



Brigham and Women's Hospital

Founding Member, Mass General Brigham

ADULT CONGENITAL HEART DISEASE

Anne Marie Valente, MD

Director, Boston Adult Congenital Heart Program

Department of Pediatric Cardiology, Boston Children's Hospital

Cardiology, Brigham and Women's Hospital

Professor, Pediatrics; Associate Professor, Medicine

Harvard Medical School



Anne Marie Valente, MD



- University of Vermont Medical School
- Medicine & Pediatrics Residency @ Duke
- Cardiovascular Medicine Fellowship @ Duke
- Pediatric Cardiology Fellowship @ Duke
- Professor of Pediatrics @ Harvard Medical School
 - Clinical Focus: Adult Congenital Heart Disease, CardioObsetrics
 - Research Focus: Imaging and Outcomes

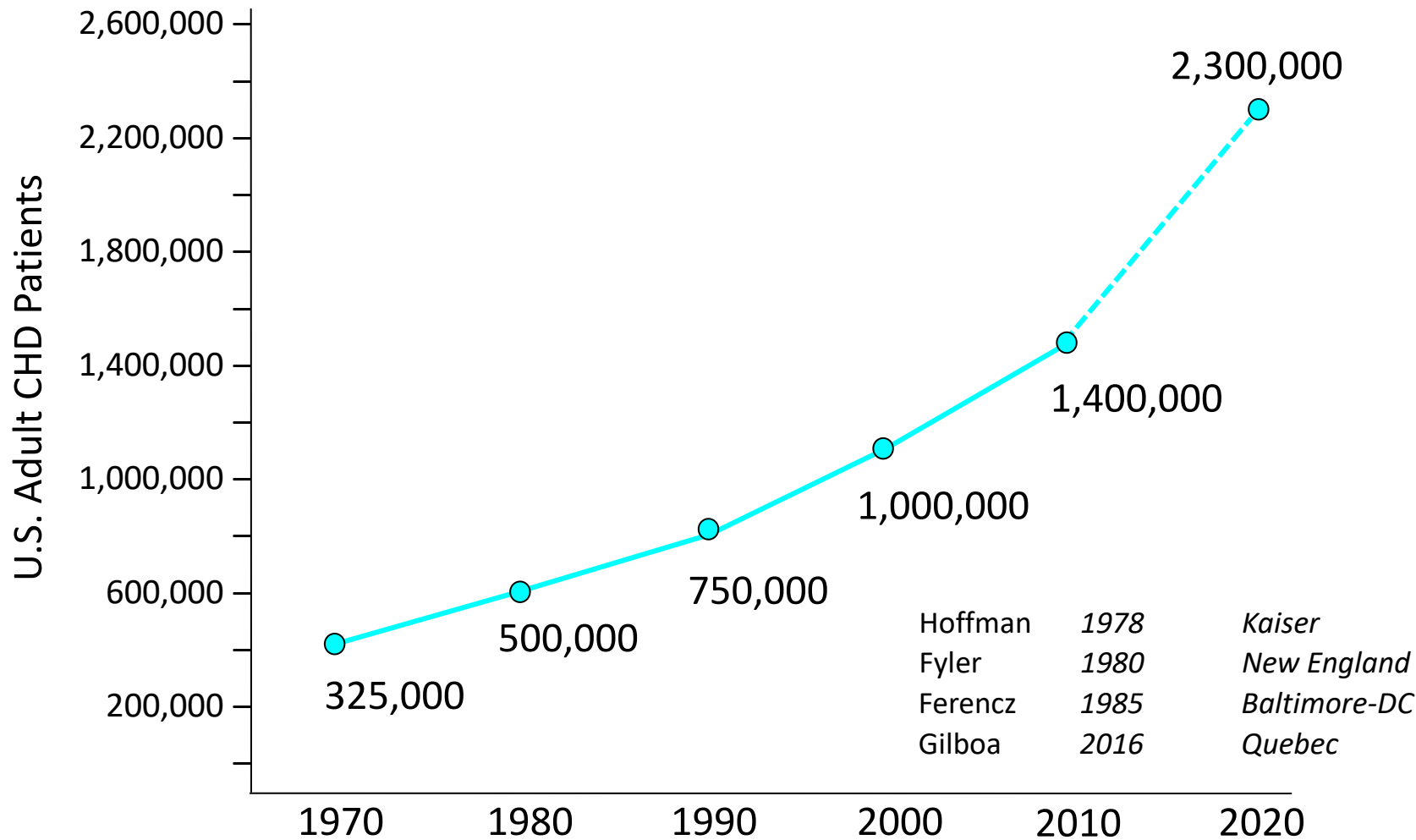
Disclosures

- Elsevier: Advisory Board

Objectives

- To understand the anatomy and physiology of the most common congenital heart conditions seen in adults
- To review current guideline recommendations for care

The ACHD Population Is Growing

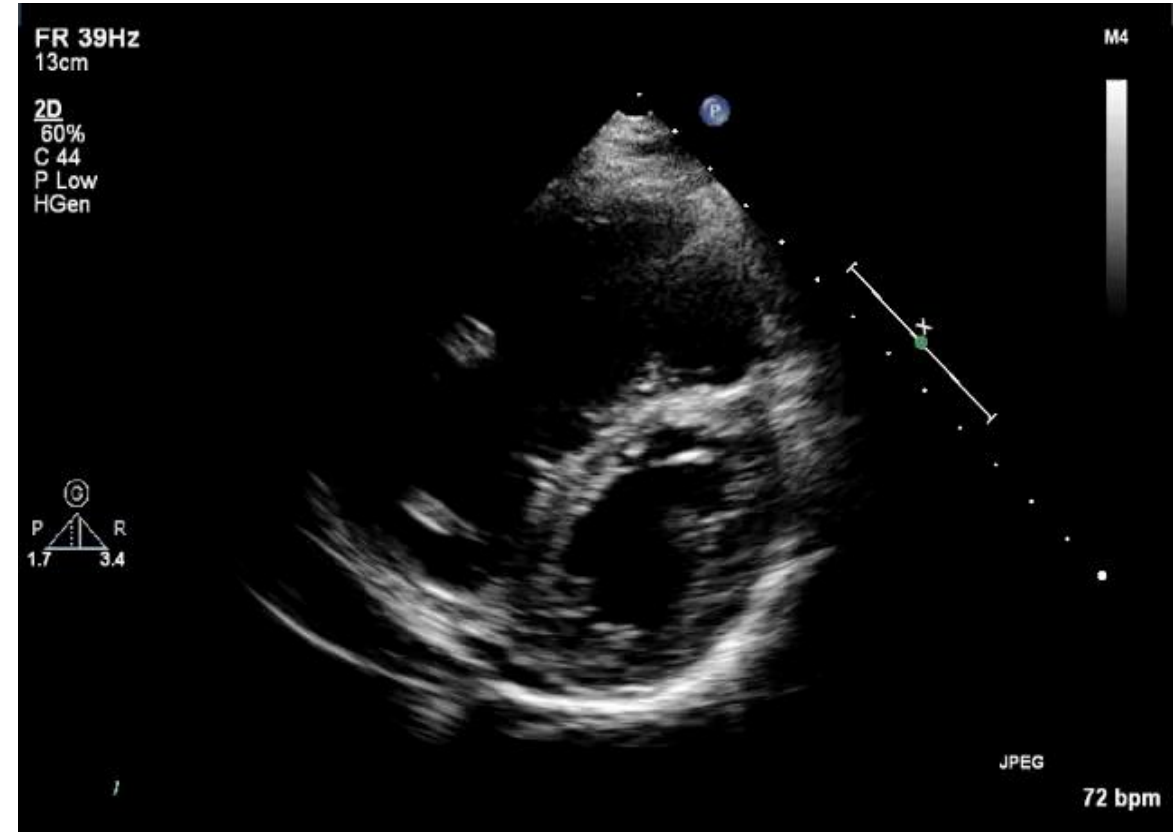


85% increase in adults living with **complex** CHD between 1985 and 2000

By 2030, it is estimated that 11% of the European ACHD population will be >60 years old

ACHD Question #1

A 30-year-old woman is referred for palpitations and dyspnea with exertion. On physical exam, the S2 is widely split and does not vary with inspiration. An echocardiogram reveals RV dilation, normal RV pressure, and no shunts are visualized. What is the most appropriate next step?



ACHD Question #1

A 30-year-old woman is referred for palpitations and dyspnea with exertion. On physical exam, the S2 is widely split and does not vary with inspiration. An echocardiogram reveals RV dilation, normal RV pressure, and no shunts are visualized. What is the most appropriate next step?

- a) Cardiac catheterization
- b) Exercise echocardiogram
- c) Cardiac MRI
- d) Echocardiogram with bubble study
- e) Cardiac CT

ACHD Answer #1

A 30-year-old woman is referred for palpitations and dyspnea with exertion. On physical exam, the S2 is widely split and does not vary with inspiration. An echocardiogram reveals RV dilation, normal RV pressure, and no shunts are visualized. What is the most appropriate next step?

