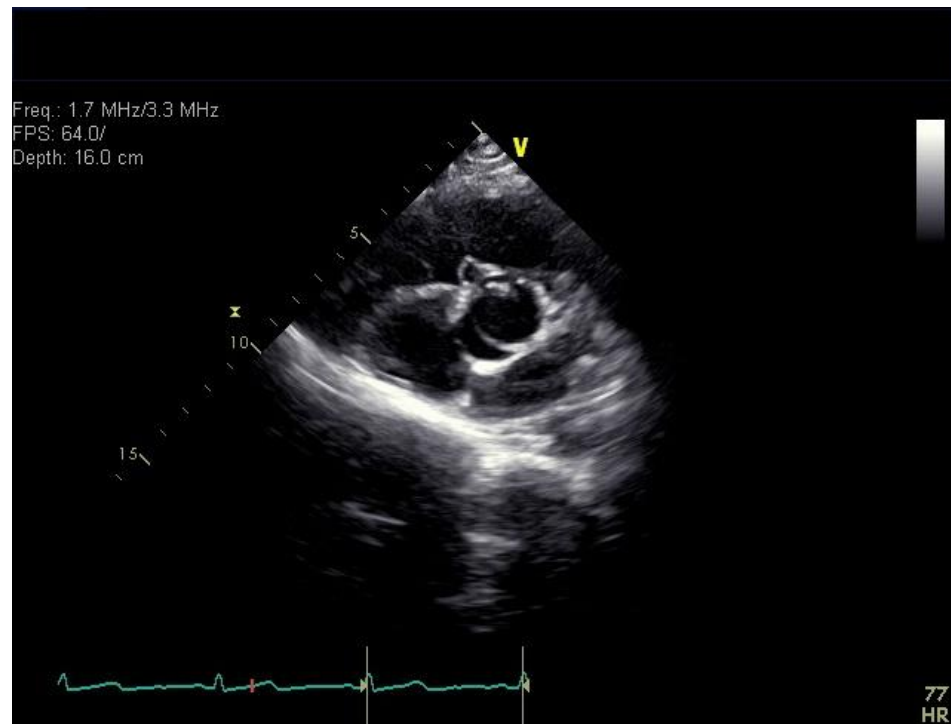


Case #3

- ▶ 48 year old asymptomatic male presents to your office for an annual physical exam. A mid-peaking crescendo-decrescendo murmur is heard at the right upper sternal border. The patient reports that his father died suddenly in his 50s.
- ▶ What ACHD diagnosis should you consider?



Bicuspid Aortic Valve



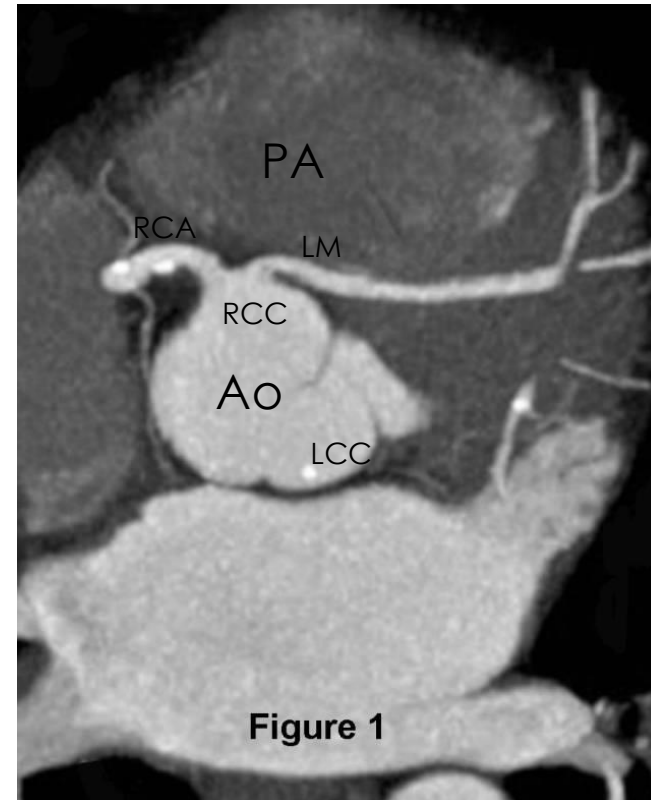
Bicuspid Aortic Valve

- ▶ Most common congenital heart defect
- ▶ 2:1 male:female predominance
- ▶ 10% of first degree relatives are affected
 - ▶ Screen all first degree family members
- ▶ Associated with aortopathy
- ▶ Surgery indicated for:
 - ▶ Severe AS or AI
 - ▶ Aortic enlargement >5.0 cm



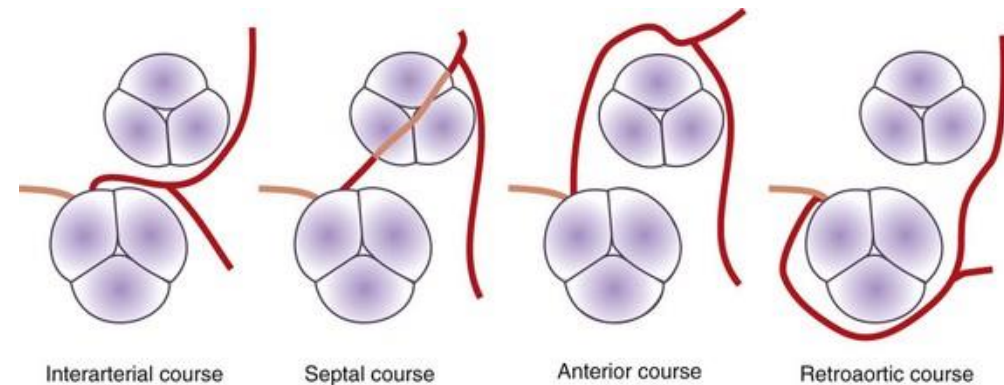
Case #4

- ▶ A 22 year old college soccer player complains of chest pain with her pre-season training. She has no cardiac risk factors and no family history of premature coronary artery disease. She is on no medications and takes no supplements.
- ▶ A treadmill exercise stress test was performed. At peak stress, she developed chest pain and had diffuse ST depressions on EKG.
- ▶ What ACHD diagnosis should you consider?



