

# Introduction to Adult Congenital Heart Disease

David Laughrun, M.D., F.A.C.C.

# Disclosure of Relevant Financial Relationships

- Employee—Mercy Des Moines

# Adult Congenital Heart Disease

- **Definition**

- Examples
- Demographics
- Evolution of Specialty Care
- Unique Medical and Non-medical Concerns

# Adult Congenital Heart Disease

- Structural heart abnormality present at birth.
- Range from Simple to Moderately Complex to Highly Complex.
- Repaired or Unrepaired.
- Cyanotic or Non-cyanotic.

# Down Syndrome

- Trisomy 21.
- AV Canal Defect, VSD, ASD, TOF, or multiple defects.



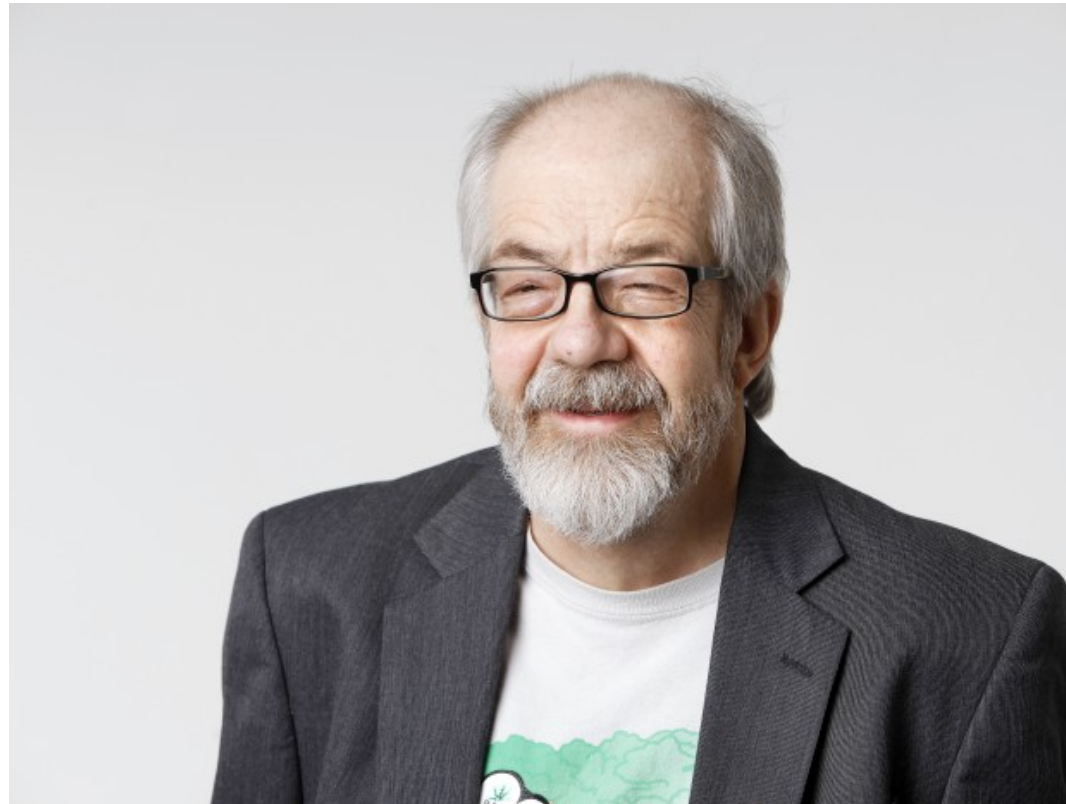
# Turner Syndrome

- Absent or abnormal X chromosome (~50% are 45XO).
- Coarctation, PAPVR.



# Noonan Syndrome

- Similar to Turner Syndrome but with normal complement of chromosomes. Mutated PTPN11 or KRAS gene. Autosomal dominant.
- PS, PAS, ASD.



# Holt-Oram (“Heart-Hand”) Syndrome

- Mutation of TBX5 gene; autosomal dominant.
- Abnormal radial, wrist, thenar and thumb bones.
- ASD, VSD(s), AV Block, atrial fibrillation.





# Adult Congenital Heart Disease: Etiology

- Genetic (complex patterns; approximately 25%).
- Environmental toxins (ETOH, maternal Rubella, drugs).
- Multifactorial (eg DM).
- *Majority of isolated cases of CHD have no apparent cause.*

## Diagnoses in Adult Patients With Simple Congenital Heart Disease\*

Native disease	Repaired conditions
Isolated congenital aortic valve disease	Previously ligated or occluded ductus arteriosus
Isolated congenital mitral valve disease (e.g., except parachute valve, cleft leaflet)	Repaired secundum or sinus venosus ASD without residua
Isolated patent foramen ovale or small atrial septal defect	Repaired ventricular septal defect without residua
Isolated small ventricular septal defect (no associated lesions)	
Mild pulmonary stenosis	
Small patent ductus arteriosus	

\*These patients can usually be cared for in the general medical community. Modified with permission from Connelly et al. Canadian Consensus Conference on Adult Congenital Heart Disease 1996. Can J Cardiol. 1998;14:395-452.

Wames, et al. J Am Coll Cardiol 2008;52. Table 5. Published ahead of print November 7, 2008, at <http://content.onlinejacc.org/cgi/content/full/jacc.2008.10.001>

## Diagnoses in Adult Patients With Congenital Heart Disease of Moderate Complexity\*

### Diagnosis

<b>Aorto–left ventricular fistulas</b>	<b>Pulmonary valve regurgitation (moderate to severe)</b>
<b>Anomalous pulmonary venous drainage, partial or total</b>	<b>Pulmonary valve stenosis (moderate to severe)</b>
<b>Atrioventricular septal defects (partial or complete)</b>	<b>Sinus of Valsalva fistula/aneurysm</b>
<b>Coarctation of the aorta</b>	<b>Sinus venosus atrial septal defect</b>
<b>Ebstein's anomaly</b>	<b>Subvalvar AS or SupraAS (except HOCM)</b>
<b>Infundibular right ventricular outflow obstruction of significance</b>	<b>Tetralogy of Fallot</b>
<b>Ostium primum atrial septal defect</b>	<b>Ventricular septal defect with: absent valve or valves, aortic regurgitation, coarctation of the aorta, mitral disease, right ventricular outflow tract obstruction, straddling tricuspid/mitral valve</b>
<b>Patent ductus arteriosus (not closed)</b>	

\*These patients should be seen periodically at regional adult congenital heart disease centers. Modified with permission from Connelly et al. Canadian Consensus Conference on Adult Congenital Heart Disease 1996. Can J Cardiol 1998;14:395–452.

AS indicates aortic stenosis; HOCM, hypertrophic obstructive cardiomyopathy; and SupraAS, supraaortic stenosis.

Warnes, et al. J Am Coll Cardiol 2008;52. Table 4. Published ahead of print November 7, 2008, at

<http://content.onlinejacc.org/cgi/content/full/jacc.2008.10.001>



## Types of Adult Congenital Heart Disease of Great Complexity\*

<b>Conduits, valved or nonvalved</b>	<b>Pulmonary atresia (all forms)</b>
<b>Cyanotic congenital heart (all forms)</b>	<b>Pulmonary vascular obstructive disease</b>
<b>Double-outlet ventricle</b>	<b>Transposition of the great arteries</b>
<b>Eisenmenger syndrome</b>	<b>Tricuspid atresia</b>
<b>Fontan procedure</b>	<b>Truncus arteriosus/hemitruncus</b>
<b>Mitral atresia</b>	<b>Other abnormalities of atrioventricular or ventriculoarterial connection not included above (i.e., crisscross heart, isomerism, heterotaxy syndromes, ventricular inversion)</b>
<b>Single ventricle (also called double inlet or outlet, common, or primitive)</b>	

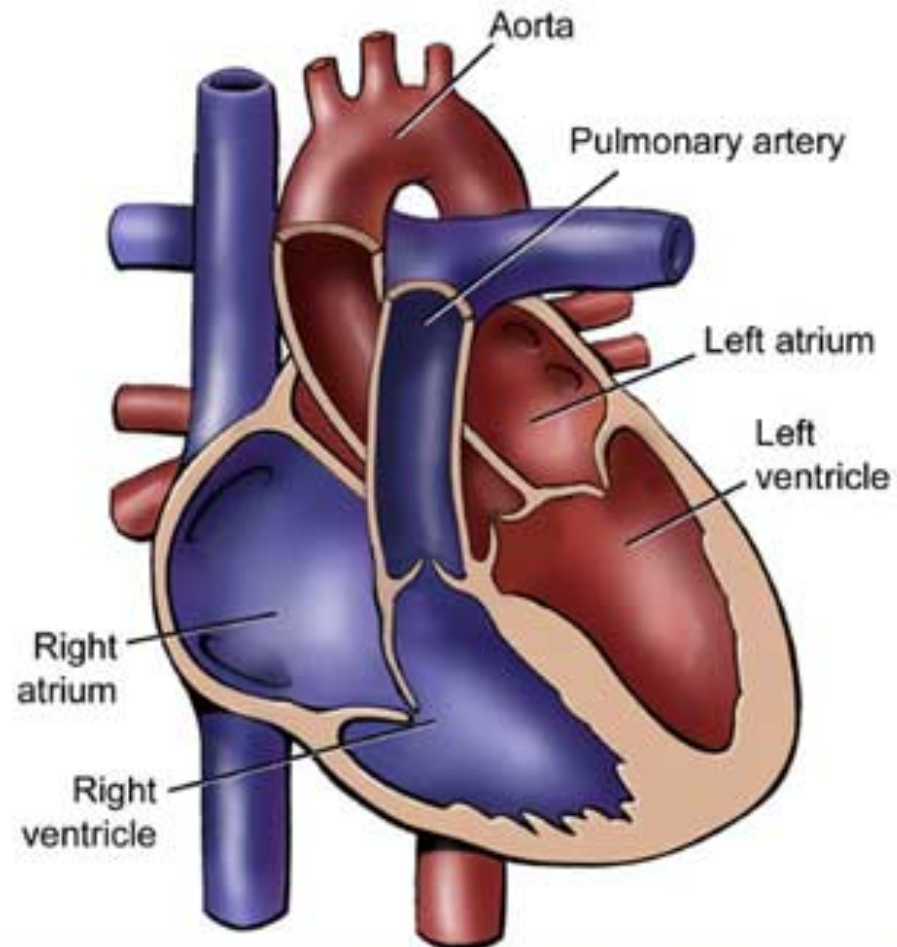
\*These patients should be seen regularly at adult congenital heart disease centers.