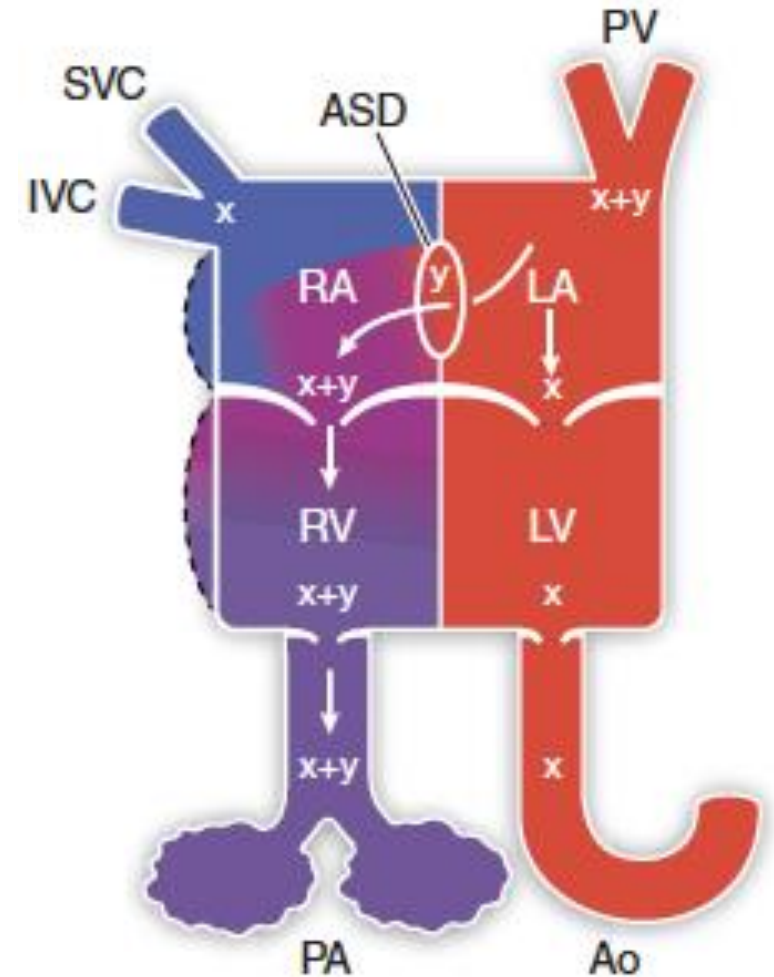
- a) Cardiac catheterization
- b) Exercise echocardiogram
- c) Cardiac MRI**
- d) Echocardiogram with bubble study
- e) Cardiac CT

Atrial Septal Defect (ASD)

- Left-to-right shunt causing pulmonary overcirculation and R heart dilation
- Palpitations, dyspnea, exercise intolerance
- Exam findings: Fixed, split S2
- EKG: RBBB, RAD*

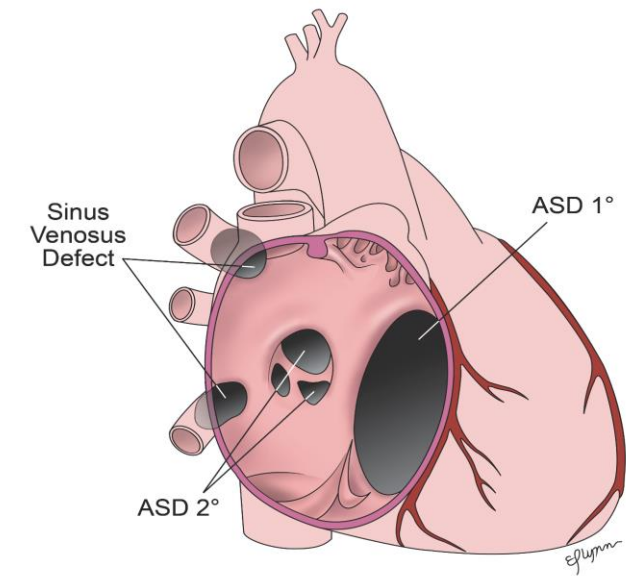
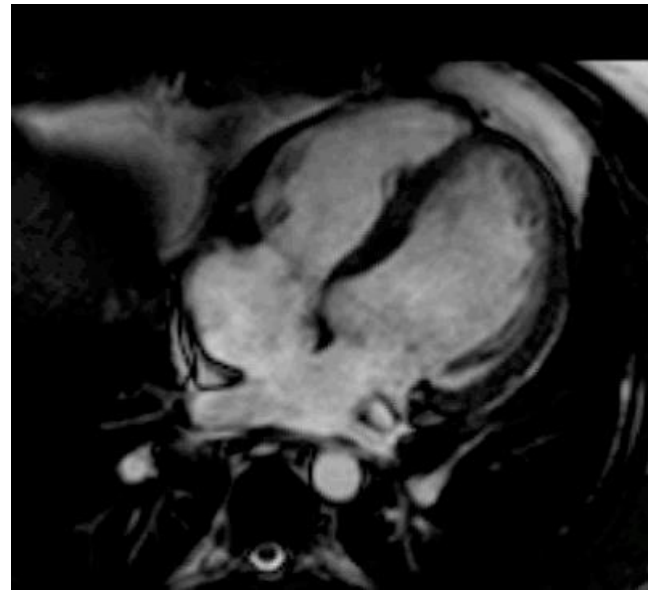
*for secundum ASDs (most common)