Anomalous Coronary Artery

- ▶ Consider in survivors of sudden death, life threatening arrhythmias, or young patients with ischemic symptoms
- ▶ CTA or MRA is ideal for screening
- ▶ Surgery for high risk lesions:
 - ▶ Left main or LAD from R coronary cusp with INTERARTERIAL course
 - ▶ Documented ischemia from any vessel coursing between the aorta and pulmonary artery
 - ▶ May also be reasonable if coronary compression or reduced blood flow is documented via IVUS, FFR





“I had a hole in my heart but it was fixed.”

- ▶ Many patients do not realize that they remain at risk of complications from their CHD
- ▶ Significant gap in care after transition from pediatric cardiologist in 65% of patients
- ▶ Patients with SIMPLE CHD should be seen at least ONCE by an ACHD physician to formulate future follow up needs
- ▶ Patients with MODERATE OR COMPLEX CHD should be followed by an ACHD specialist at least every 12-24 months

Simple Congenital Defects

- ▶ Unrepaired:
 - ▶ Isolated aortic valve disease
 - ▶ Bicuspid AV
 - ▶ Isolated PFO or small ASD
 - ▶ Isolated small VSD (without associated lesions)
 - ▶ Mild Pulmonic stenosis
 - ▶ Small PDA
- ▶ Repaired:
 - ▶ Ligated/Occluded PDA
 - ▶ Repaired secundum or sinus venosus ASD without residua
 - ▶ Repaired VSD without residua

Moderate Congenital Defects

- ▶ Anomalous pulmonary venous return
- ▶ Atrioventricular septal defect (partial or complete)
- ▶ Coarctation of the Aorta
- ▶ Ebstein Anomaly
- ▶ Ostium primum ASD
- ▶ PDA (not closed)
- ▶ Pulmonary valve disease (mod to severe)
- ▶ Sinus venosus ASD
- ▶ Subvalvular or supra-ventricular aortic stenosis
- ▶ Tetralogy of Fallot
- ▶ VSD with associated lesions

Complex Congenital Defects

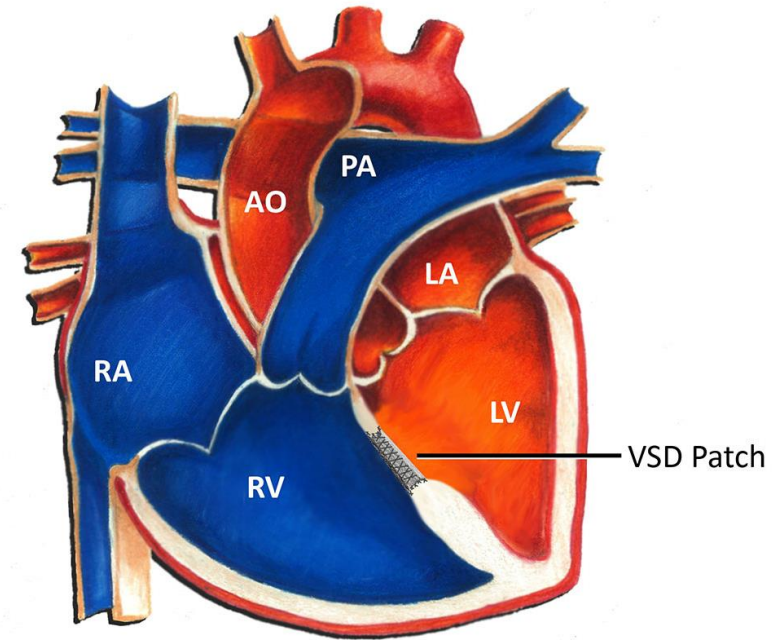
- ▶ All conduits
- ▶ All forms of cyanotic heart disease
- ▶ Double outlet ventricle
- ▶ Eisenmenger Procedure
- ▶ Mitral atresia
- ▶ Fontan procedure
- ▶ Single ventricle
- ▶ Pulmonary atresia
- ▶ Transposition of the Great Arteries
- ▶ Tricuspid Atresia
- ▶ Truncus Arteriosus