Modified from Connelly et al. Canadian Consensus Conference on Adult Congenital Heart Disease 1996. Can J Cardiol 1998; 14:395-452.

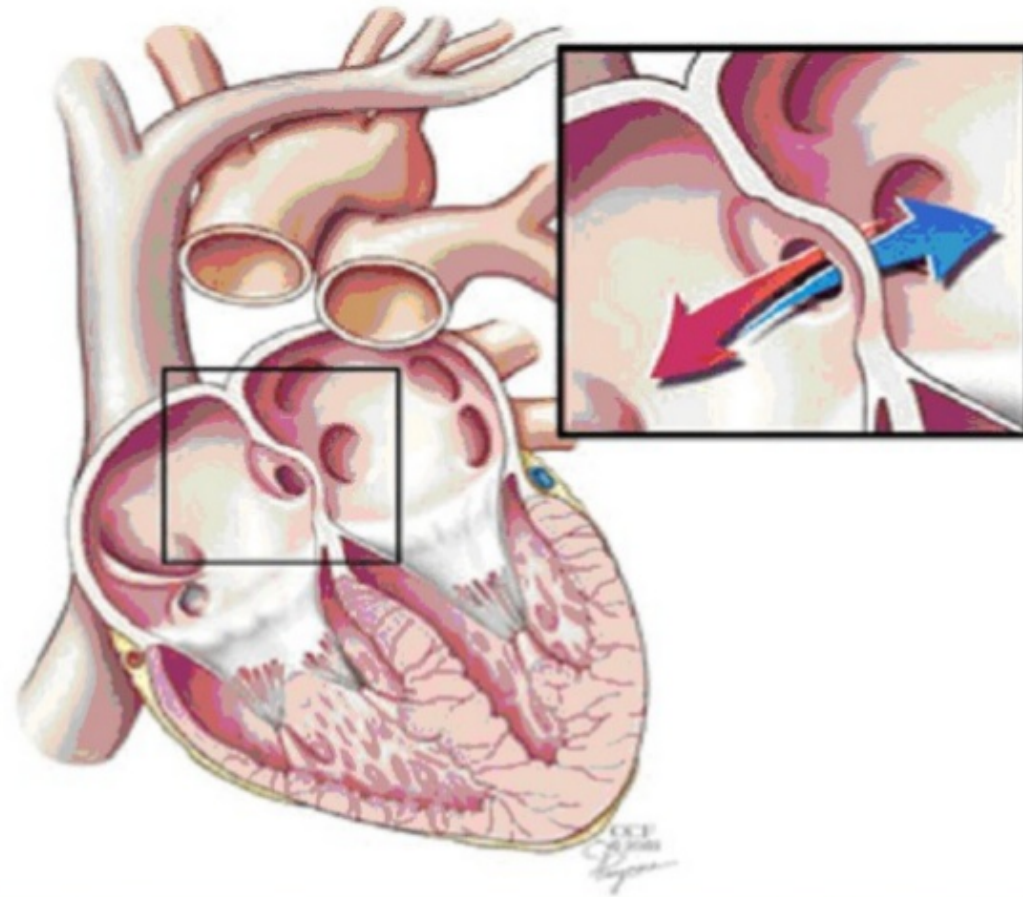
# Adult Congenital Heart Disease

- Definition
- **Examples**
- Demographics
- Evolution of Specialty Care
- Unique Medical and Non-medical Concerns