If LAD -> think primum ASD

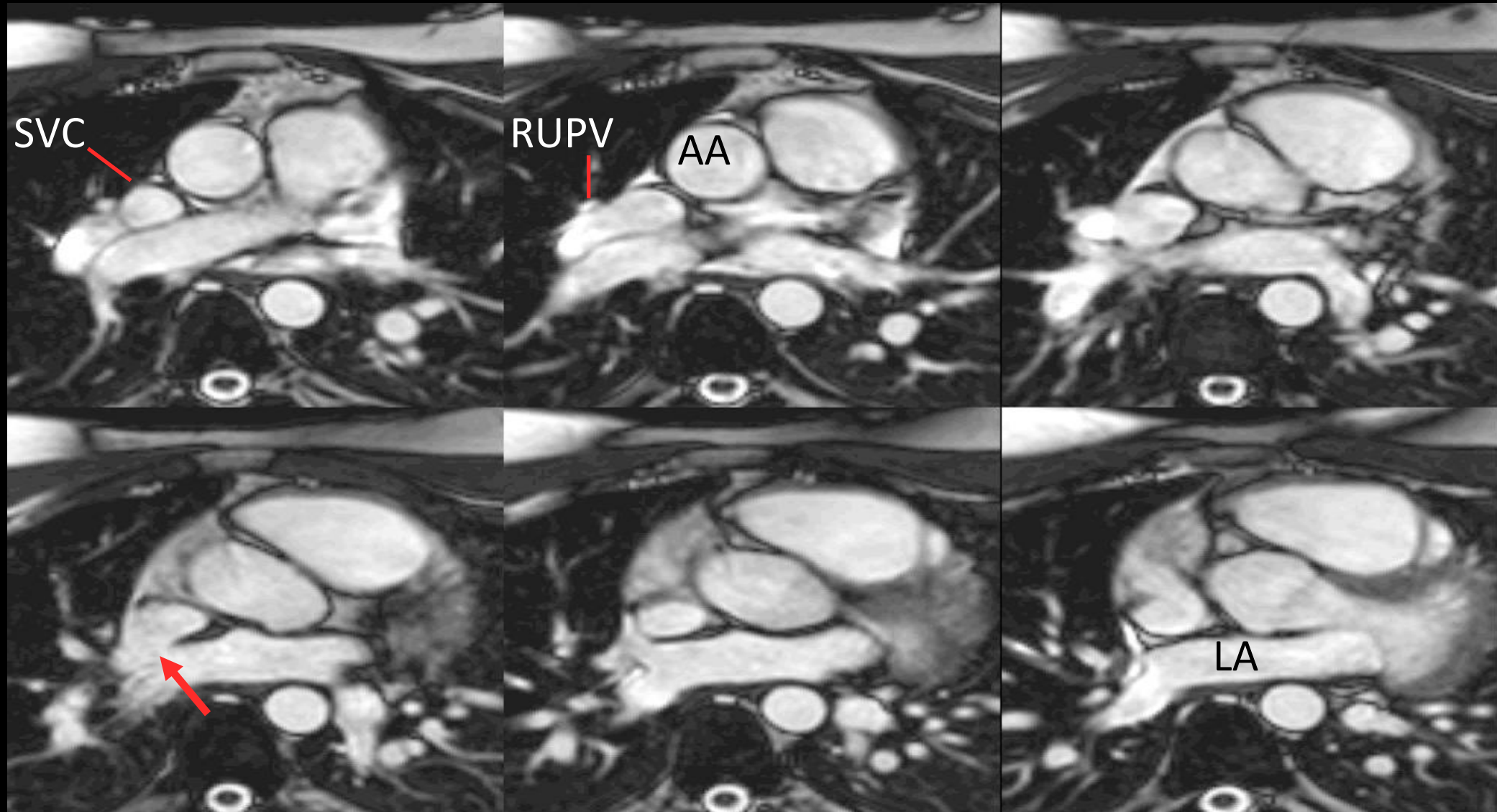


Atrial Septal Defects: One Size Does Not Fit All

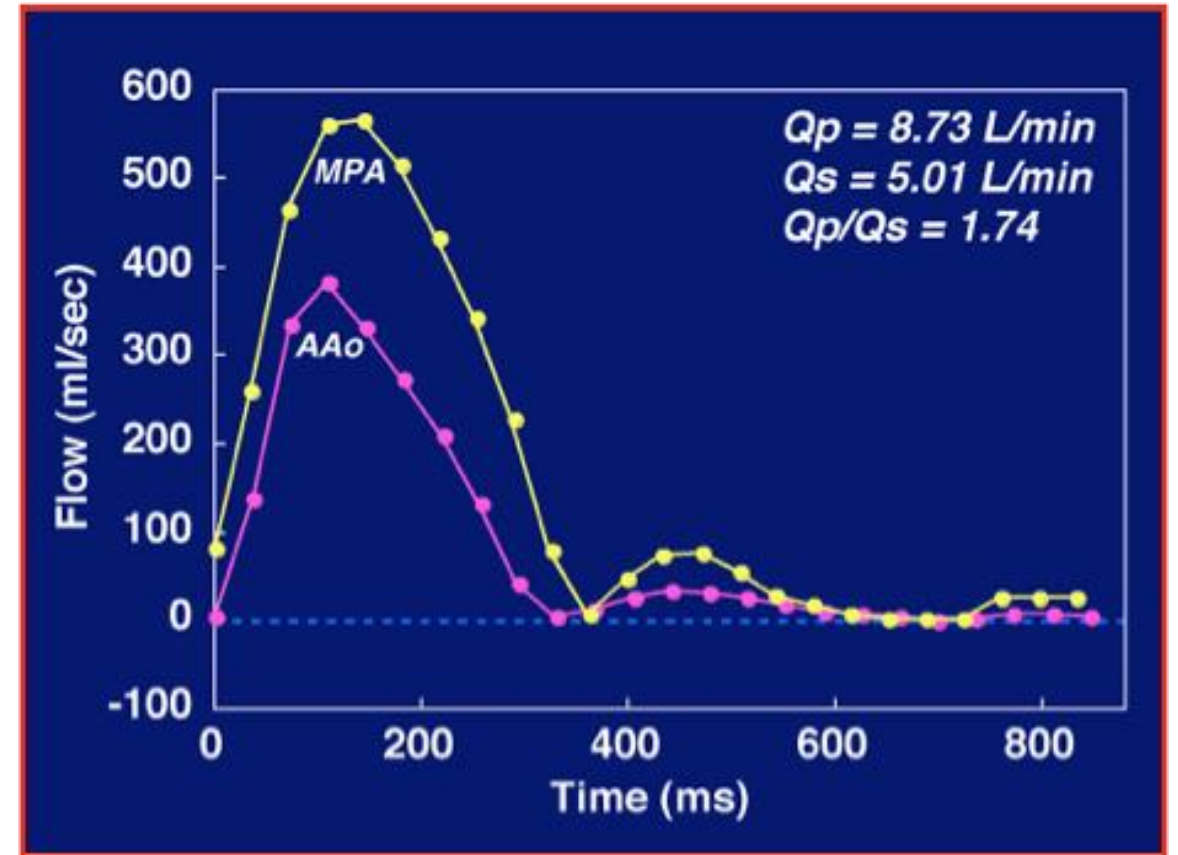
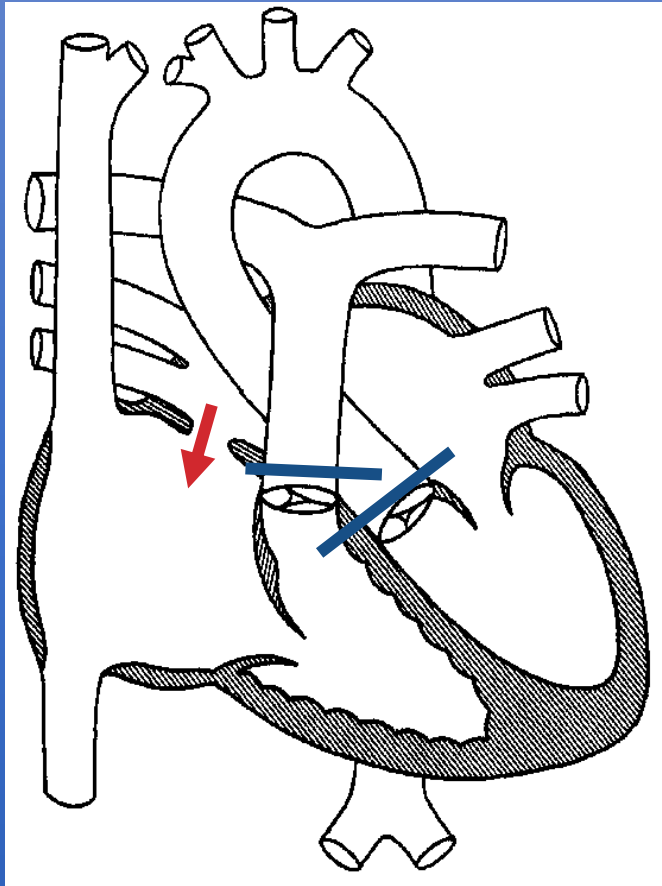
- **Secundum ASD:** most common
- **Primum ASD:** on the spectrum of AVC defects; associated with a cleft mitral valve
- **Sinus venosus defect*:** associated with partial anomalous pulmonary venous return
 - *Not in the atrial septum