Atrial Septal Defect

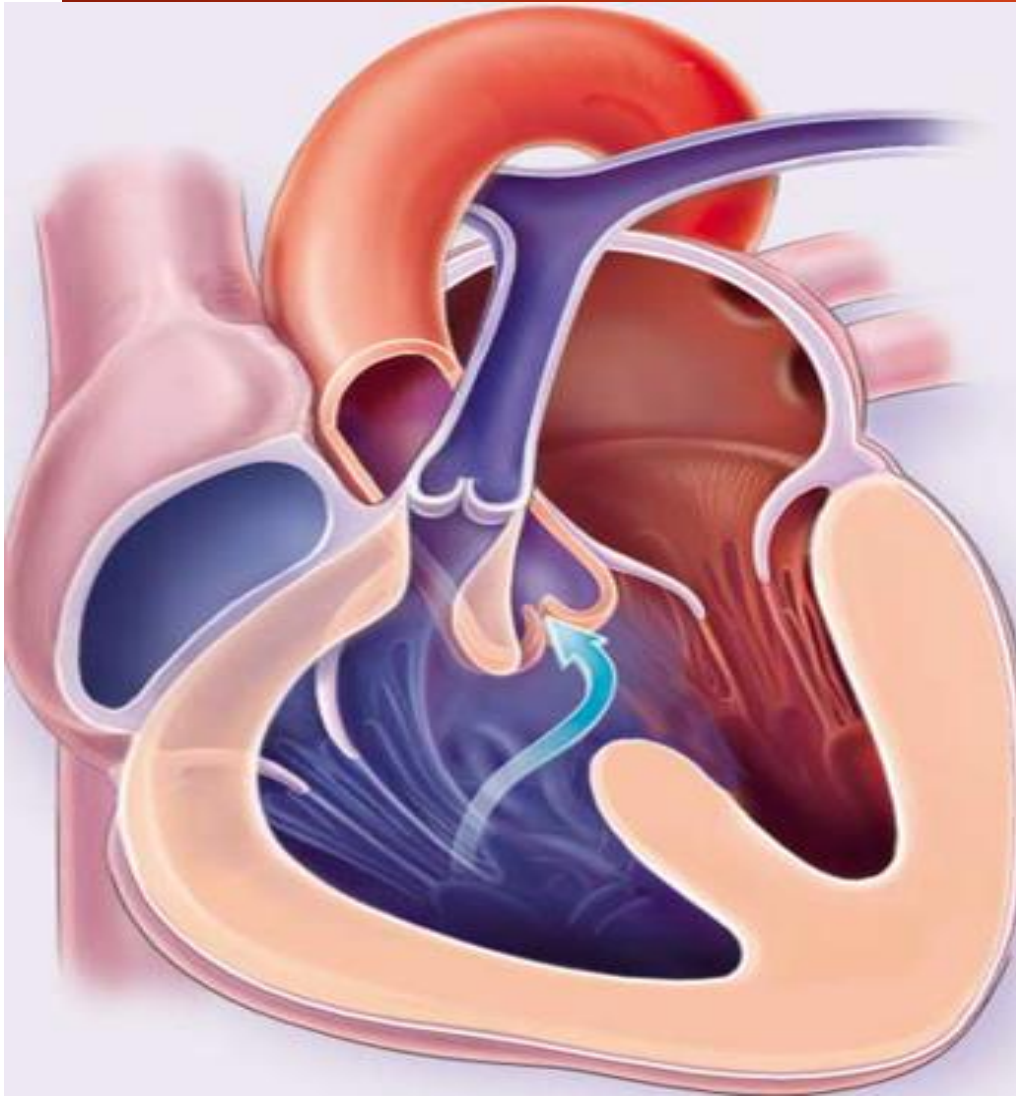
- ▶ Most patients will do well, especially if closed early
- ▶ Pulmonary arterial hypertension can develop after closure of ASD, even if ASD was closed early
 - ▶ Much more common if closed after age 25
 - ▶ Lifelong annual follow up recommended for ASDs closed in adults
- ▶ Consider pulmonary hypertension if exercise tolerance, dyspnea, or right heart failure symptoms develop in patient with prior ASD

Ventricular Septal Defect

- ▶ VSD may be large (NON-restrictive) or small (restrictive)
- ▶ Restrictive VSD is noisy (pansystolic murmur), high velocity, low volume jet
 - ▶ No hemodynamic consequence
 - ▶ No need to repair – innocent bystander
- ▶ Non-restrictive VSD will have been repaired
 - ▶ Unrepaired will lead to left ventricular volume overload and/or Eisenmenger Syndrome
 - ▶ Post-repair, there may be a residual PATCH LEAK
 - ▶ If no evidence of LV volume overload, can watch



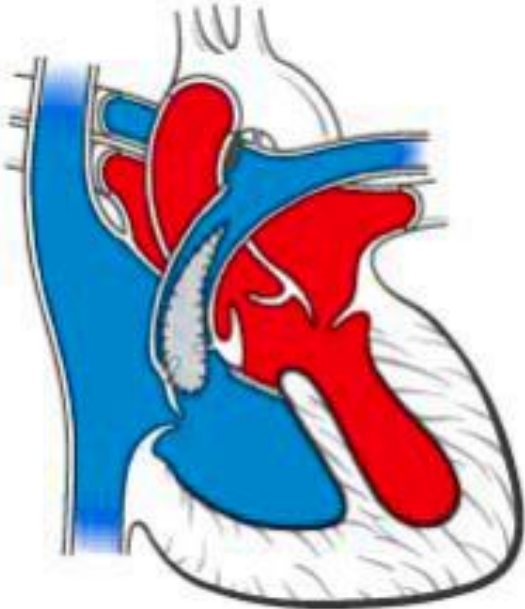
Tetralogy of Fallot



- ▶ Anterior Malalignment VSD
- ▶ Overriding Aorta
- ▶ RVOT Obstruction
- ▶ RVH
- ▶ ASSOCIATED LESIONS
 - ▶ 25% Right Sided Aortic Arch
 - ▶ 10% ASD ("Pentalogy")
 - ▶ 10% Coronary Anomalies