# Normal Heart

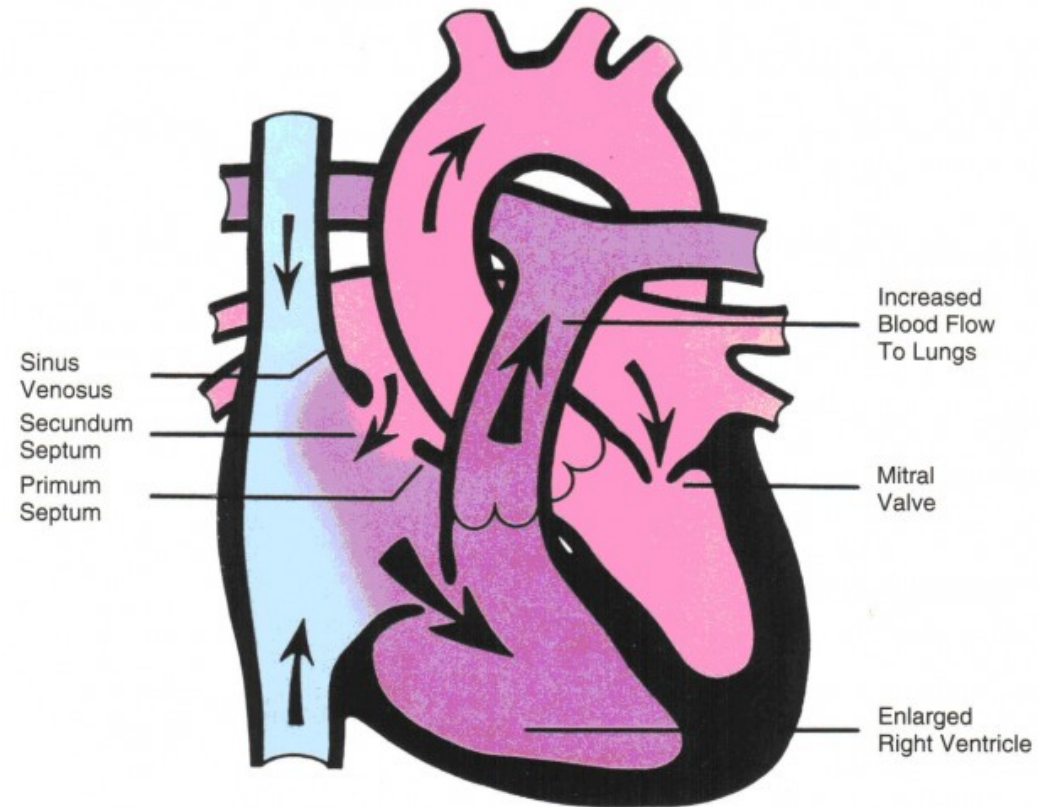


# ACHD-Simple: Secundum Atrial Septal Defect





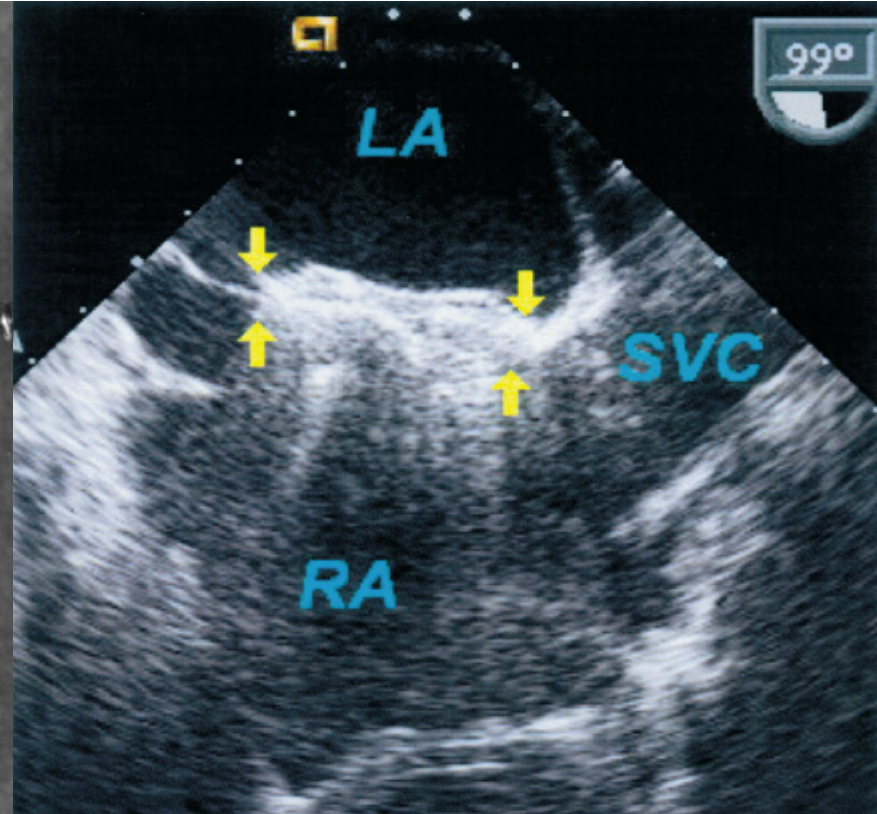
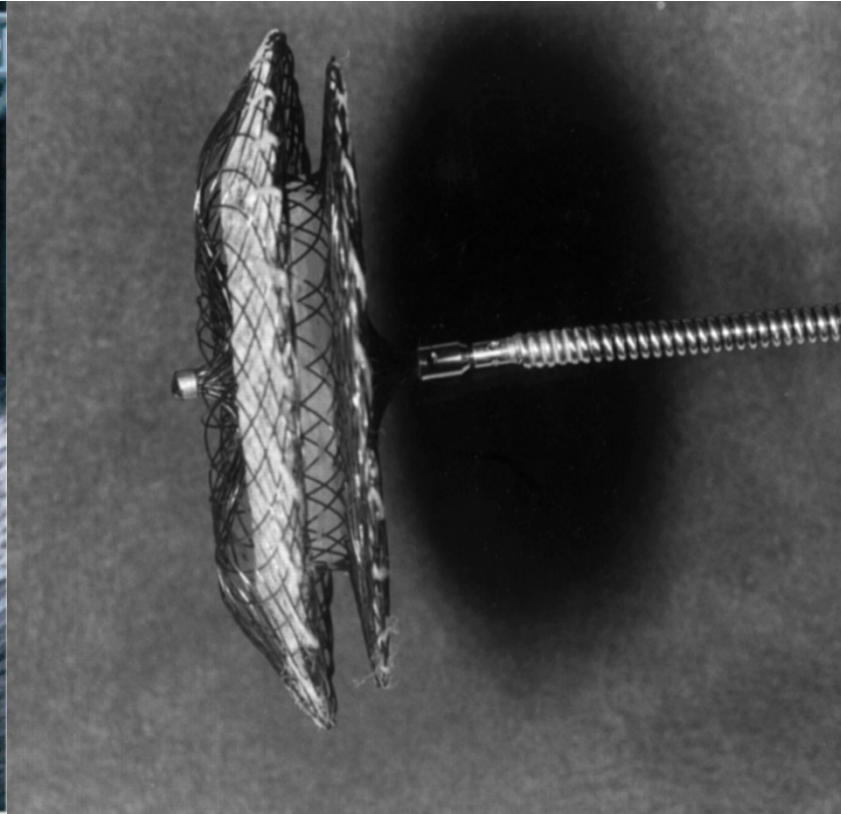
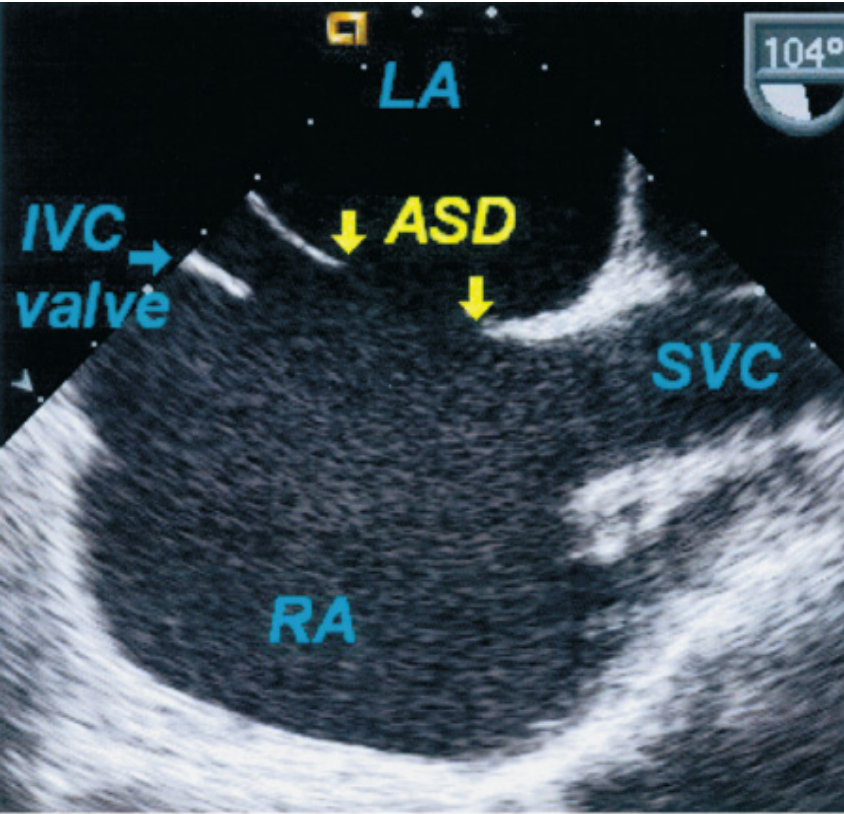
# Secundum ASD: Percutaneous or Surgical Closure if RA and RV Enlargement



**SECUNDUM ATRIAL SEPTAL DEFECT**



# Secundum ASD: Percutaneous Closure



**A**

Source: Fauci AS, Kasper DL, Braunwald E, Hauser SL, Longo DL, Jameson JL, Loscalzo J: *Harrison's Principles of Internal Medicine*, 17th Edition: <http://www.accessmedicine.com>  
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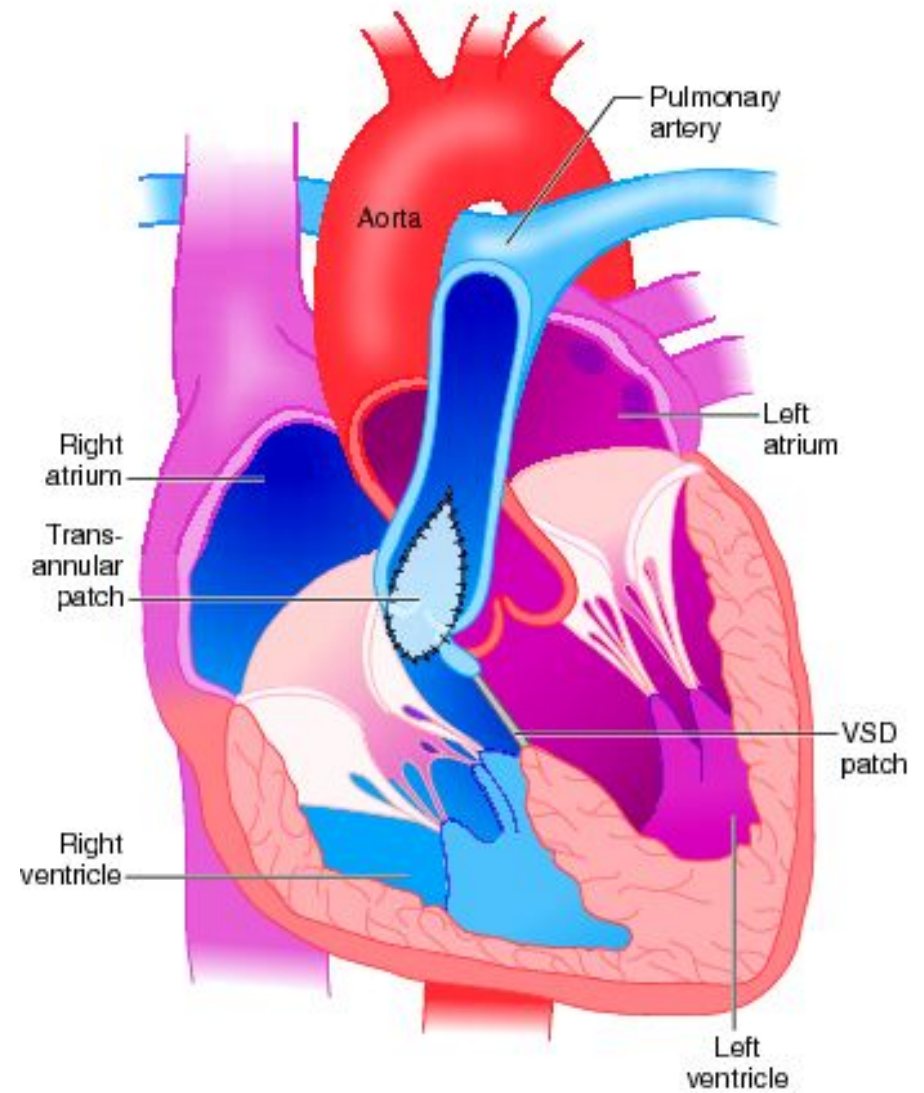
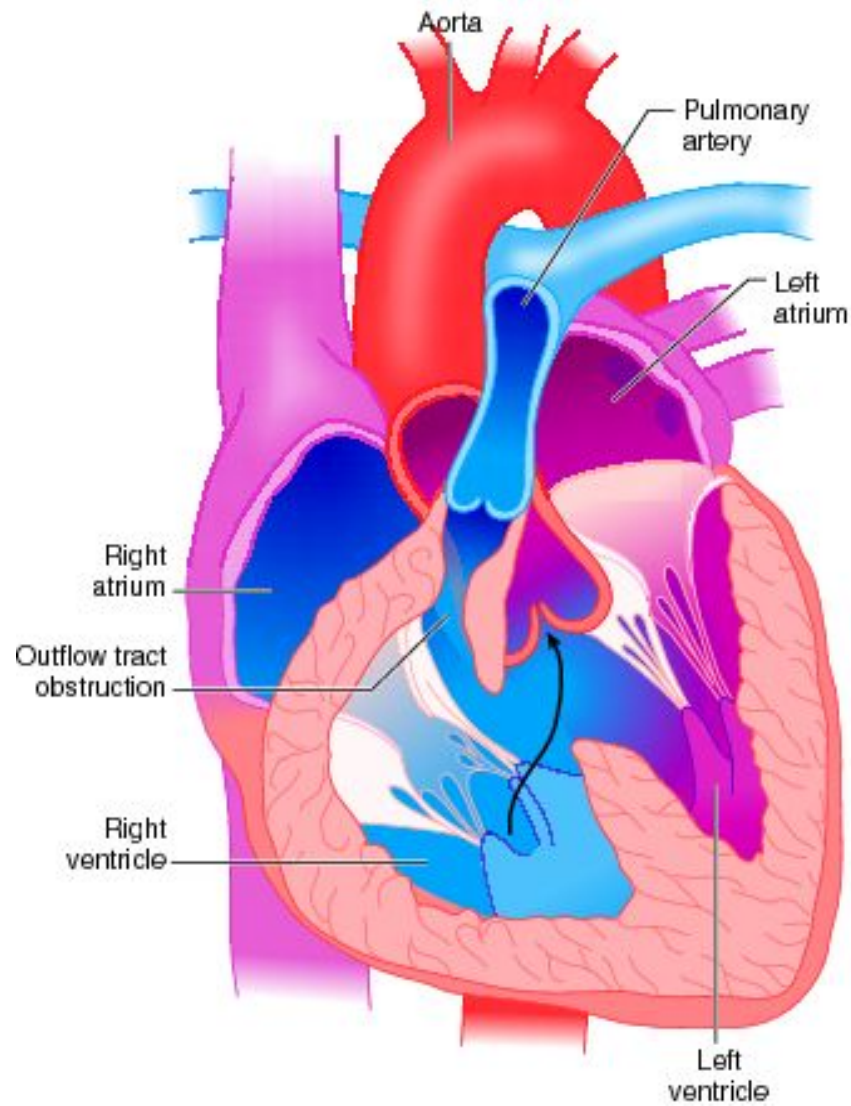
**B**