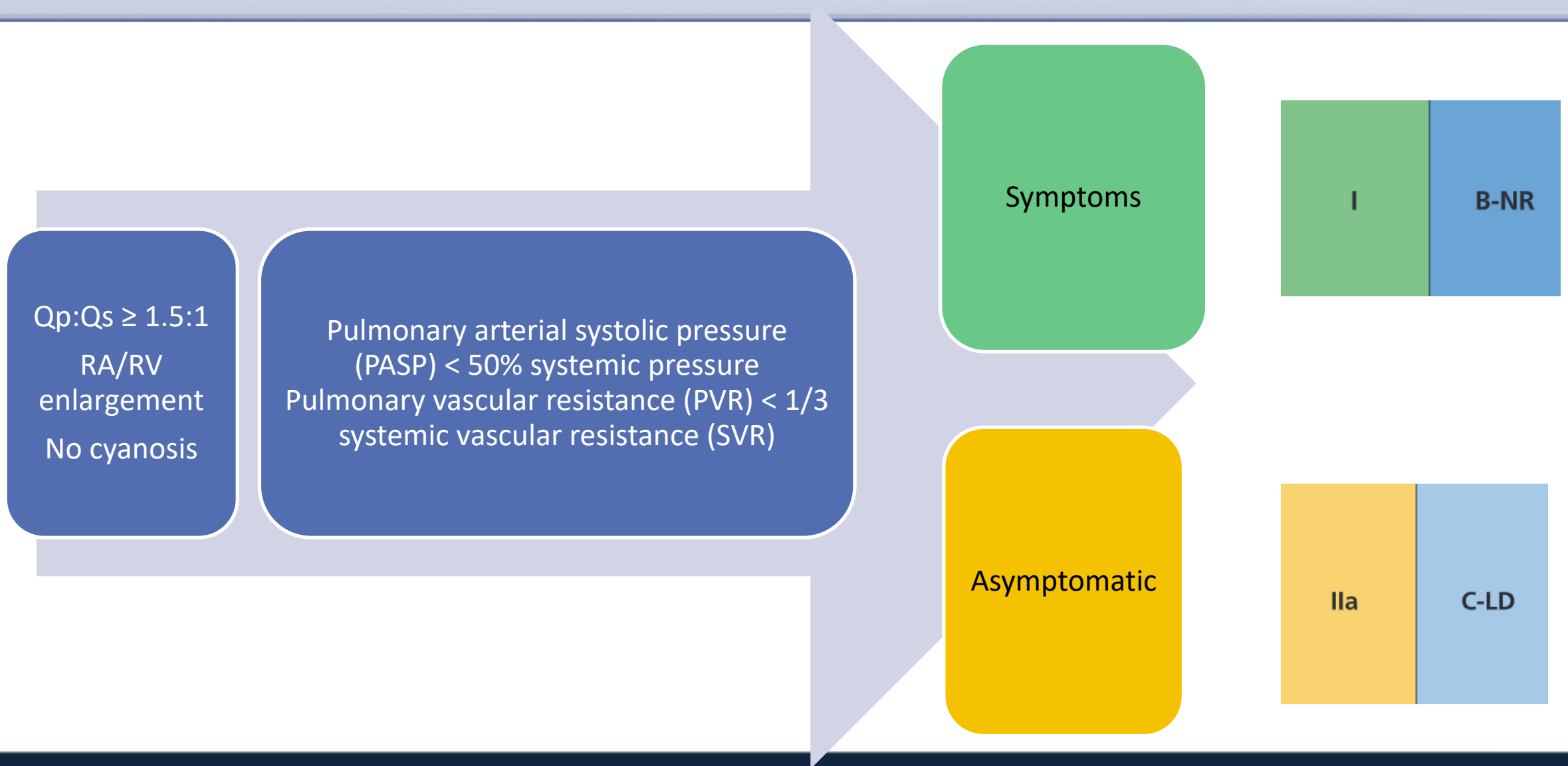
Superior Sinus Venosus Defect



ASD: Quantification of Shunt



ASD: Guidelines for Closure



ASD: When Not to Close



Net right-to-left shunt OR



PASP > 2/3 systemic pressure OR



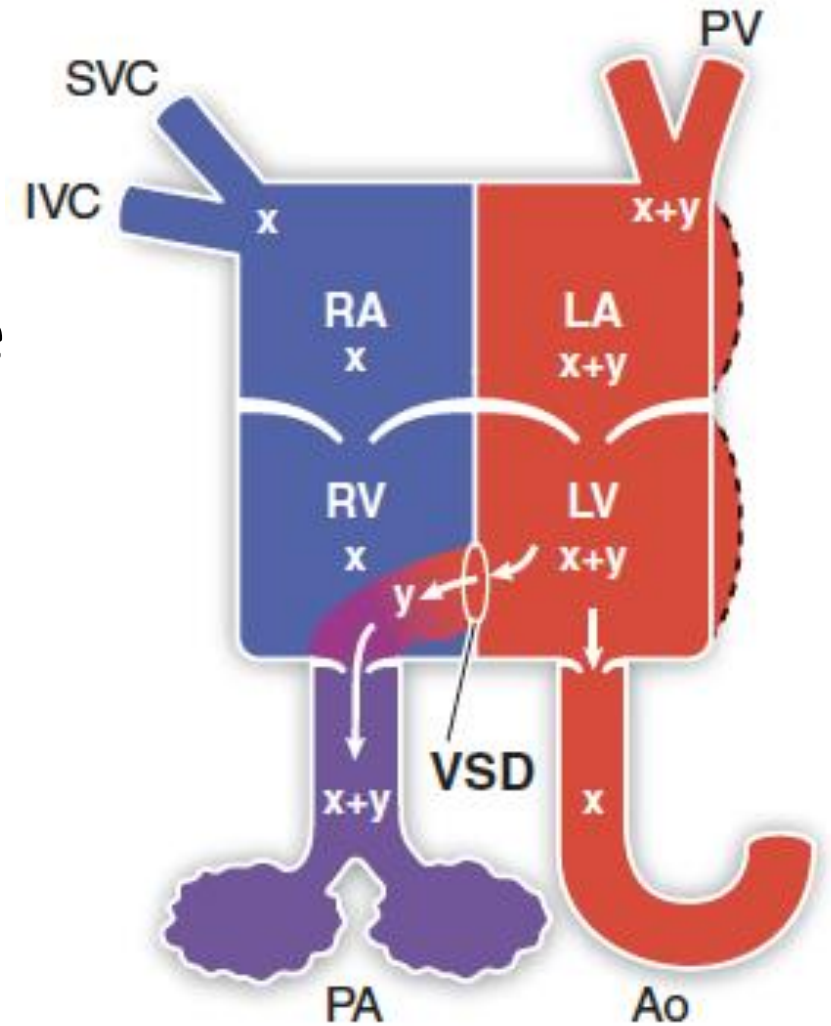
PVR > 2/3 SVR

III: Harm

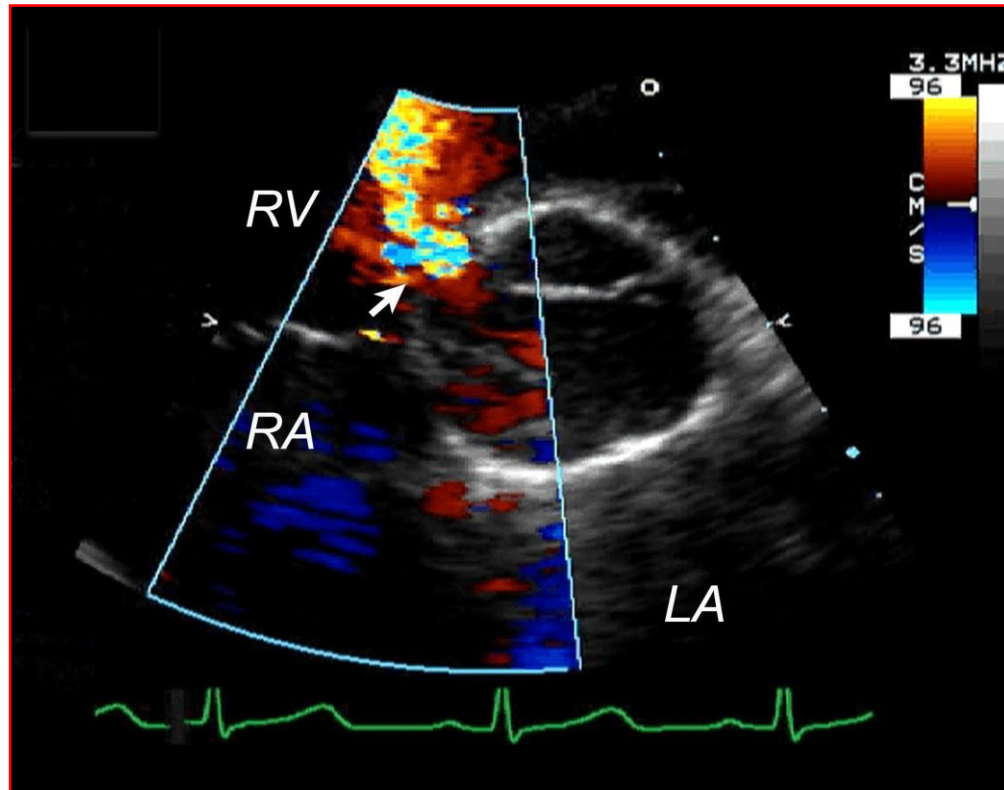
C-LD

Ventricular Septal Defect

- Left-to-right shunt causing left heart dilation
- Large VSDs cause symptoms of heart failure and poor growth in children
- Many VSDs found in adults are pressure- and flow-restrictive and do not cause LV dilation or symptoms
- If not restrictive -> Eisenmenger syndrome

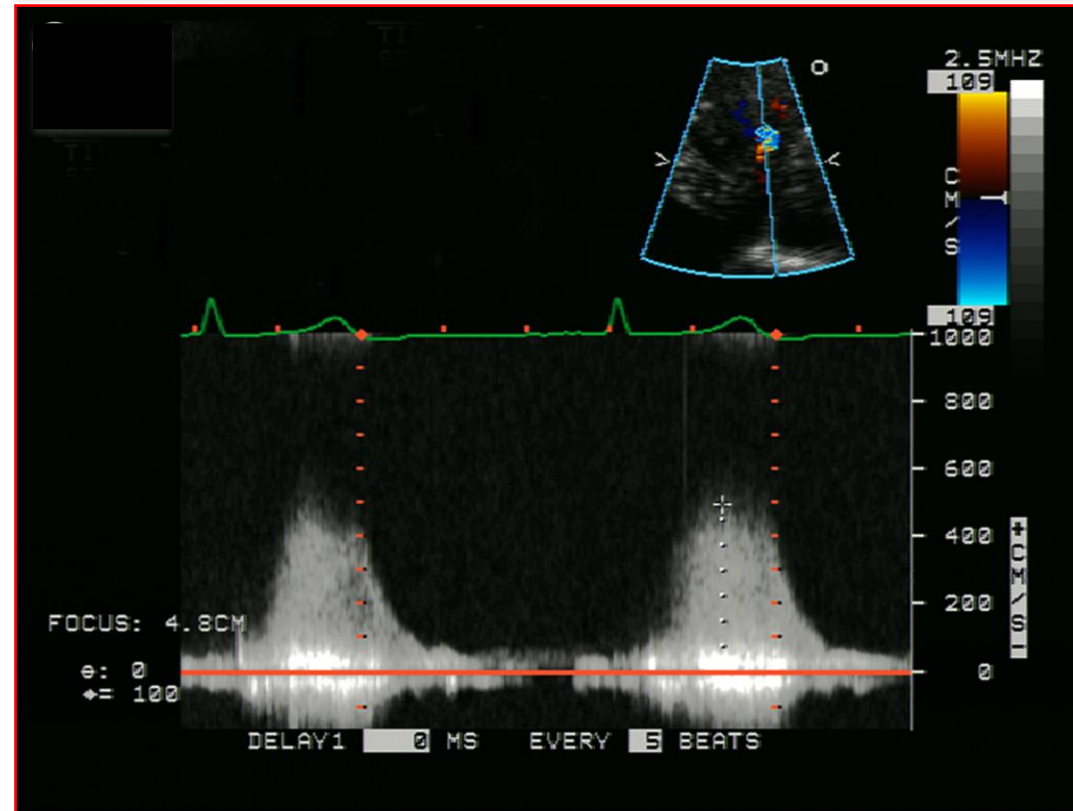


Pressure-Restrictive VSD



$4v^2$
 $\sim 5 \text{ m/s} = 4(25) = 100 \text{ mmHg}$
gradient between the RV and LV

The key is to have a HIGH velocity on Doppler interrogation of the VSD flow



Expected Chamber Enlargement with Cardiac Shunts

To dilate a chamber, it needs to fill during diastole

VSD ejects *through* the right ventricle (and into the PA) during systole

Shunted blood returns to the left heart and fills (and dilates) during diastole

Shunt	RA	RV	PA	LA	LV	Aorta
ASD	+	+	+			
VSD		+/-	+	+	+	
PDA			+	+	+	+

Right-to-Left Shunts

Deoxygenated blood from right side of the heart shunts over to the left side



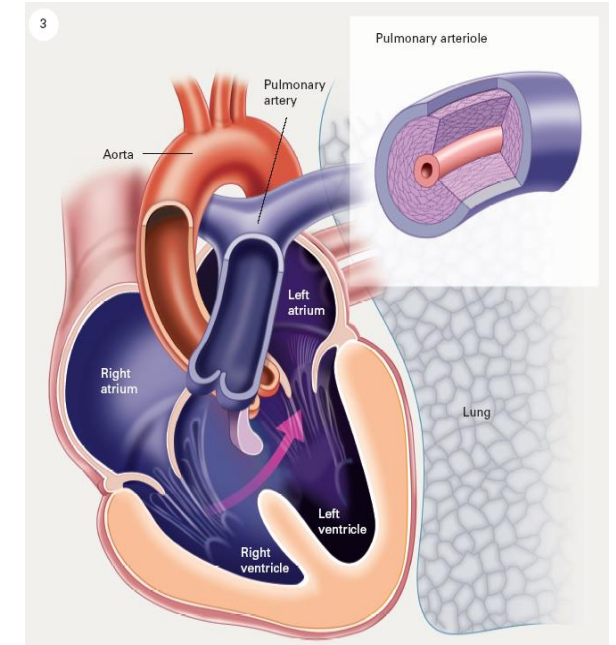
Takes blood from the right side of the heart and pulmonary blood flow decreases



Since pulmonary blood flow decreases, no increase in ventricular volumes



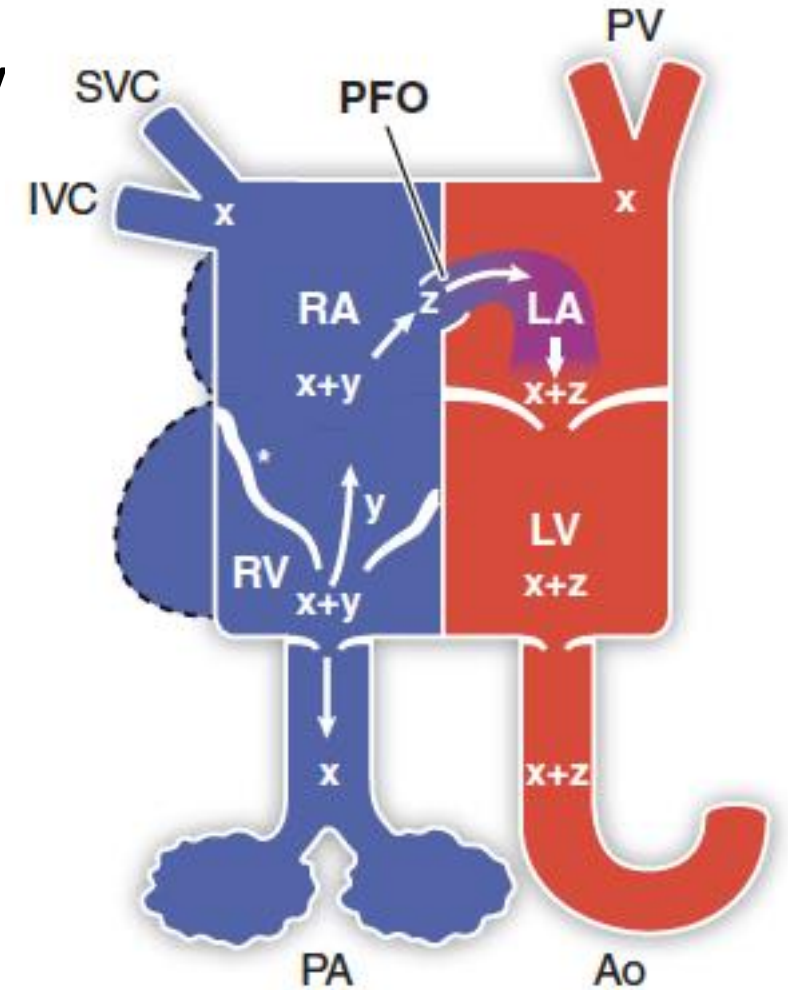
Systemic arterial desaturation (cyanosis)