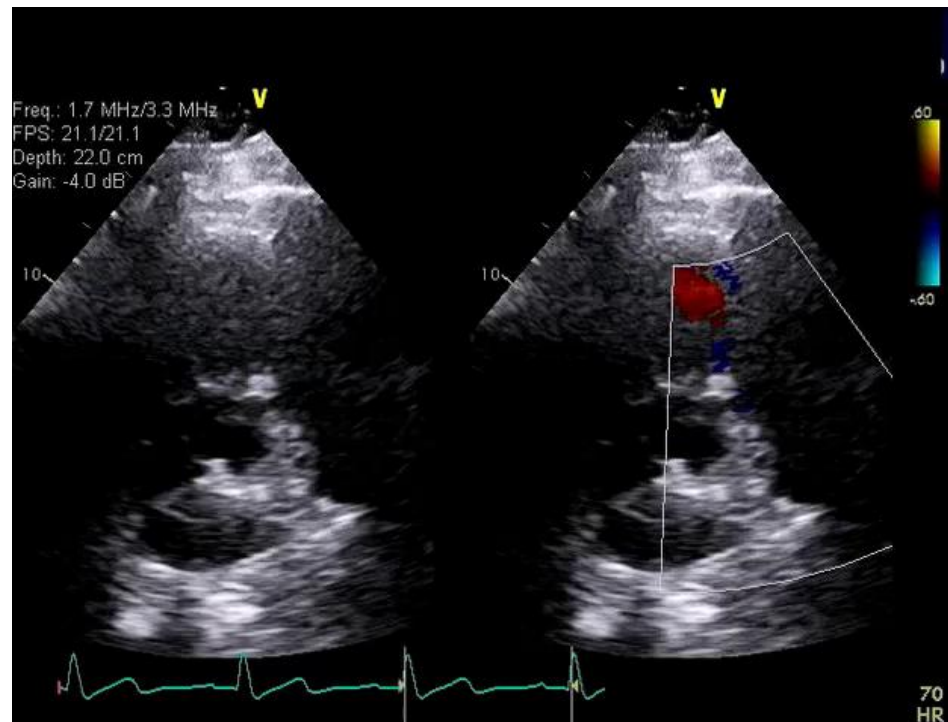
Repair and Late Complications

Tetralogy of Fallot - Repair of small PV Tet

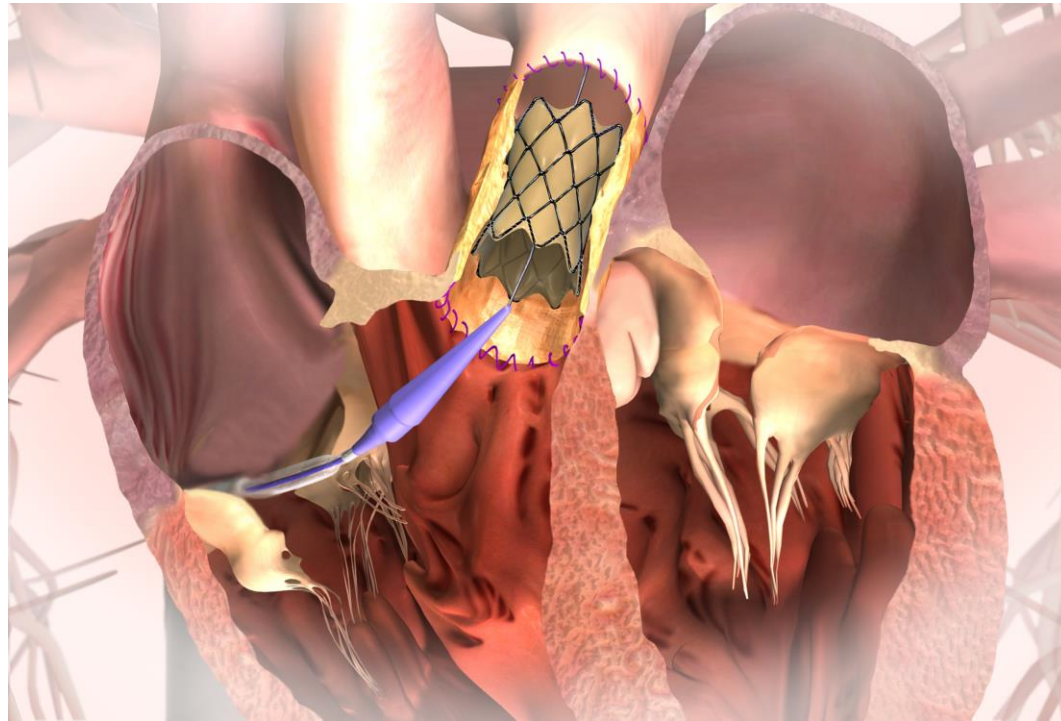


- ▶ Pulmonary Insufficiency
- ▶ Pulmonary Insufficiency
- ▶ Pulmonary Insufficiency
- ▶ Pulmonary Stenosis
- ▶ Ventricular Arrhythmias
- ▶ Left Ventricular Dysfunction