Source: Fauci AS, Kasper DL, Braunwald E, Hauser SL, Longo DL, Jameson JL, *Harrison's Principles of Internal Medicine*, 17th Edition: <http://www.accessmedicine.com>  
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# Repaired Secundum ASD Followup

- **Percutaneous Closure:** 3 months to 1 year then “periodically” thereafter. Watch for: migration, erosion, thrombosis.
- **Surgical Closure:** Indefinite followup if adult at time of surgery, pulmonary HTN, arrhythmias, RV dysfunction or associated lesions.

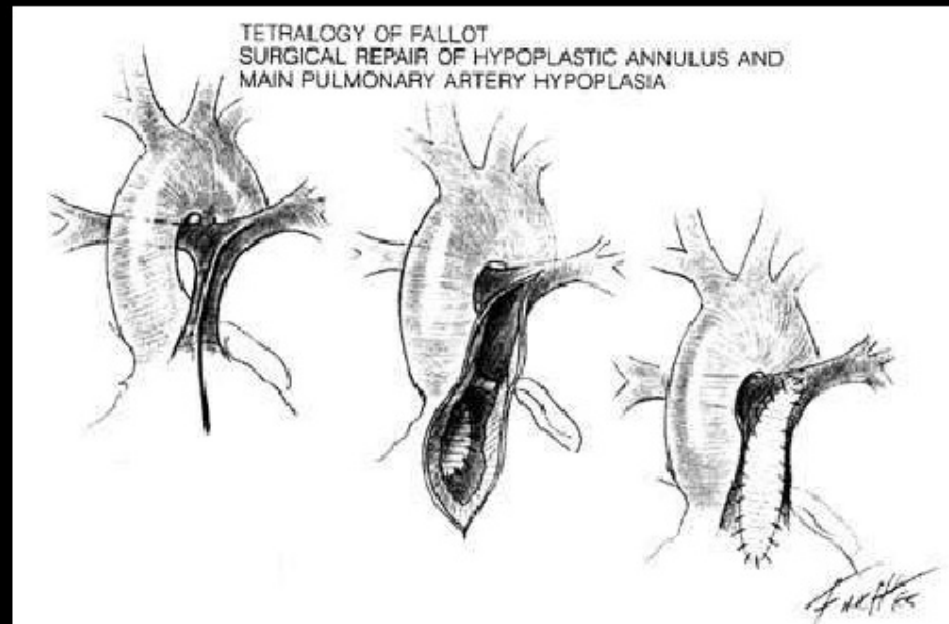
# ACHD-Moderately Complex: Repaired Tetralogy of Fallot