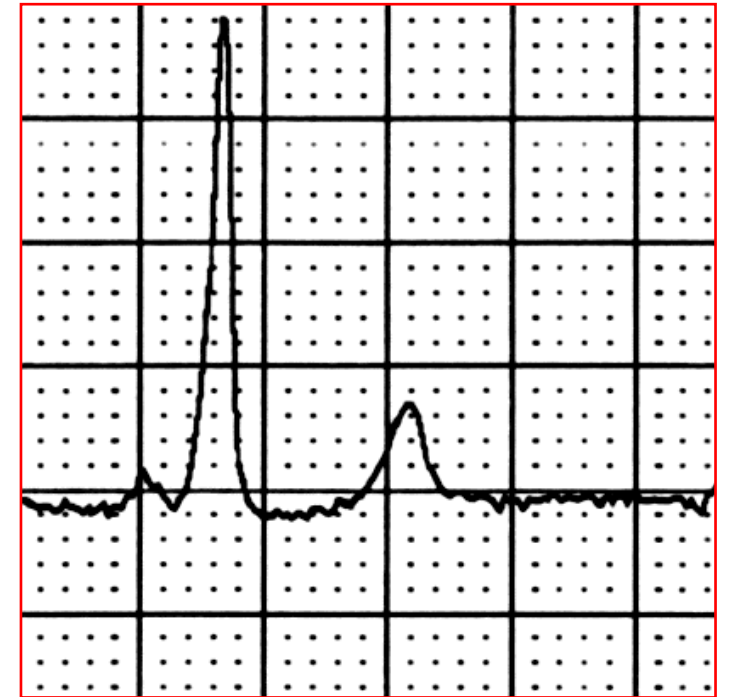
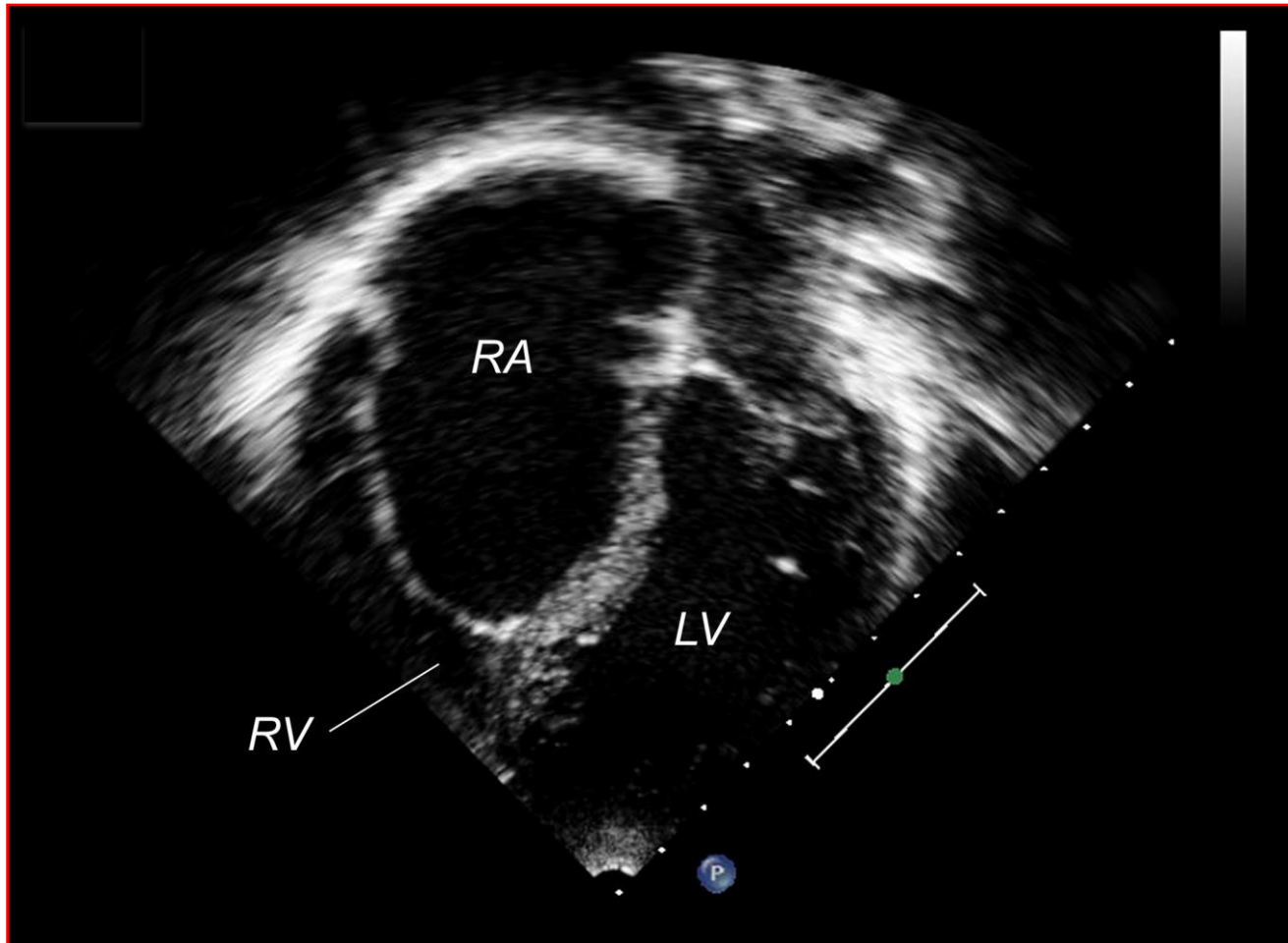
PVR ↑'s: shunt reverses:
R-to-L → Eisenmenger
syndrome: ↑ cyanosis

Ebstein Anomaly

- Failure of delamination of the septal leaflet of the TV
- Results in “atrialized” portion of the RV
- Symptoms depend on severity of TR and associated lesions
 - Signs of R heart failure
 - Cyanosis (if ASD/PFO)
- Exam: Widely split S1 “sail sign”
- 20% of patients have WPW



Ebstein Anomaly



ACHD Question #2

A 45-year-old with tetralogy of Fallot presents to your office for clearance prior to a screening colonoscopy. She had undergone a bioprosthetic pulmonary valve replacement 8 years ago for pulmonary regurgitation. She has no known drug allergies. Examination is significant for a 2/4 diastolic murmur, and echocardiogram reveals stable bioprosthetic pulmonary valve function with moderate regurgitation. What is the most appropriate medication to be given prior to her colonoscopy?

- a) Amoxicillin 2 grams 30 minutes prior to procedure
- b) Amoxicillin 2 grams one hour prior to and one hour after procedure
- c) Clindamycin 600 mg 30 minutes prior to procedure
- d) No antibiotic prophylaxis

ACHD Answer #2

A 45-year-old with tetralogy of Fallot presents to your office for clearance prior to a screening colonoscopy. She had undergone a bioprosthetic pulmonary valve replacement 8 years ago for pulmonary regurgitation. She has no known drug allergies. Examination is significant for a 2/4 diastolic murmur, and echocardiogram reveals stable bioprosthetic pulmonary valve function with moderate regurgitation. What is the most appropriate medication to be given prior to her colonoscopy?

- a) Amoxicillin 2 grams 30 minutes prior to procedure
- b) Amoxicillin 2 grams one hour prior to and one hour after procedure
- c) Clindamycin 600 mg 30 minutes prior to procedure
- d) No antibiotic prophylaxis**

SBE Prophylaxis Guidelines

High-risk Patients

Prosthetic valves, including mechanical, bioprosthetic, homografts

Prosthetic material for valve repair, including annuloplasty rings

A prior history of IE

Unrepaired cyanotic CHD

Repaired CHD with residual shunts or valvular regurgitation at or adjacent to the site of the prosthetic patch or prosthetic device

Repaired CHD with catheter-based intervention involving an occlusion device or stent for 6 months

Valve regurgitation due to a structurally abnormal valve in a transplanted heart

High-risk Procedures

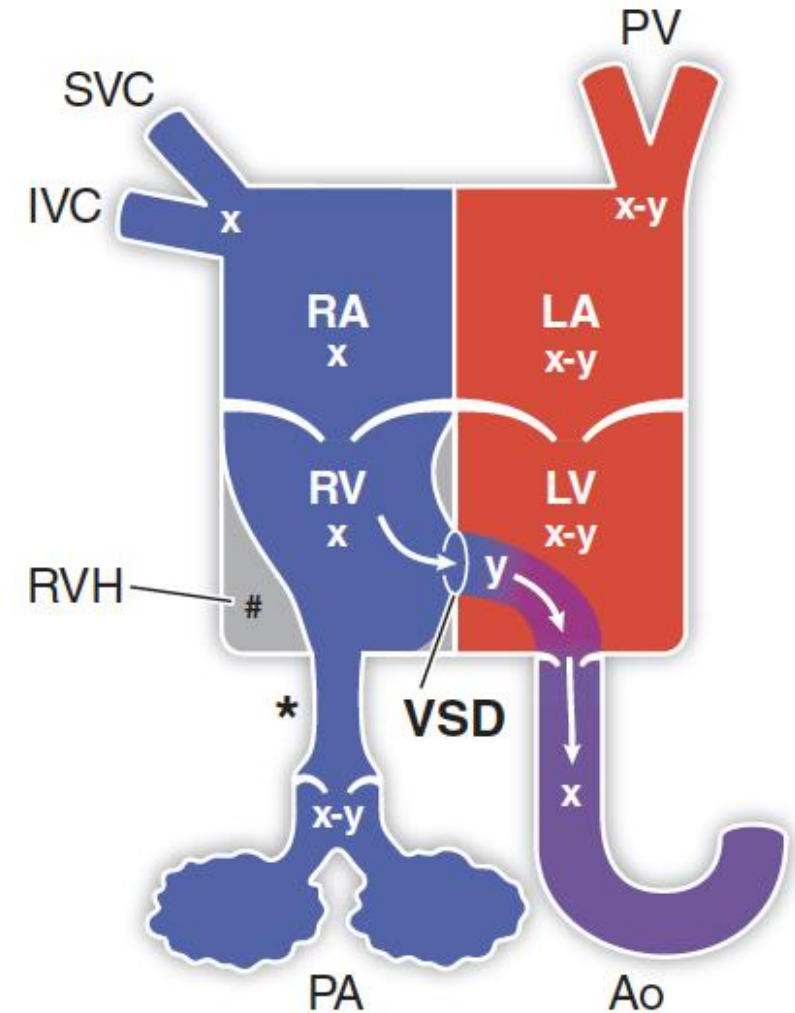
Dental work including abscess drainage, extractions, or routine cleaning

NOT NEEDED FOR:

Procedures in GI or GU tract, vaginal or cesarean delivery, respiratory tract procedures