Pulmonary Insufficiency



Percutaneous Pulmonary Valve



Coarctation of the Aorta

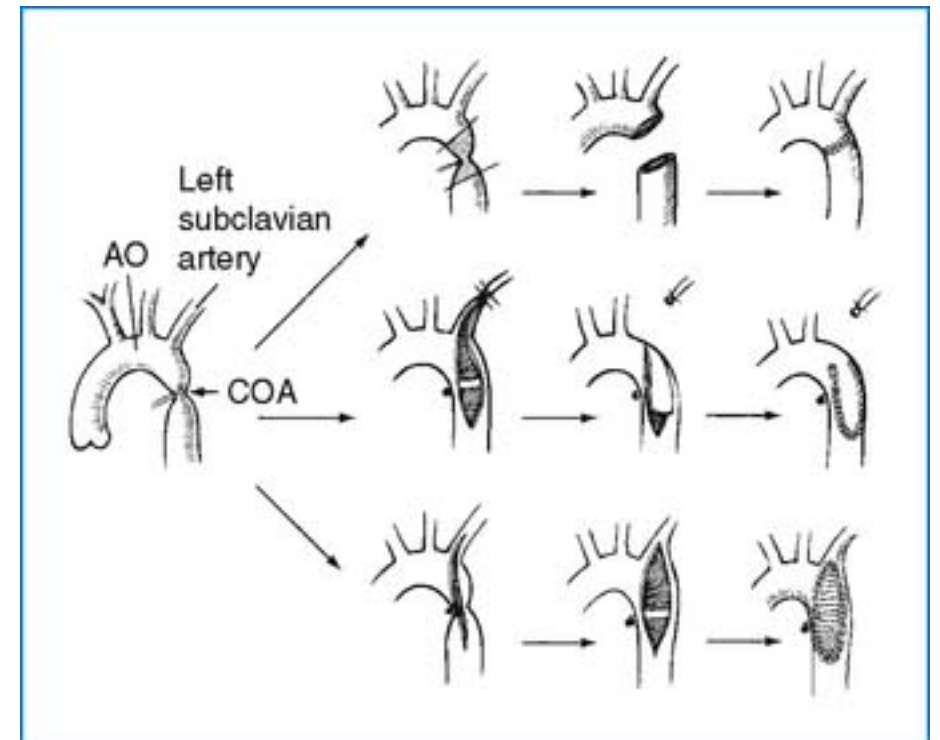
- ▶ 26 year old man with history of coarctation repair has complaints of exertional chest pain.
- ▶ Resting blood pressure is 133/77 on the right and 108/58 on the left.
- ▶ Baseline echocardiogram shows a 16 mmHg gradient across the coarctation repair site
- ▶ Treadmill stress testing shows no evidence of ischemia, but peak blood pressure 225/118.

Coarctation of the Aorta: Hypertension

- ▶ Hypertension may occur via several mechanisms
 - ▶ Re-coarctation at site of prior repair
 - ▶ >20 mmHg gradient considered clinically significant
 - ▶ Evaluation by MRI or CT is recommended every 5 years or less
 - ▶ Even in the presence of a good repair, chronic hypertension may develop
 - ▶ Close BP monitoring is essential
 - ▶ Exercise-induced hypertension may be present with normal resting BPs
 - ▶ Periodic exercise testing is recommended

Coarctation: Aortopathy

- ▶ Re-stenosis, aortic dilatation, and aneurysm formation may all occur
 - ▶ Periodic MRI/CT imaging
 - ▶ Aortic tissue is not “normal” – this is an aortopathy
- ▶ Be aware of BP differential due to repair method
- ▶ Lifelong follow up with cardiology, including consultation with ACHD cardiologist, is recommended for all patients with coarctation



SBE Antibiotic Prophylaxis: Who, What, When, Where, Why?

▶ Who:

- ▶ Prosthetic heart valves
- ▶ Prior endocarditis
- ▶ Heart transplant patients with valve regurgitation due to structurally abnormal valve
- ▶ Congenital heart disease with
 - ▶ UNREPAIRED CYANOTIC CHD (Includes palliative shunts/conduits)
 - ▶ Repaired CHD with prosthetic material within the first 6 months of the procedure
 - ▶ Repaired CHD with RESIDUAL DEFECTS at the site of a prosthetic patch/device



"Yes, I floss regularly — right before every appointment."

SBE Antibiotic Prophylaxis: Who, What, When, Where, Why?