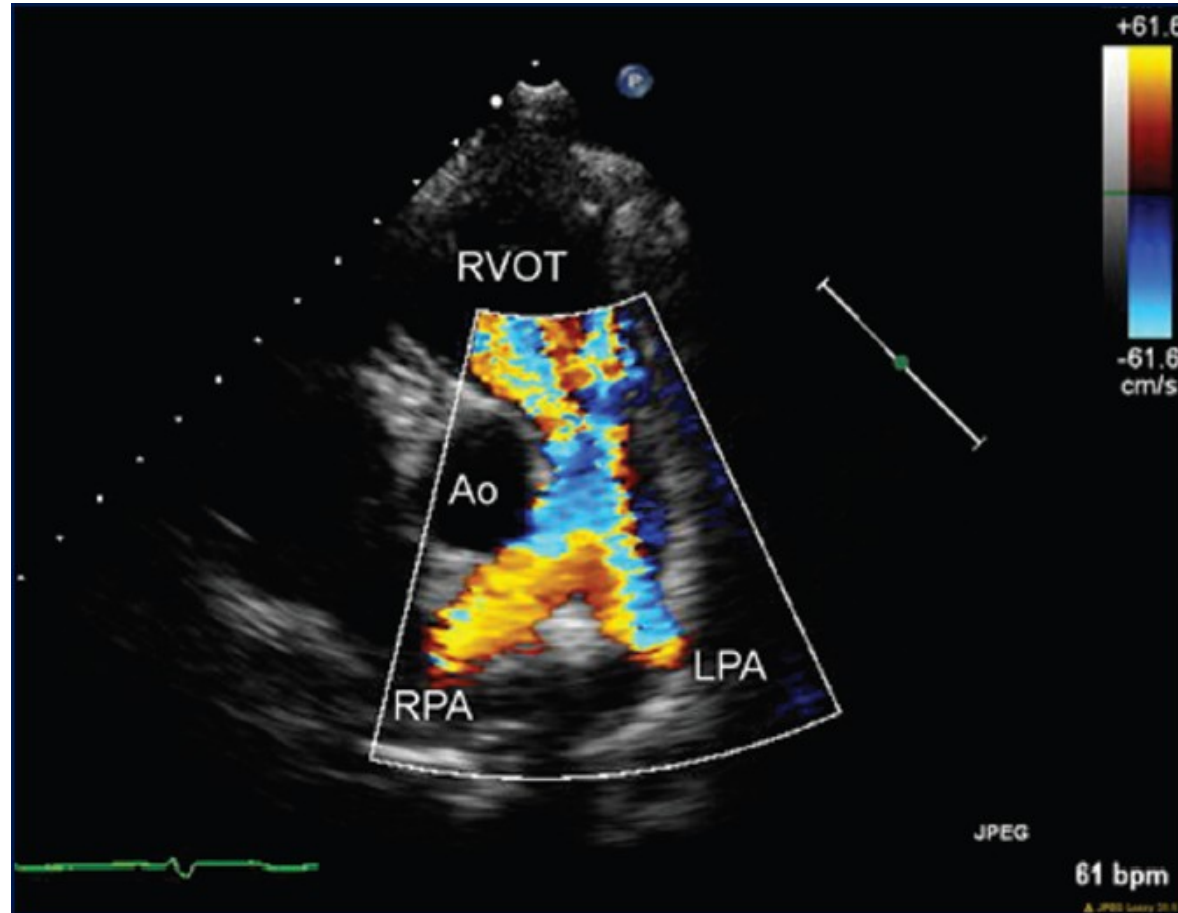


# TOF Repair of RVOT and PA Obstruction

## Surgical repair: Tof F



# Pulmonary Regurgitation s/p TOF Repair

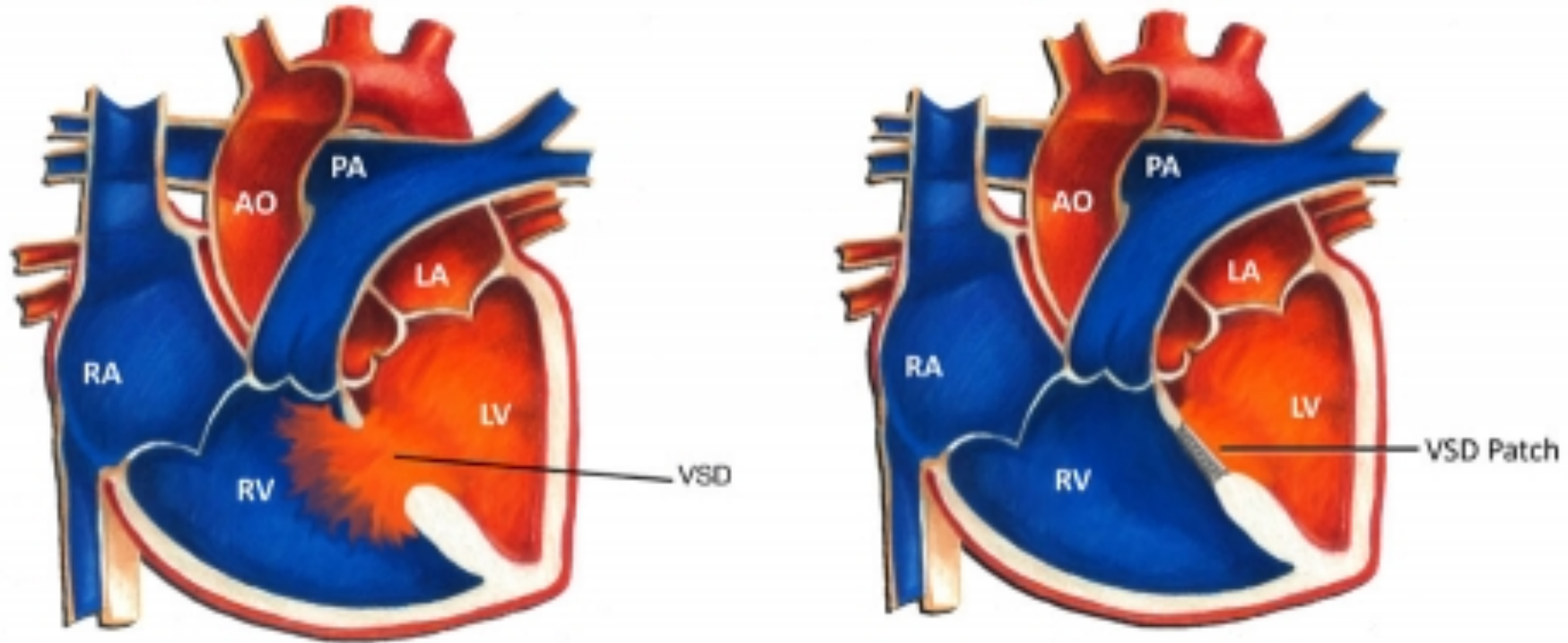


# Repaired TOF Patient: Long Term Followup

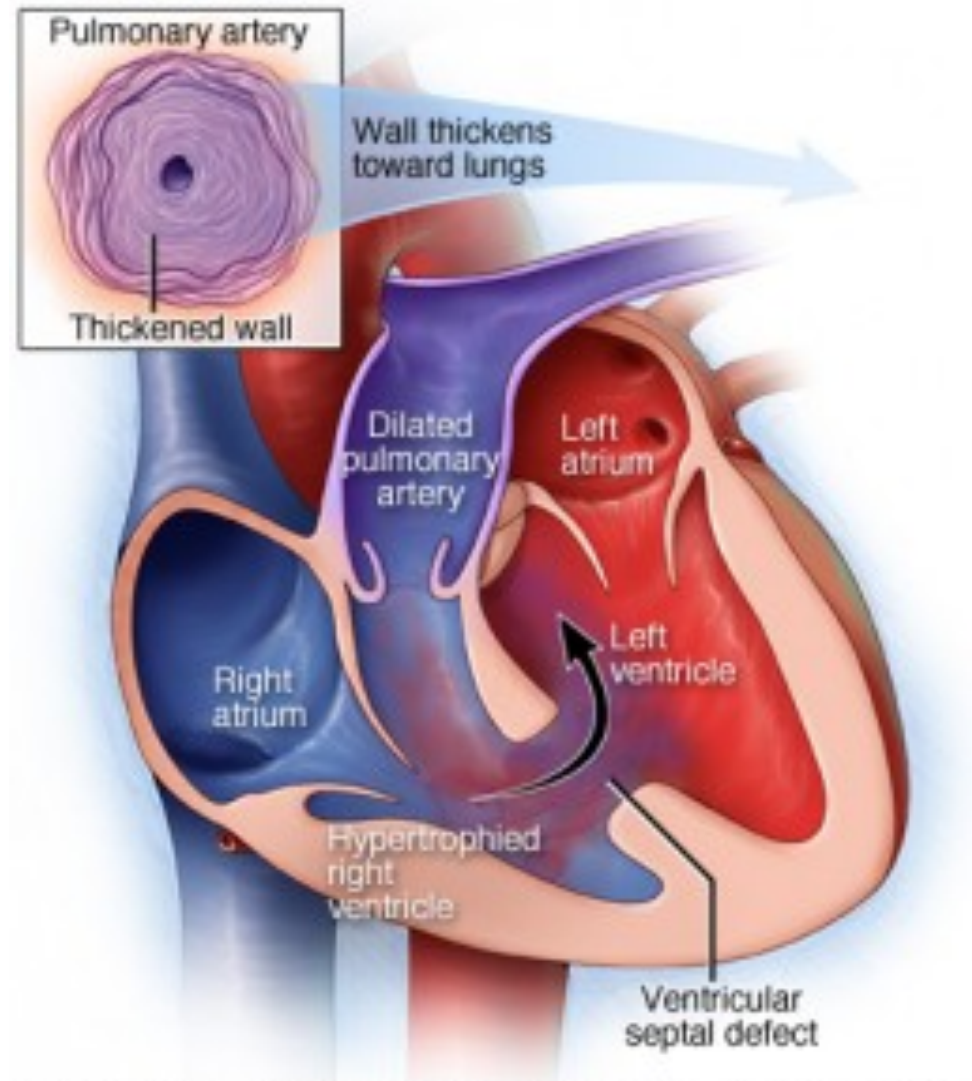
- Re-operation for pulmonic regurgitation, residual RVOT obstruction or aortic regurgitation?
- Heritable cause (eg: 22q11 deletion)?
- Arrhythmias?
- Increased risk of SCD.
- Favorable long term prognosis (86% 30 years postop) but:
- *Repaired TOF is not cured TOF.*

# ACHD-Highly Complex: VSD with Eisenmenger Physiology

- VSD corrected *before* Eisenmenger physiology develops has an excellent long term outlook.



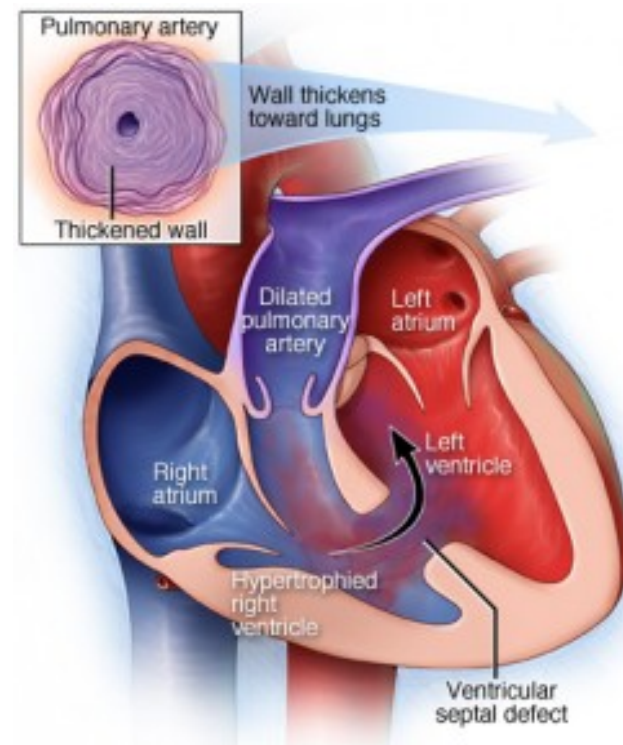
# VSD with Eisenmenger Physiology





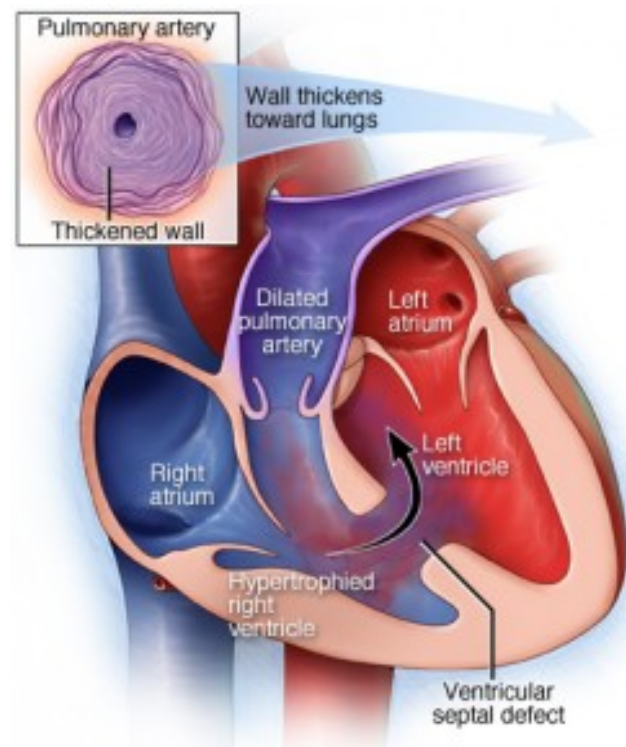
# VSD with Eisenmenger Physiology

- Initial L to R shunt leads to medial thickening of pulmonary vasculature.
- Resultant increase in pulmonary vascular resistance ultimately leads to R to L shunt.
- O<sub>2</sub>-unresponsive hypoxemia results.



# VSD with Eisenmenger Physiology

- When PVR exceeds 70% of SVR due to irreversible changes in the pulmonary vasculature, the risk of surgical repair of the VSD becomes prohibitive due to the likelihood of postoperative death from RV failure.