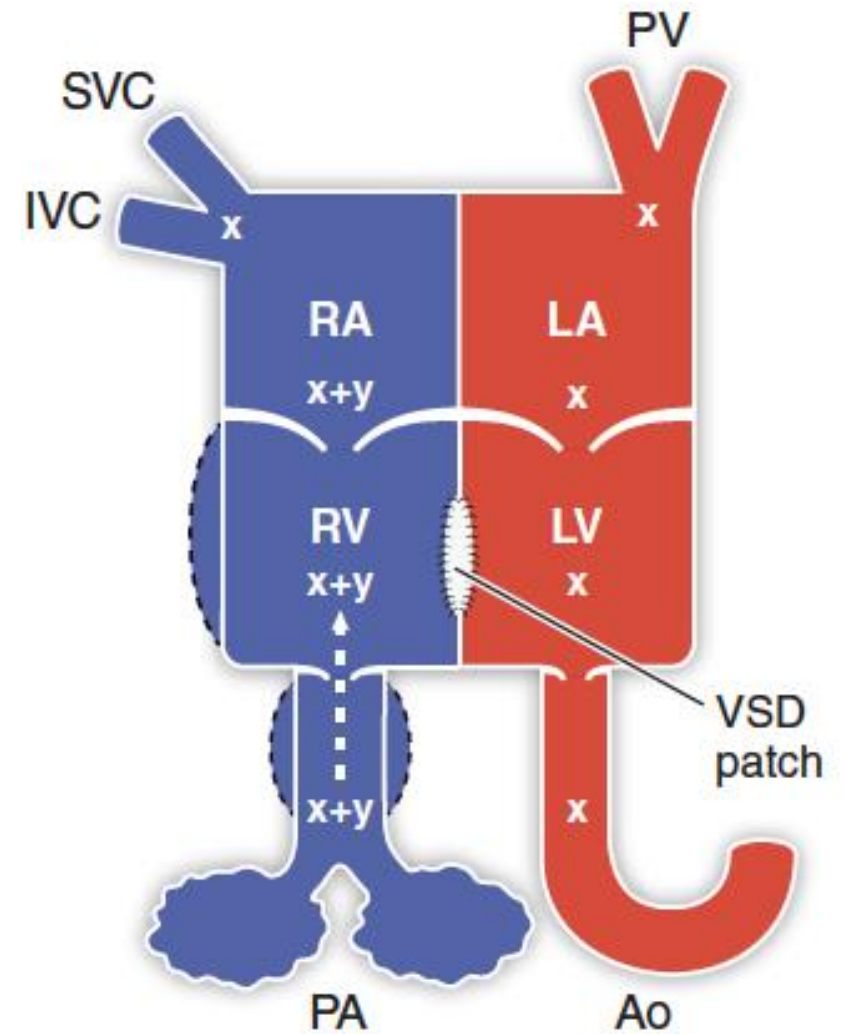
Tetralogy of Fallot (TOF)

- The most common form of cyanotic CHD
- Spectrum of severity
- Surgical repairs have changed over time
 - Palliative shunts
 - Transannular patch
 - RV-PA conduit
- Many adults undergo PVR later in life



Repaired TOF: Sequelae in Adulthood

- RA/RV dilation
- RV dysfunction
- **Pulmonary regurgitation**
- Tricuspid regurgitation
- Branch PA stenosis
- Residual VSD
- LV dysfunction
- Aortic root dilation
- Arrhythmias
- Sudden death



Indications for PVR in Repaired TOF

I

B-NR

- PVR for relief of **symptoms** not otherwise explained in adults with rTOF with \geq moderate PR

IIa

B-NR

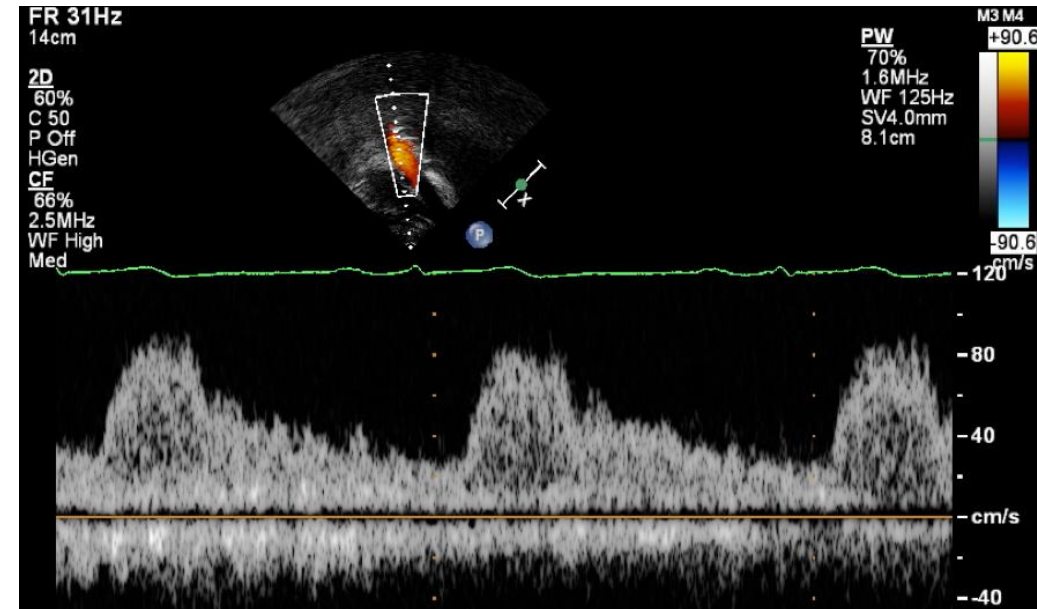
- PVR is reasonable for preservation of ventricular size & function in asymptomatic adults with ventricular enlargement or dysfunction with \geq moderate PR



ACHD Question #3

A 28-year-old is seen for evaluation of headaches. On exam, BP is 172/94 mmHg. A 3/6 systolic ejection murmur is heard at the left sternal border. An echocardiogram is performed which demonstrates normal left ventricular function, left ventricular hypertrophy, and no significant valve disease. Abdominal aorta Doppler is shown here. What is the most common additional physical examination finding with this condition?

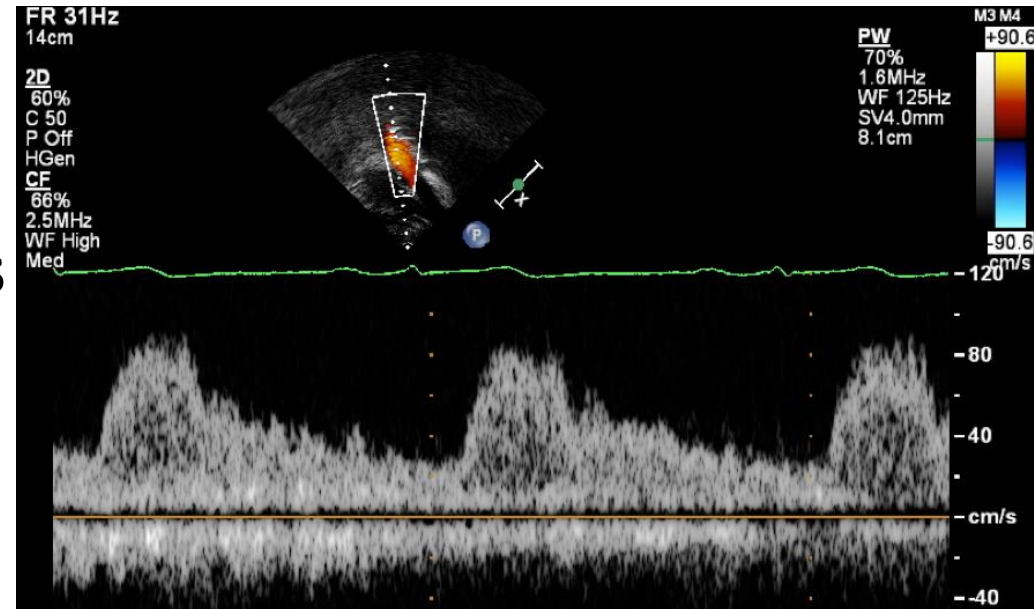
- a) Clubbing of the fingernails
- b) Bifid uvula
- c) Diminished pulses in the lower extremities
- d) An abdominal bruit



ACHD Answer #3

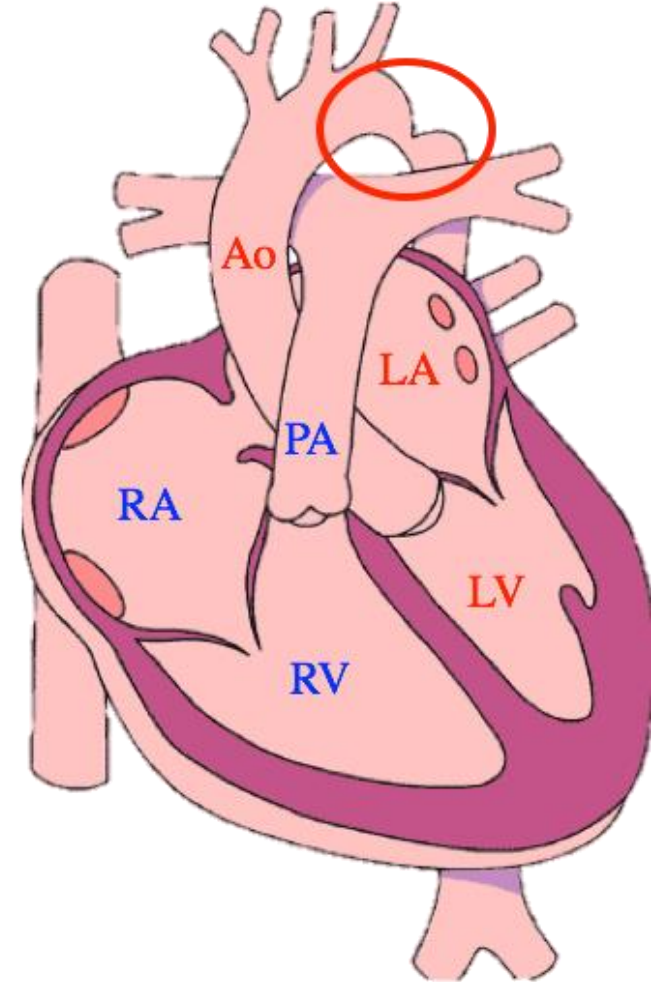
A 28-year-old is seen for evaluation of headaches. On exam, BP is 172/94 mmHg. A 3/6 systolic ejection murmur is heard at the left sternal border. An echocardiogram is performed which demonstrates normal left ventricular function, left ventricular hypertrophy, and no significant valve disease. Abdominal aorta Doppler is shown here. What is the most common additional physical examination finding with this condition?