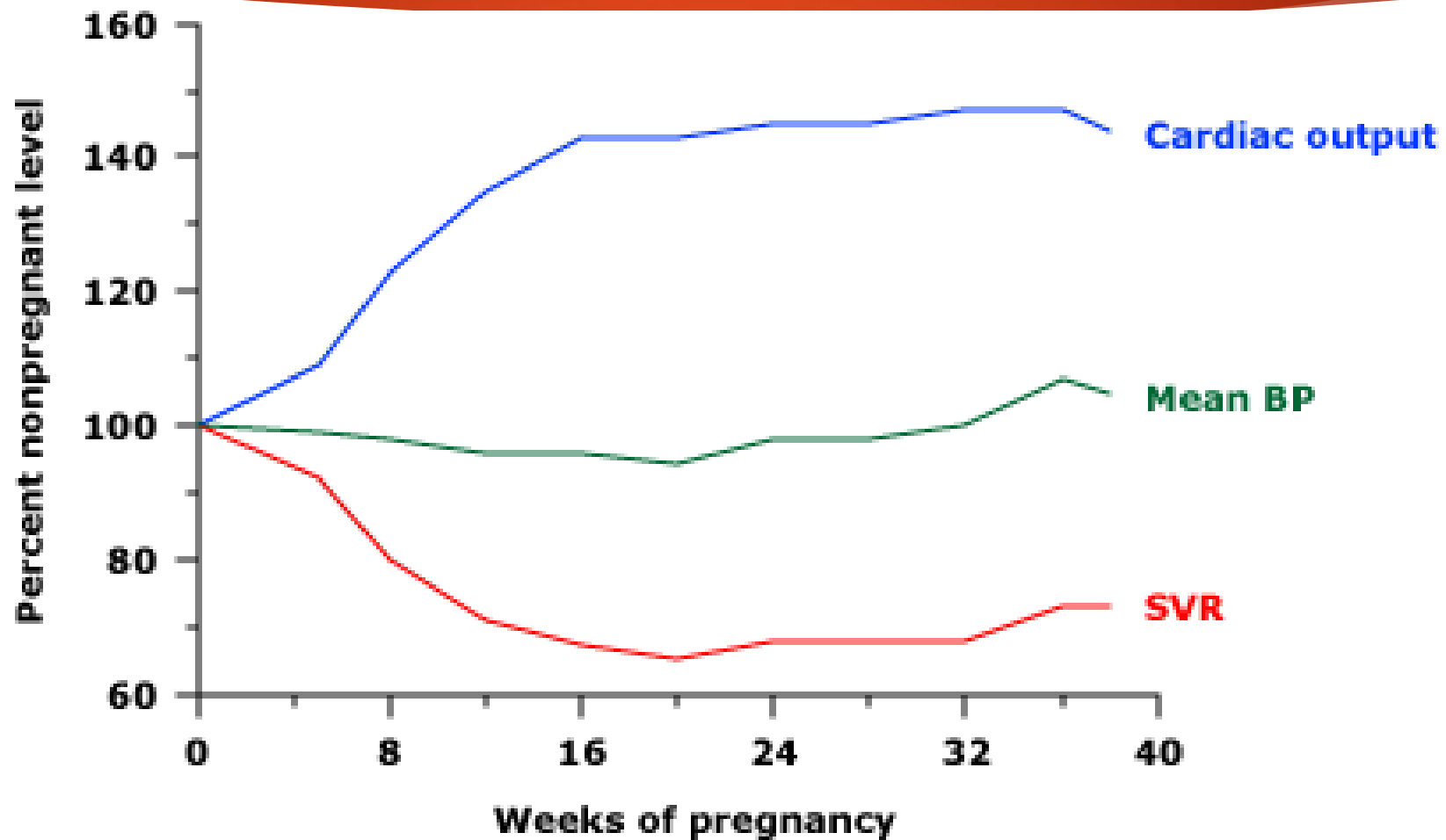
- ▶ What:
 - ▶ Amoxicillin 2 g po x 1 OR
 - ▶ Clindamycin 600 mg po x 1
- ▶ When:
 - ▶ 30-60 mins prior to **DENTAL PROCEDURES**
 - ▶ **NOT** required prior to non-dental procedures – EGD, colonoscopy, cystoscopy, vaginal delivery, IUD placement, etc.



ACHD and Pregnancy

- ▶ Heterogeneous Risk
- ▶ Even complete repairs can have residual lesions
 - ▶ PI/RV failure in ToF
 - ▶ PAH in ASD
 - ▶ Re-coarctation or aortic aneurysm in repaired coarctations
- ▶ Loss to follow up is common
- ▶ GOAL: PLANNING for most, PREVENTING for some

Hemodynamics in Pregnancy



Importance of counseling and risk assessment

- ▶ ACC/AHA Guidelines recommend counseling women with heart disease on contraception and pregnancy
 - ▶ Heterogeneous risk → Planning vs. Preventing
 - ▶ Teratogenic medications
 - ▶ Pre-conceptual imaging/interventions
 - ▶ Anticoagulation planning
 - ▶ Risk of fetal CHD

Cardiovascular Complications in Pregnancy

- ▶ Arrhythmias
- ▶ Heart Failure
- ▶ Thromboembolism
 - ▶ DVT/PE
 - ▶ MI
 - ▶ CVA
 - ▶ Valve Thrombosis

WHO Classification: Pregnancy Risk

- ▶ Class IV – Pregnancy is Contraindicated
 - ▶ Pulmonary Hypertension
 - ▶ Marfan's Syndrome with Dilated Aorta (>45 mm)
 - ▶ Bicuspid AV with Dilated Aorta (>50 mm)
 - ▶ Severe Left Sided Obstruction
 - ▶ Severe LV Dysfunction (EF <30% or NYHA III-IV)
 - ▶ Prior peripartum cardiomyopathy with any residual impairment of LV function

Cessation of Teratogenic Meds

Contraindicated

- ▶ ACE Inhibitors
- ▶ ARBs
- ▶ Statins
- ▶ Aldosterone Antagonists

Weigh Benefits and Risks

- ▶ Beta-Blockers
 - ▶ Exception: Avoid ATENOLOL
 - ▶ Preferred: Propranolol, Metoprolol, Nadolol
- ▶ Aspirin
- ▶ Calcium Channel Blockers
- ▶ Diuretics
- ▶ Antiarrhythmics
 - ▶ Avoid AMIODARONE if possible
- ▶ Digoxin
- ▶ Plavix

Caring for the Pregnant Woman with Heart Disease

- ▶ Multidisciplinary Approach
 - ▶ Experienced cardiologist, MFM, OB anesthesia, Neonatology, Cardiac anesthesia
- ▶ Pre-conceptual Counseling
- ▶ Delivery Planning
- ▶ Managing Cardiac Complications
- ▶ Contraception Counseling

Contraception

- ▶ Women with heart disease should receive counseling on contraception
 - ▶ PLANNING pregnancy for lower-risk patients
 - ▶ PREVENTING pregnancy for highest-risk patients
- ▶ Many women do not recall discussing with their cardiologist
- ▶ Others recall inaccurate information

Vigl, M., et al. (2010). "Contraception in women with congenital heart disease." *Am J Cardiol* **106**(9): 1317-1321.