# Eisenmenger Syndrome



# VSD with Eisenmenger Physiology

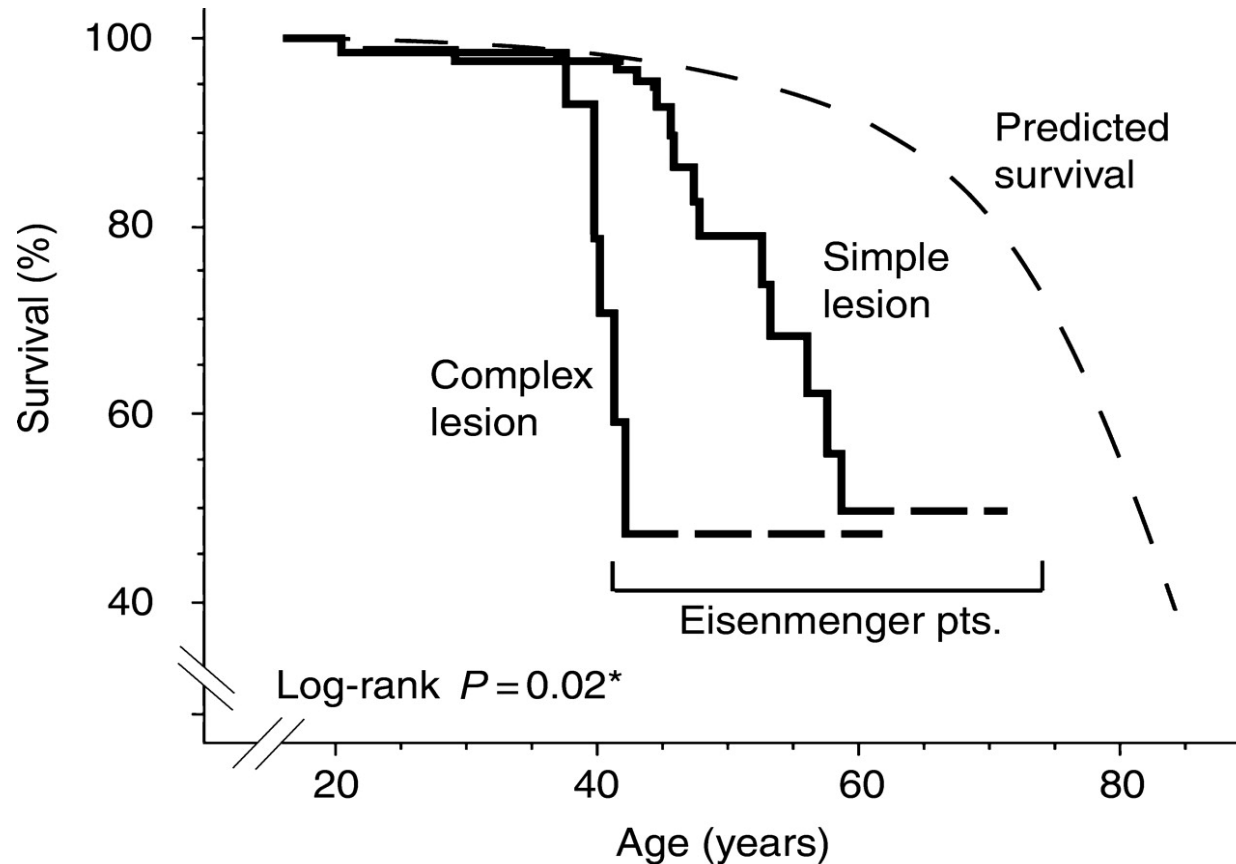
- Progressive dyspnea on exertion.
- Secondary erythrocytosis and iron deficiency can lead to hyperviscosity problems (cerebrovascular, renal).
- Right heart failure.
- Paradoxical embolism.
- Angina (RV ischemia or coronary artery compression by dilated PA).
- Death from: SCD, hemoptysis, HF, pregnancy, non-cardiac surgery, brain abscess, infectious endocarditis, stroke.

# VSD with Eisenmenger Syndrome: Management

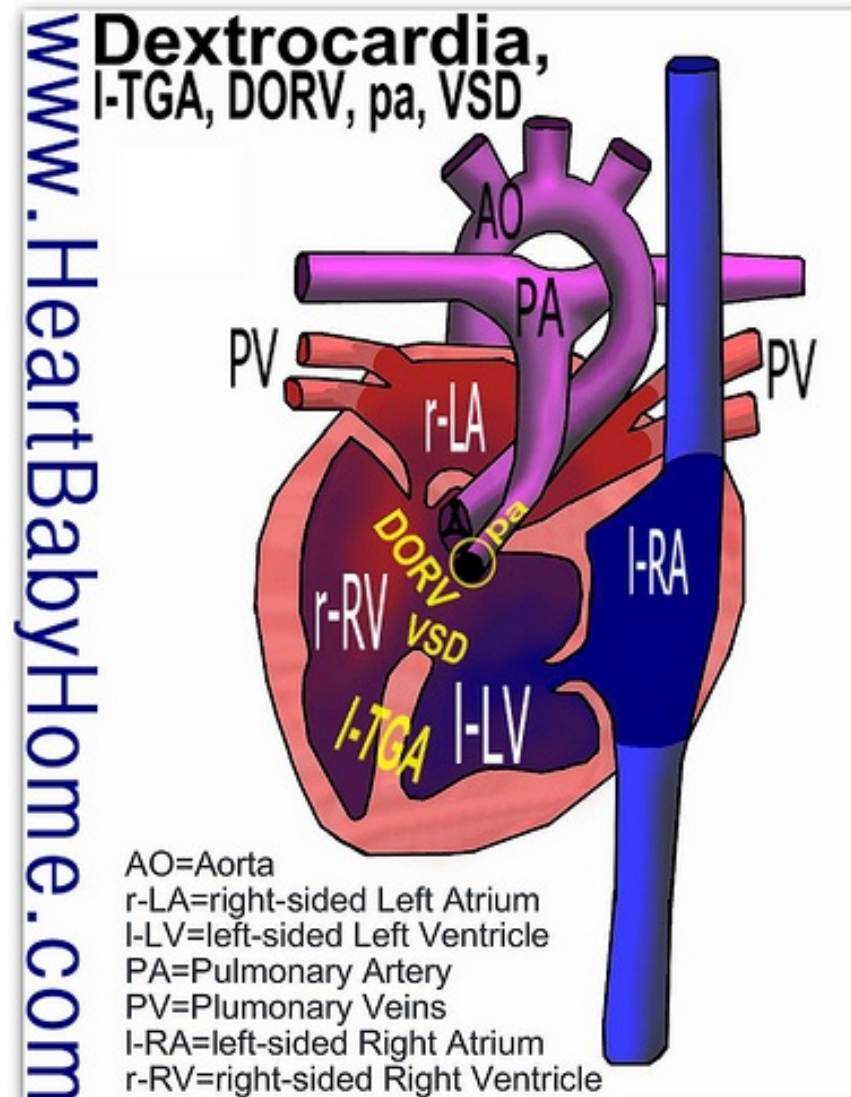
- Absolute avoidance of pregnancy.
- Avoid: air bubbles in IV, dehydration, moderate or greater exercise (especially isometric), excessive heat, high altitude.
- Maintain adequate iron stores.
- Uncommonly use therapeutic phlebotomy (Hb>20 with symptoms).
- Medical treatment of PAH.
- Consider heart-lung transplant or VSD repair-lung transplant.

# VSD with Eisenmenger: Transplant Considerations

- 10 year survival s/p HLT approximately 20%
- Without HLT:

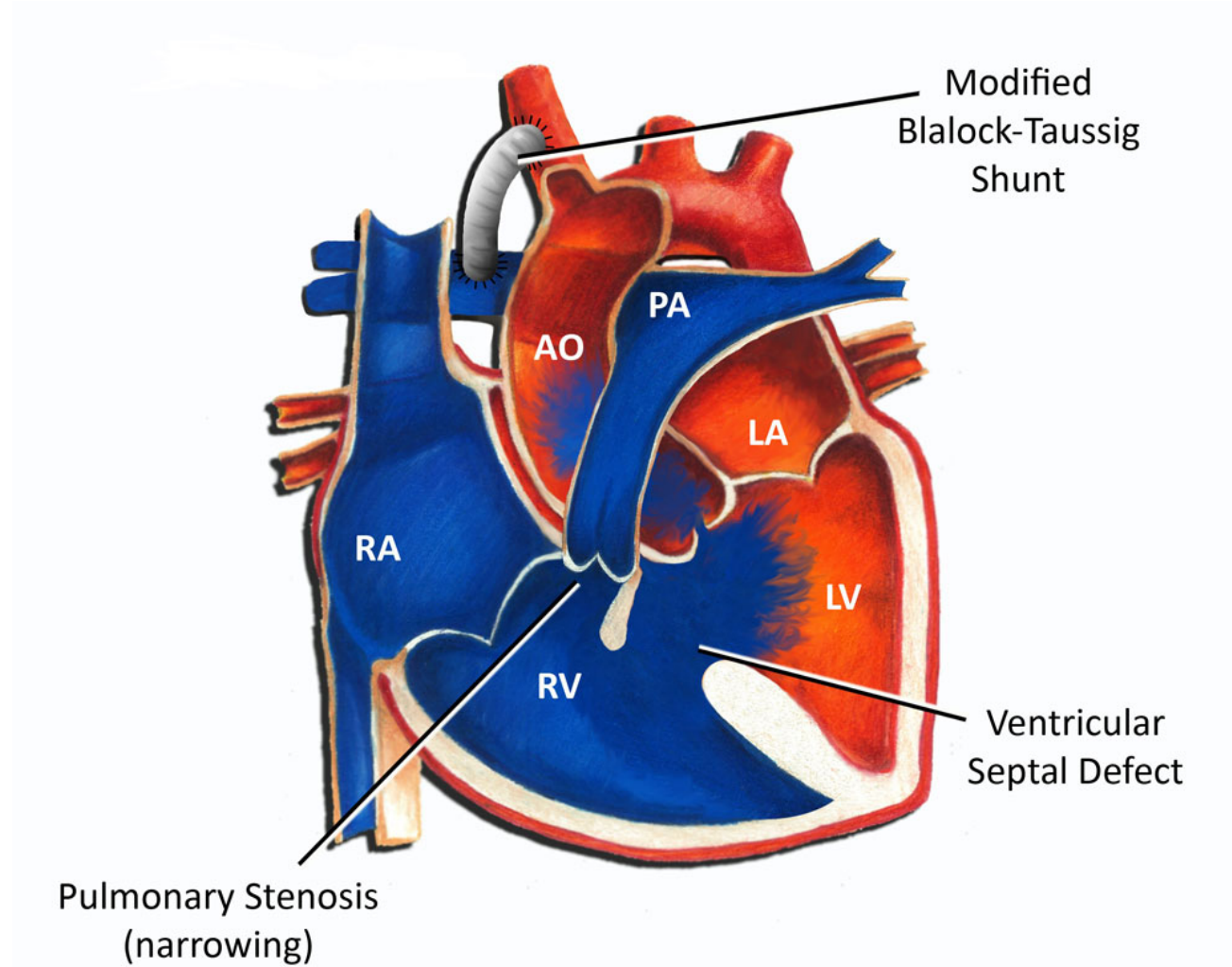


# ACHD-Highly Complex: Dextrocardia, DORV, VSD, L-TGA, Pulmonary Atresia



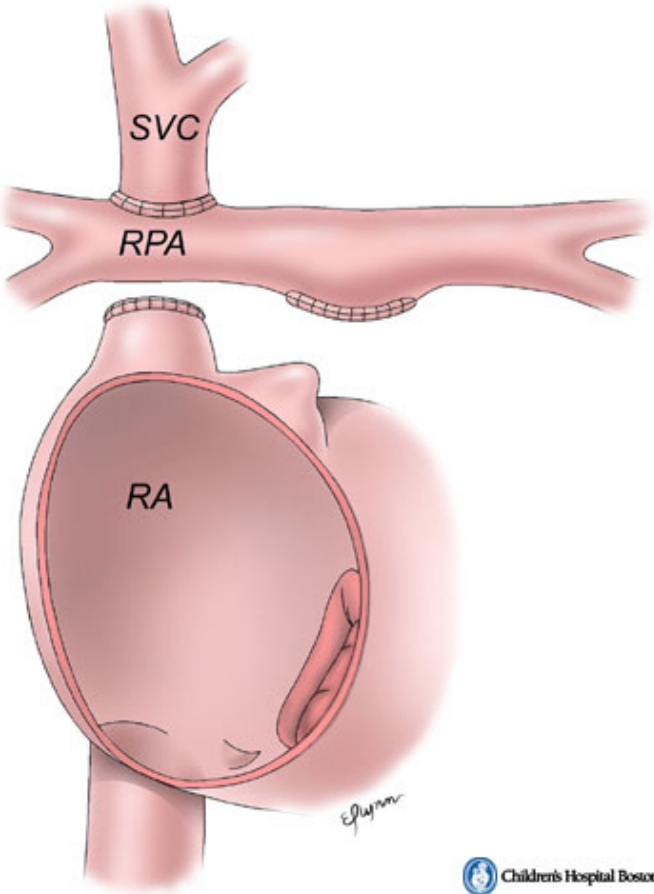


# Modified Blalock-Taussig Shunt: Subclavian Artery to Pulmonary Artery



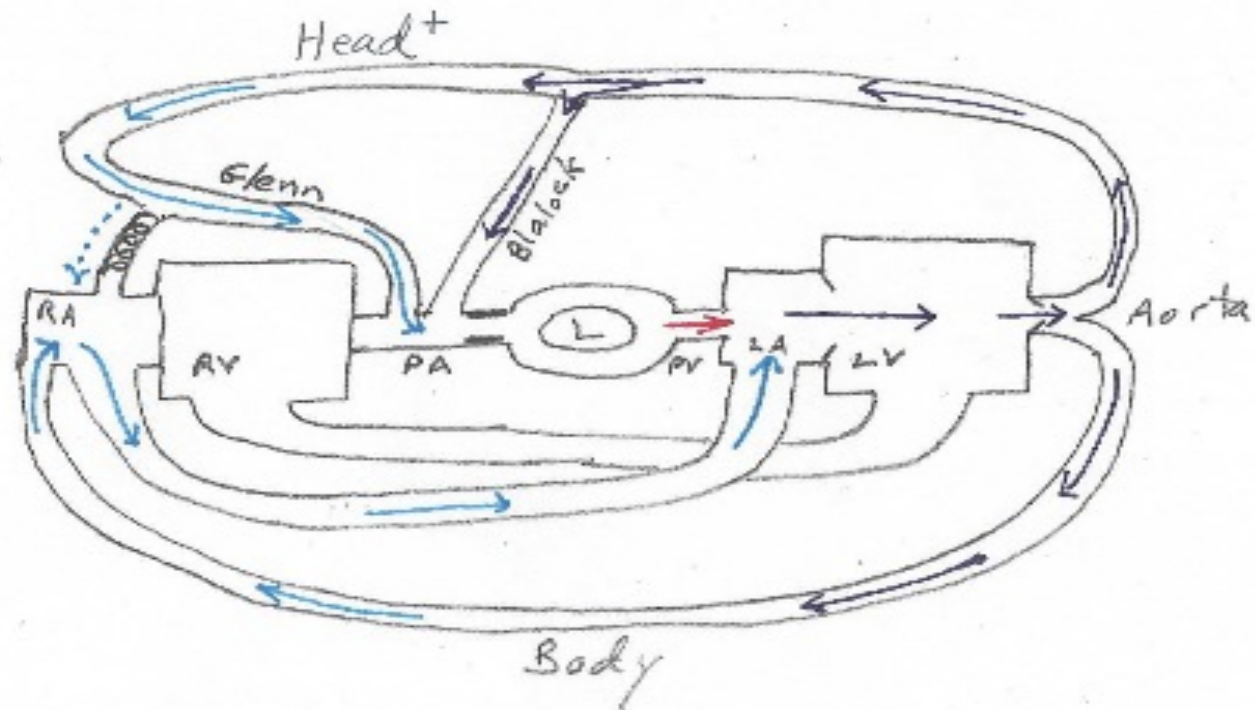
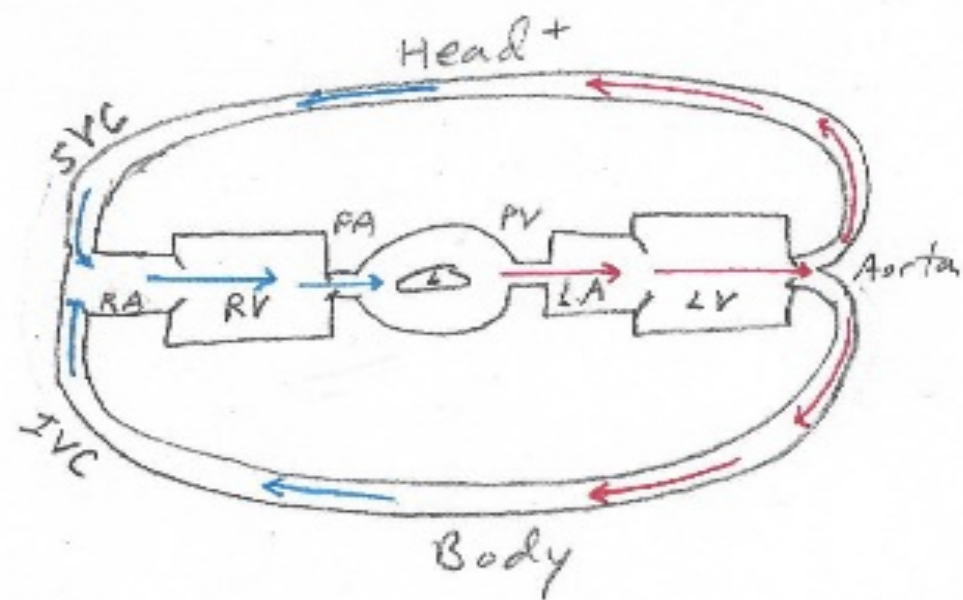


# Glenn Shunt: Superior Vena Cava to Pulmonary Artery



# ACHD-Highly Complex: Dextrocardia, DORV, VSD, L-TGA, Pulmonary Atresia

- Infant: L Glenn Shunt
- Infant: R Blalock-Taussig Shunt
- 11 y.o.: R sided unifocalization surgery with bovine pericardial graft.
- 12 y.o.: Patch closure of morphologic R AV valve + excision of interatrial septum.
- 23 y.o.: Successful pregnancy.
- 27 y.o.: Pulmonary artery stent + coiling of Glenn “pop-off” collaterals.
- 30 y.o.: Coiling of new Glenn collaterals.



- No interatrial septum
- VSD
- Pulmonary atresia
- Closed R AV Valve

Although Vivien Thomas (Mos Def), a black man in the 1930s, is originally hired as a janitor, he proves himself adept at assisting the "Blue Baby doctor," Alfred Blalock (Alan Rickman), with his medical research. When Blalock insists that Thomas follow him to Johns Hopkins University, they must find a way to skirt a racist system to continue their study of infant heart disease. Thomas is indispensable to Blalock's progress, but Blalock is the only one who is allowed to receive the acclaim.



A breakthrough that changed  
the face of medicine.  
A unique partnership that broke  
the rules.

Alan Rickman                      Mos Def

# SOMETHING THE LORD MADE

HBO FILMS    SUNDAY, MAY 30, 9PM    HBO

# Vivien Thomas

- Instructor of Surgery and Honorary Doctorate Johns Hopkins University 1976.