- a) Clubbing of the fingernails
- b) Bifid uvula
- c) Diminished pulses in the lower extremities**
- d) An abdominal bruit



Coarctation of the Aorta

- Presents with hypertension
- Exam: Brachial-femoral delay and diminished LE pulses
- Associations:
 - Bicuspid aortic valve
 - Ascending aortic dilation
 - Aneurysms
 - LVH
 - Coronary artery disease
 - Cerebral aneurysm



ACHD Question #4

A 35-year-old with D-loop transposition of the great arteries who underwent an arterial switch in infancy reports chest heaviness with exercise. EKG reveals lateral ST depressions. Echocardiogram shows a LV EF of 42% and no significant valve dysfunction. What is the best next step?

- a) Right heart catheterization
- b) Lung perfusion scan
- c) Empiric treatment for GERD
- d) Coronary angiography
- e) Cardiac MRI

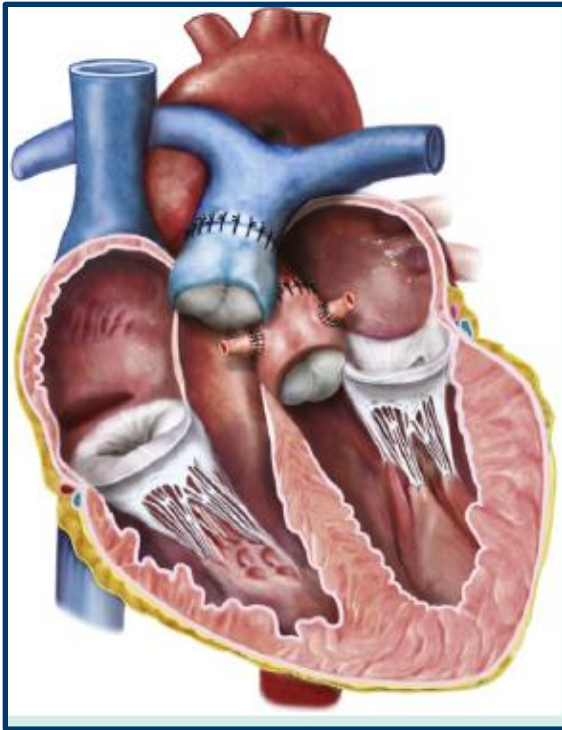
ACHD Answer #4

A 35-year-old with D-loop transposition of the great arteries who underwent an arterial switch in infancy reports chest heaviness with exercise. EKG reveals lateral ST depressions. Echocardiogram shows a LV EF of 42% and no significant valve dysfunction. What is the best next step?

- a) Right heart catheterization
- b) Lung perfusion scan
- c) Empiric treatment for GERD
- d) Coronary angiography**
- e) Cardiac MRI

ACHD Answer #4

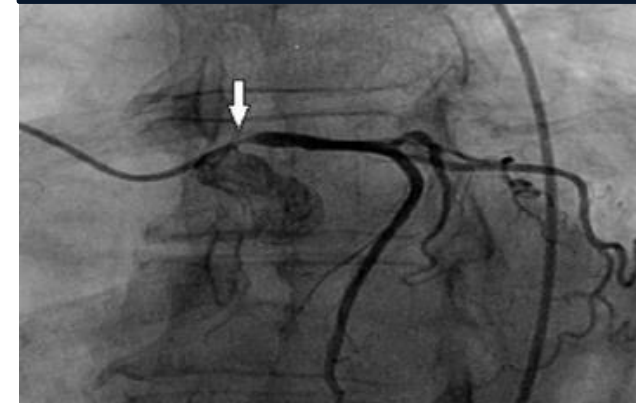
Have a High Suspicion for Coronary Stenosis After Arterial Switch



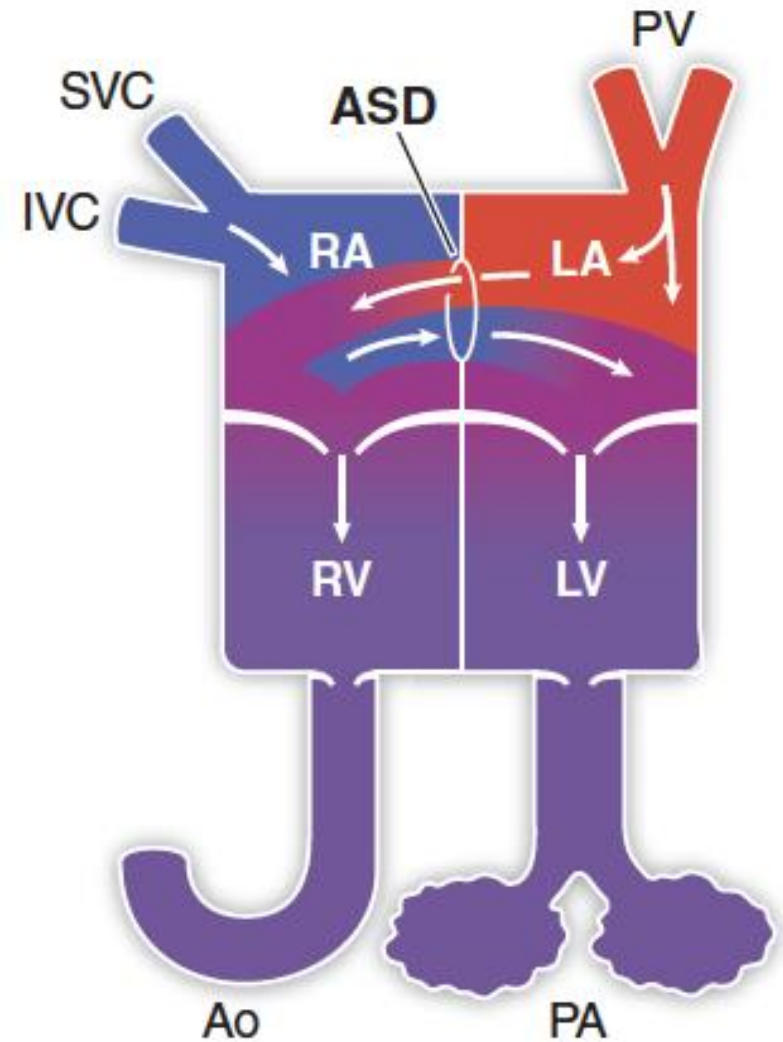
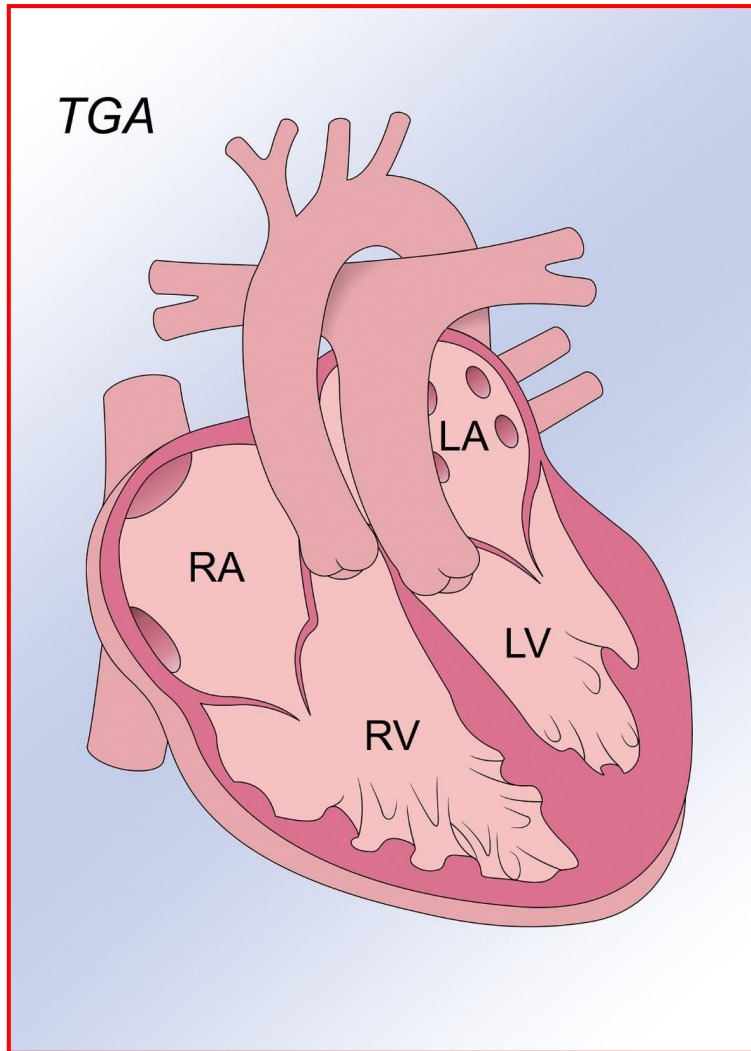
Arterial switch operation requires neonatal coronary re-implantation.

Coronary events most common early but can present in young adulthood

May present as angina, reduced EF, or sudden cardiac death

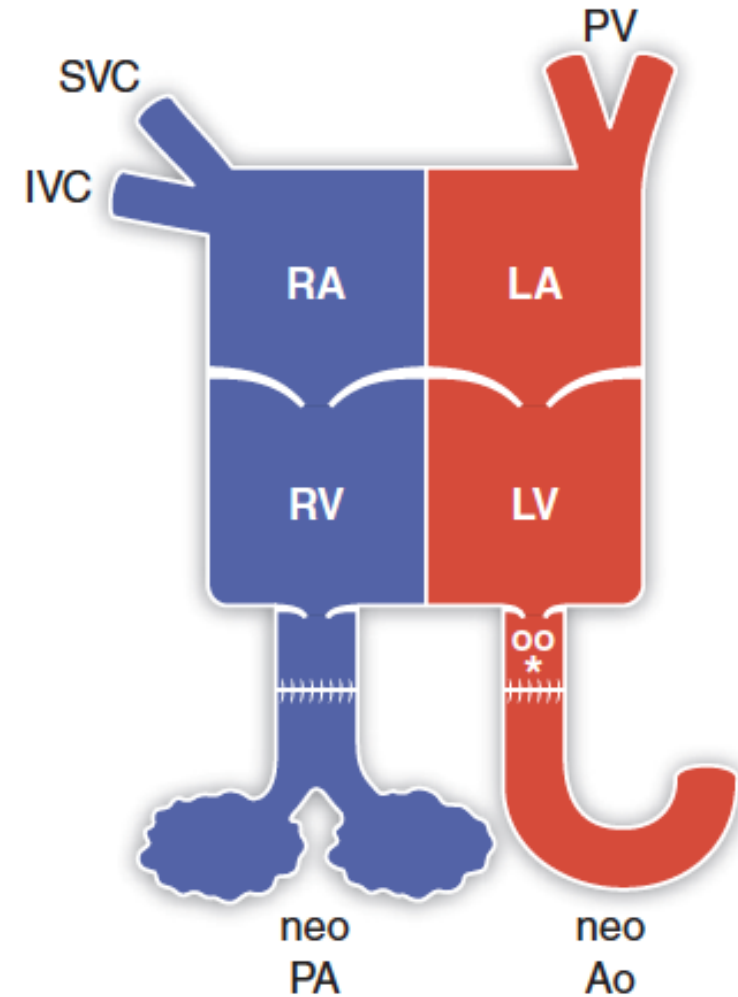


Transposition of the Great Arteries (TGA)