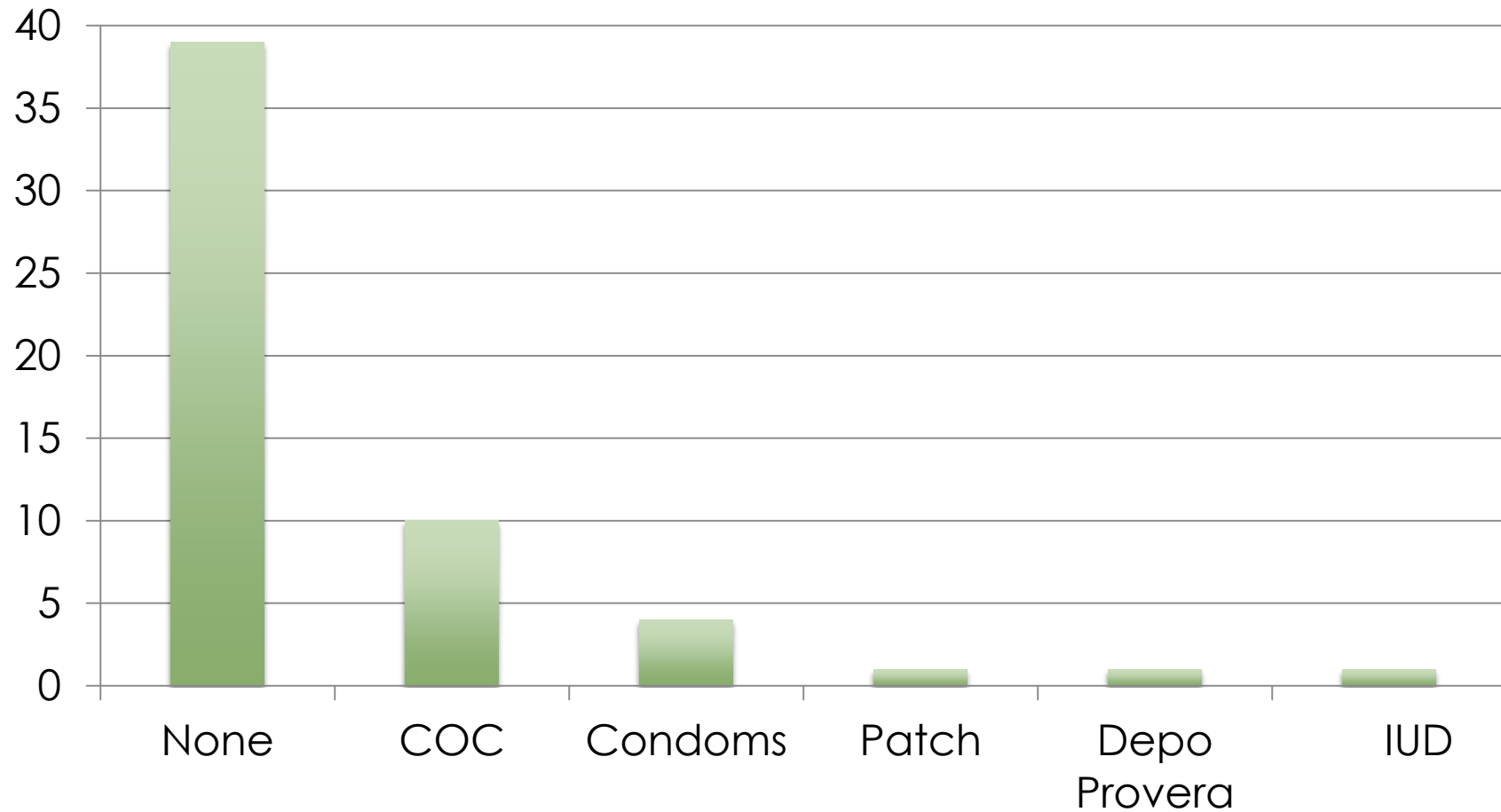
Kovacs, A. H., et al. (2008). "Pregnancy and contraception in congenital heart disease: what women are not told." *J Am Coll Cardiol* **50**(7): 577-579.

Unplanned Pregnancies in Women With ACHD

Percent of Women Having At Least One Unplanned Pregnancy (Range 1-10)