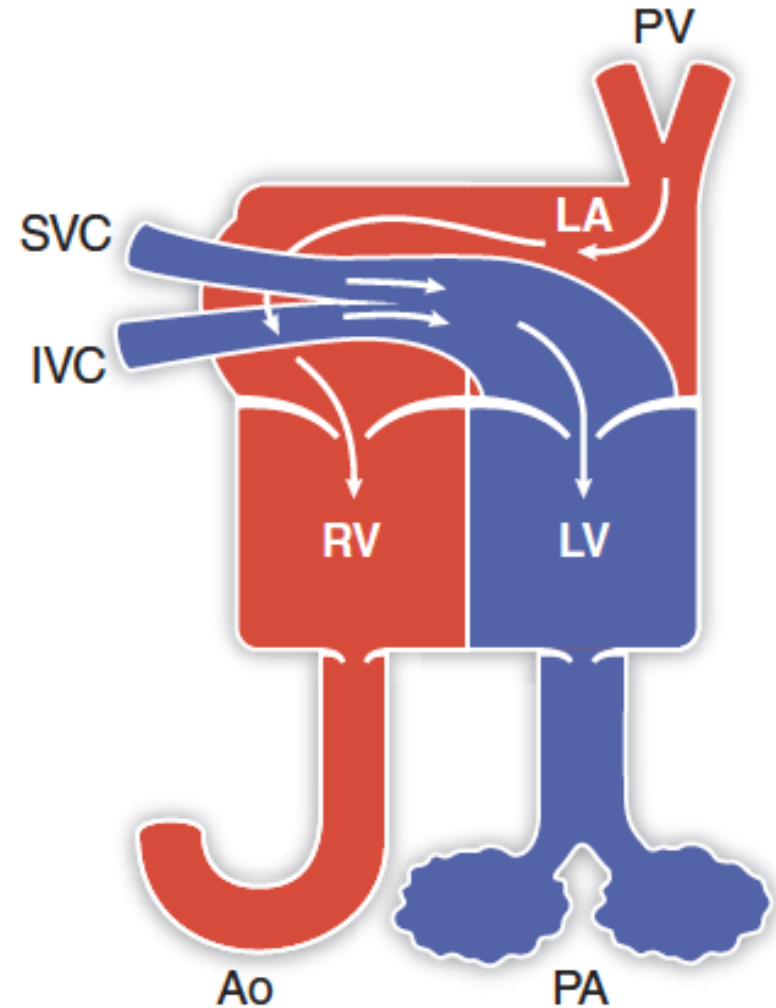
TGA s/p Arterial Switch

- Jatene procedure (1975)
- The great arteries are transected and switched – the coronaries are re-implanted
- Long-term complications:
 - Branch PA stenosis
 - Neo-aortic root dilation
 - Neo-aortic regurgitation
 - Coronary artery stenosis



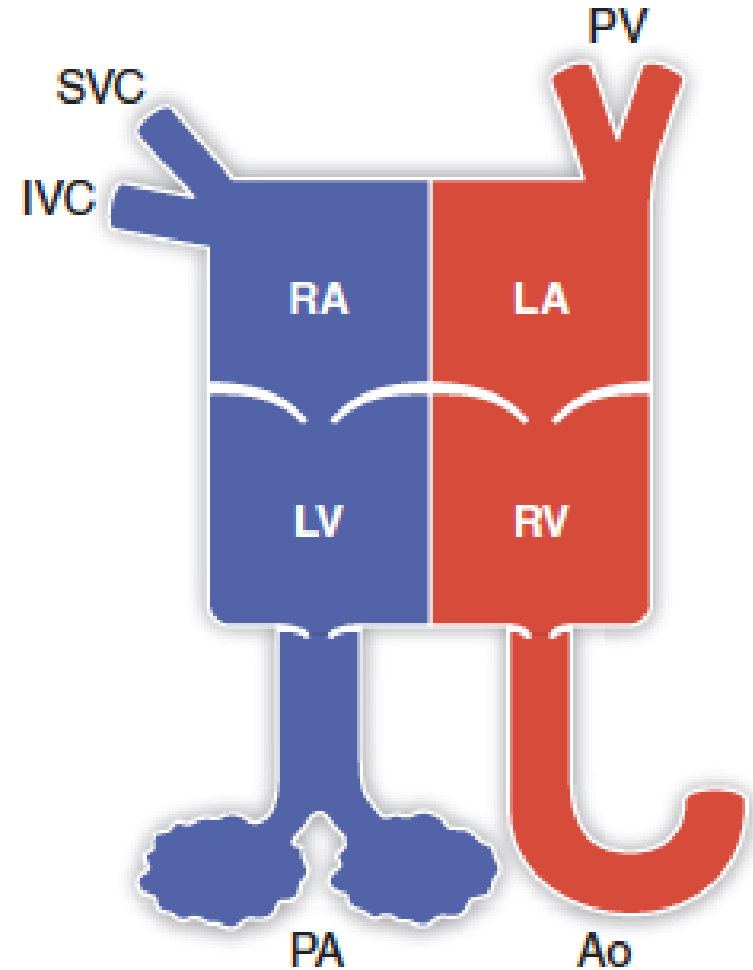
TGA s/p Atrial Switch (Older Patients)

- Senning procedure (1957)
- Mustard procedure (1963)
- Redirects blood flow to allow systemic venous return to get to the lungs
- Long-term complications:
 - **Systemic RV dysfunction**
 - Tricuspid regurgitation
 - Atrial baffle complications
 - Atrial arrhythmias



Congenitally Corrected Transposition of the Great Arteries (cc-TGA)

- AV and VA discordance
- Symptoms depend on associated lesions:
 - ASD
 - Tricuspid valve dysplasia \pm TR
 - Pulmonary stenosis
- 2% incidence of **complete heart block** per year in adulthood
- May be diagnosed in adulthood



MOC Reflective Statement

- Most adults with congenital heart disease need lifelong care
- Adults with surgical repairs in childhood may need interventions later in life
 - Tetralogy of Fallot: pulmonary valve replacement
 - Transposition of the Great Arteries:
 - older adults (atrial switch): arrhythmias, heart failure
 - Younger adults (arterial switch): coronary issues
- Congenital heart disease may present in adulthood:
 - Dilated right heart: atrial septal defects
 - Hypertension: aortic coarctation
 - Heart block: congenitally corrected transposition

References

- Stout K et al. 2018 AHA/ACC Guideline for the Management of Adults With Congenital Heart Disease. *Circulation*. 2019 Apr 2;139(14):e698-e800
- Valente AM, Landzberg MJ. Adult Congenital Heart Disease. Chapter 264. Harrison's Textbook of Internal Medicine, ed. Loscalzo J. McGraw-Hill, 2018.

Thank You

Boston Adult Congenital Heart (BACH) Program

Michael J. Landzberg, MD
Mary Mullen, MD
Michael N. Singh, MD
Fred Wu, MD
Michelle Gurvitz, MD
Fernando Baraona Reyes, MD
Amrit Misra, MD
Chrisopher Valle, MD
Caitlyn Joyce, PA-C
Nancy Barker, PA-C
Alex Alexander, PA-C
Siobhan Murray, PA-C
Susan Blensdorf, NP
Matthew Howley, RN
Hannah Smith, RN
Alexa Standley, RN
Jessica Joyce, MHA

Avihai Shimon, BA
Mildred Joseph
Autumn Scarcella
Daniella Morla
BACH Collaborators
Katherine E. Economy, MD
Sarah Rae Easter, MD
Jean Marie Carabuena, MD
Sitaram Emani, MD
Michael Kwon, MD
Margaret Holland, PA-C
Tom Tadros, MD
Ted O'Leary, MD
Akshay Desai, MD
Michael Givertz, MD
Jonathan Brown, PhD
Elazer R. Edelman, MD, PhD

Current and Former BACH Trainees

Patricia Frangini, MD
Ami B. Bhatt, MD
Yuli Y. Kim, MD
Gabriele Egidy Assenza, MD
Eric V. Krieger, MD
Alexander Opotowsky, MD
Anitra Romfh, MD
Sarah Partington, MD
Shailendra Upadhyay, MD
Ahmed Alomrani, MBBS
Yonatan Buber, MD
Keri Shafer, MD
Daniel Halpern, MD
Laith Alshawabkeh, MD
Saurabh Rajpal, MD
Matthew Carazo, MD
Valeria Duarte, MD
Shivani Aggarwal, MD
Ada Stefanescu-Schmidt, MD
Nael Aldweib, MD
Carla Rodriguez-Monserrate, MD
Allison Tsao, MD
Matthew Lippmann, DO
Tony Pastor, MD
Logan Eberly, MD
Christopher DeZorzi, MD
Charmaine Lam, MD
Michelle Moore-Padilla, MD
Maram Sati, MD
Madeline Duncan, BS
Vedang Diwanji, BS
Navya Krishna, BS

ACHD Question #5

A 32-year-old with developmental delay presents to clinic with symptoms of fatigue. He has a history of an unrepaired atrioventricular septal defect (AV canal defect). Laboratory results demonstrate hypothyroidism. Which genetic abnormality does he most likely have?

- a) Chromosome 22q11.2 deletion
- b) Chromosome X monosomy
- c) Chromosome 7q11.23 ELN mutation
- d) Chromosome 21 trisomy
- e) NOTCH1 mutation

ACHD Answer #5

A 32-year-old with developmental delay presents to clinic with symptoms of fatigue. He has a history of an unrepaired atrioventricular septal defect (AV canal defect). Laboratory results demonstrate hypothyroidism. Which genetic abnormality does he most likely have?

- a) Chromosome 22q11.2 deletion
- b) Chromosome X monosomy
- c) Chromosome 7q11.23 ELN mutation
- d) Chromosome 21 trisomy**
- e) NOTCH1 mutation