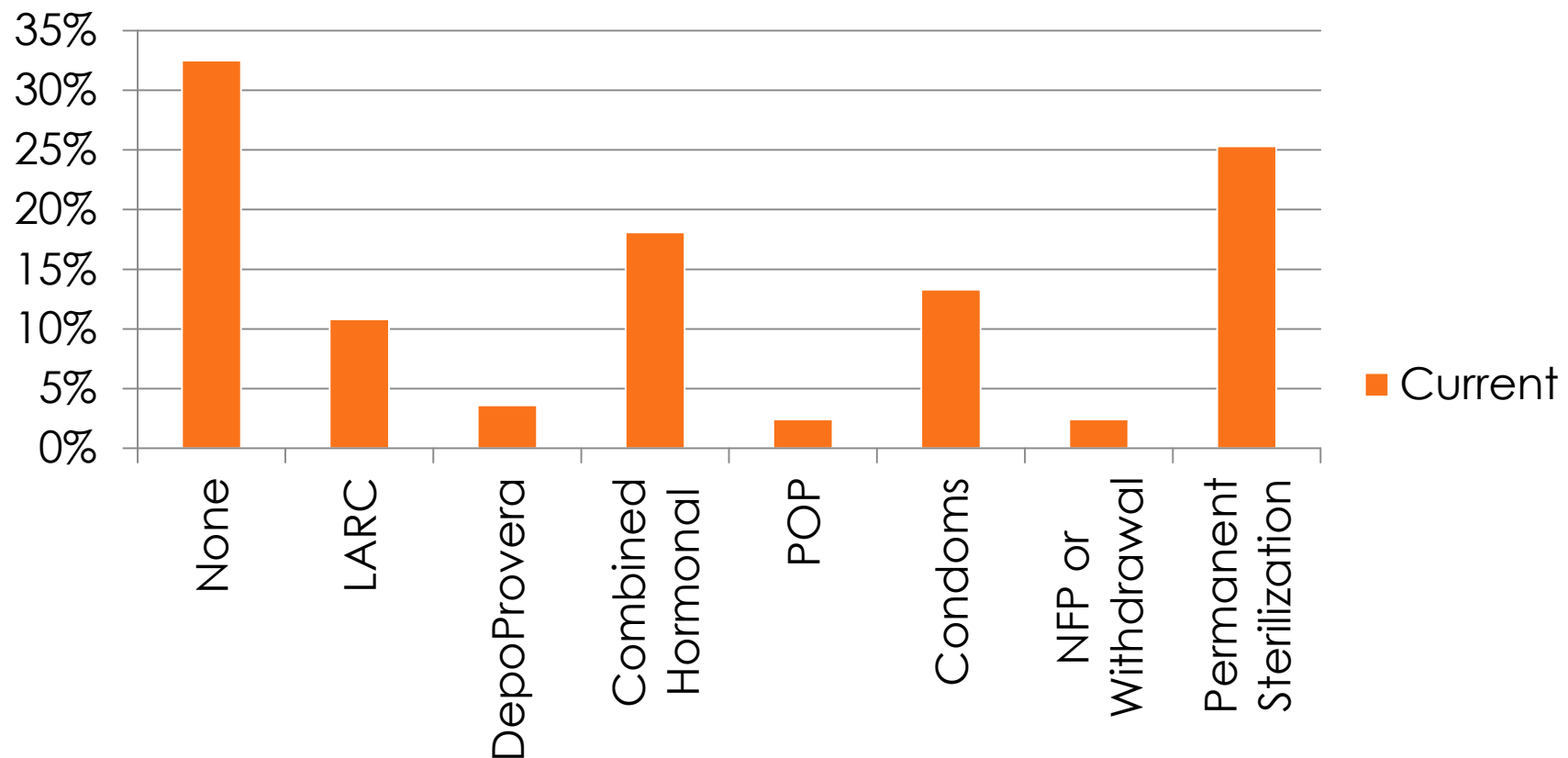


Method of Contraception at Time of Unintended Pregnancy



Contraceptive Use in Sexually Active Women with ACHD

Primary Method of Contraception



Contraception

- ▶ Is it safe?
- ▶ Does it work?

Safety Concerns

- ▶ Combined hormonal methods
 - ▶ Pill, patch, and ring
 - ▶ Associated with increased risk of thromboembolism
 - ▶ Absolute or relative contraindication in some cardiovascular conditions
- ▶ ***STILL SAFER THAN PREGNANCY...DON'T STOP BEFORE INITIATING AN ALTERNATIVE METHOD***

WHO COC Risk: Contraindications

- ▶ Pulmonary hypertension or Fontan Palliation
- ▶ Atrial Fibrillation
- ▶ Mechanical Valves
- ▶ R to L Shunt
- ▶ Coronary or Aortic Diseases
- ▶ Previous Thromboembolism
- ▶ LV Dysfunction
- ▶ Hypertension (relative)

Tiers of Contraceptive Effectiveness

- ▶ I – Failure Rate <1%
 - ▶ Permanent sterilization
 - ▶ Long Acting Reversible Contraception (LARC)
- ▶ II – Failure Rate 6-12%
 - ▶ Combined Hormonal Contraceptives
 - ▶ Progestin Only Contraceptives
- ▶ III – Failure Rate 12-24%
 - ▶ Barrier methods
 - ▶ Withdrawal
 - ▶ Fertility awareness methods
- ▶ None – 85% pregnancy rate within 1 year

Long Acting Reversible Contraception

- ▶ 4 Options:
 - ▶ Levonorgestrel impregnated IUD
 - ▶ Mirena, Skyla, Liletta, Kyleena
 - ▶ Copper IUD
 - ▶ Etonogestrel impregnated rod
- ▶ As/more effective than tubal ligation
 - ▶ Lower risk procedure
- ▶ Estrogen-free
- ▶ Completely reversible
- ▶ FDA approved for 3 to 10 years

Recommendations: Contraception for Women with Heart Disease

- ▶ Method of contraception assessed and documented annually
- ▶ Long acting reversible contraception should be preferred method for:
 - ▶ WHO Class III-IV
 - ▶ All patients taking potentially teratogenic medications

Summary

- ▶ Adults with CHD is a rapidly growing population
 - ▶ You will see them!
 - ▶ You will diagnose them!
- ▶ Adults with CHD are thriving, but require lifelong specialized cardiology care
- ▶ In addition to usual care, adults with CHD require special attention
 - ▶ SBE prevention
 - ▶ Family Planning
 - ▶ Late sequelae of disease

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PATIENT CARE

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- Adult Congenital Heart Disease
- Cardio-Neuromuscular Disorders
- Heart Rhythm Disorders
- Cardio-Oncology Center of Excellence
- Hypertrophic Cardiomyopathy
- Thoracic Aortic Disease
- Valvular Heart Disease Center
- Women's Heart Disease

Adult Congenital Heart Disease



The Adult Congenital Heart Disease Center, which was started in 1992, is one of the largest centers for adults with congenital heart defects in the United States, according to the listings of the International Society for Adults with Congenital Heart Disease. The center provides care for approximately 2000 patients.

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- Valvular Heart Disease Center
- Women's Heart Disease

Women's Heart Disease



Heart disease is the leading killer of women in the United States, yet it often is under-recognized and under-treated. We recognize the importance of preventing, identifying and treating women with cardiovascular disease.

Contact: 314-362-1292 or kathryn.lindley@wustl.edu
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