# Adult Congenital Heart Disease

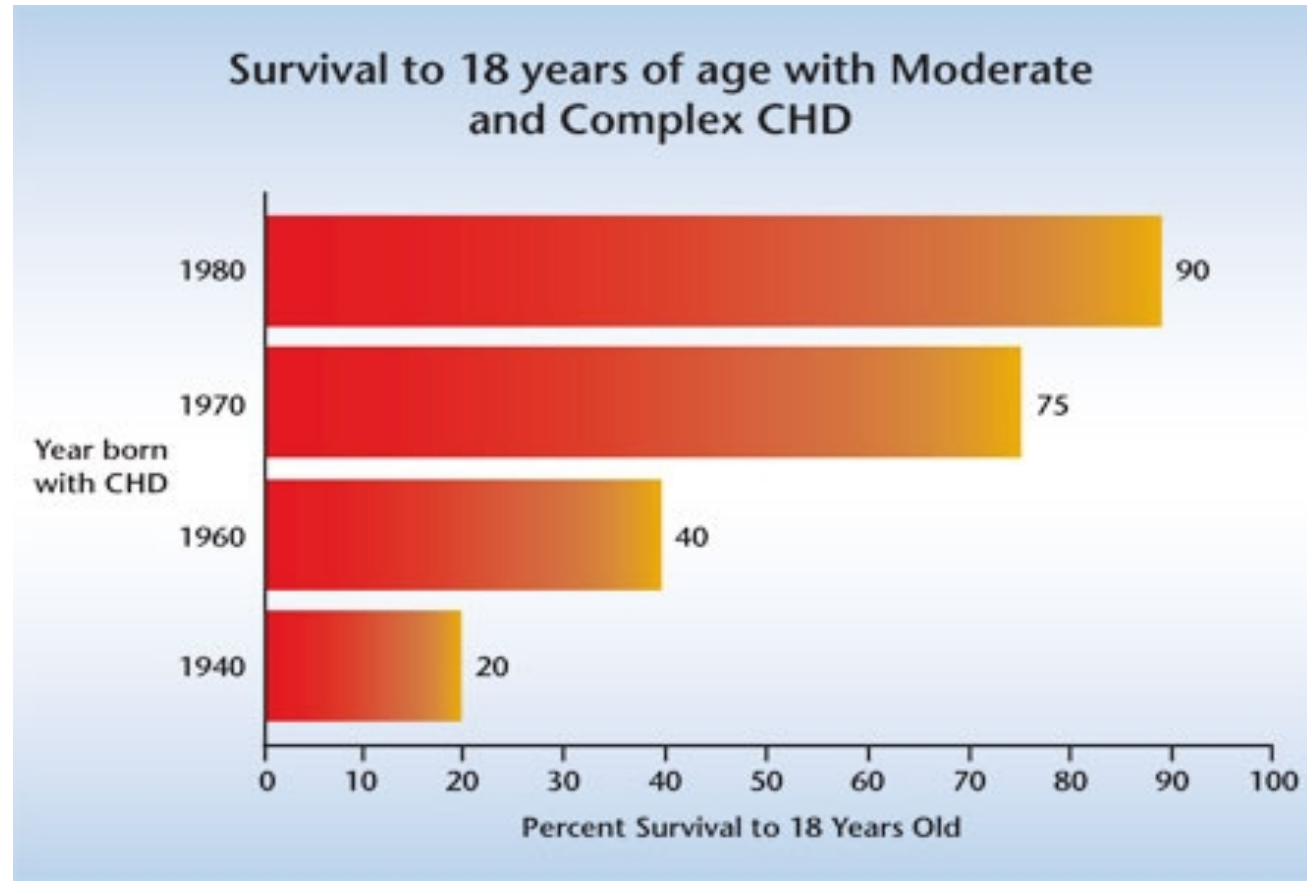
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# ACHD-Demographics

- Approximately 1 in 100 births have some form of heart defect.
- In 1960, <40% survived to adulthood.
- Today, >90% survive to adulthood.
- >1 million adults in U.S. living with CHD.
- ACHD population growing at an estimated 5% per year.

# Improved CHD Survival

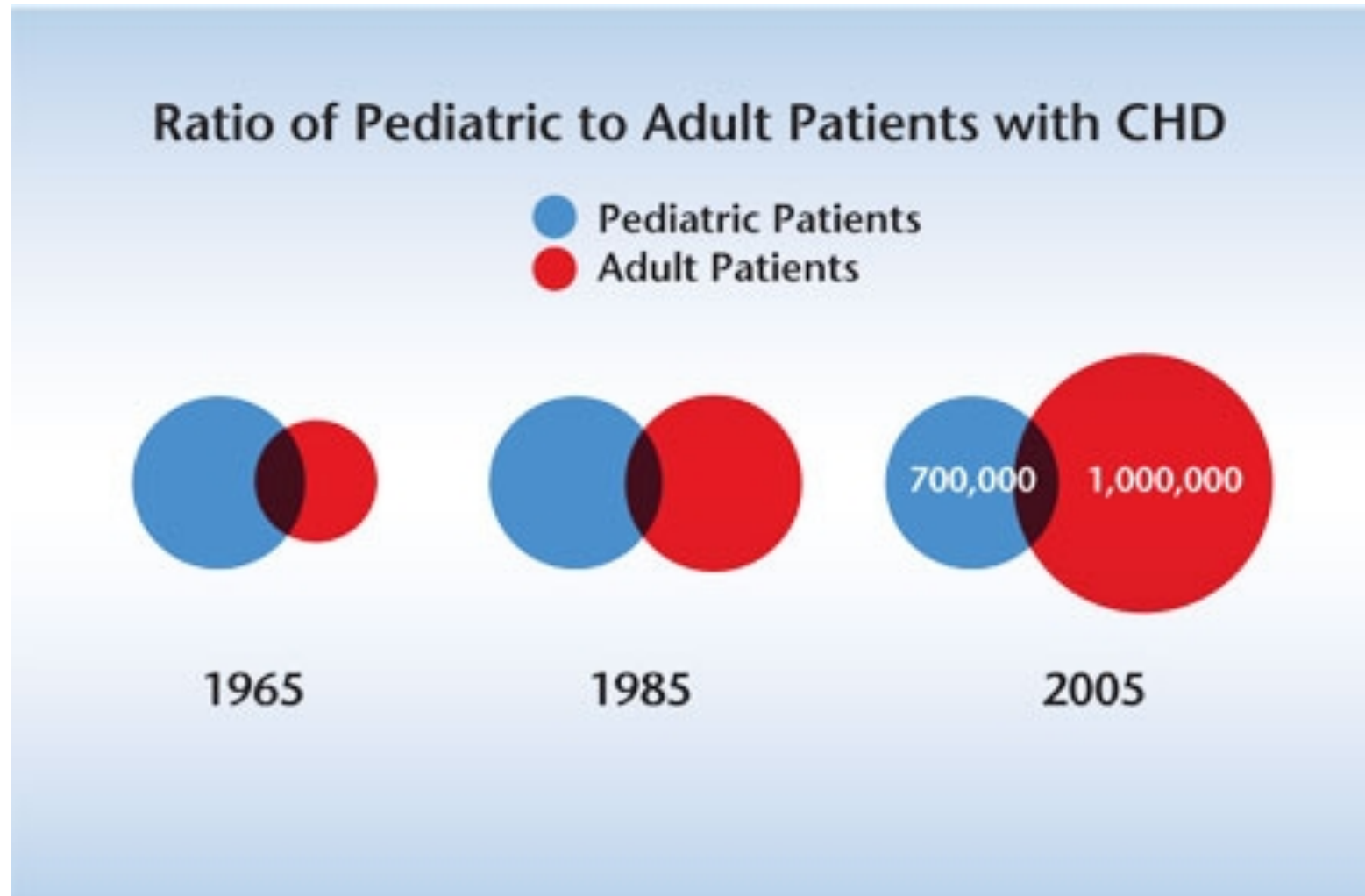
- Improved imaging and early diagnosis.
- Improved surgical and interventional techniques.
- Advances in critical care and EP.





# ACHD-Demographics

- More adults than children are now living with CHD.



# Adult Congenital Heart Disease

- Definition
- Examples
- Demographics
- **Evolution of Specialty Care**
- Unique Medical and Non-medical Concerns



# ACHD-Evolution of Specialty Care

- Estimated >50% of CHD patients are lost to follow-up after adolescence. Only 10% receive subspecialty care.
- Adult Cardiology Fellowships require only 6 hours of lecture training in CHD.
- In 2012, 76% of Pediatric Cardiologists surveyed cited a lack of qualified ACHD providers.
- *Currently there are many patients with too few specialists and programs to take care of them.*

# ACHD-Evolution of Specialty Care

- **“Bethesda 32” 2000:** ACC concludes the U.S. is not meeting the needs of adults with CHD. Recommends ACHD Centers.
- **ACC Guidelines 2008:** Specific personnel and services recommended for ACHD Centers. Disease specific guidelines for the care of adults with CHD.
- **ABIM October 2015:** First offering of Board Exam in ACHD.
- ABIM has applied to ACGME for **accreditation of postgraduate training programs** in the U.S.
- **ACHA 2015:** begin process of **accrediting ACHD Centers.**

# Adult Congenital Heart Disease

- Definition
- Examples
- Demographics
- Evolution of Specialty Care
- **Unique Medical and Non-medical Concerns**

# Unique Concerns for ACHD

- Congenital syndromes.
- Endocarditis, brain abscess.
- Endocarditis prophylaxis
- Secondary erythropoiesis with iron deficiency.
- Noncardiac surgery risk.
- Depression and anxiety.
- Hemostasis.
- Renal function.
- Gallstones.
- Pulmonary vascular disease.
- Restrictive lung disease.
- Orthopedic/rheumatologic disease.
- Varicose veins.
- Hepatic congestion/cirrhosis.
- Thromboembolic disease.
- Protein losing enteropathy.
- ?Operation, re-operation, intervention, transplant.

- Insurance.
- Medical records.
- Career choice.
- Finances.
- Transition of Care

- Mortality
- Pregnancy and Contraception
- Exercise and Sports
- Arrhythmias and risk of SCD



# Exercise and Sports

- Symptoms account for only 30% of all barriers to exercise.
- Other barriers: lack of experience with exercise in childhood, fear, coexisting disabilities, cultural attitudes.
- Provider should emphasize what *to do* for exercise and de-emphasize restrictions.
- “Bethesda 36” Guidelines available for competitive athletics. Competition may hinder prudent recognition of symptoms.
- No such guidelines for noncompetitive exercise.

# Regular, Moderate, Symptom-Limited Exercise

(Braunwald)

- Reduces cardiovascular morbidity and mortality in CAD patients.
- Improves functional capacity, quality of life and risk factors in patients with HTN, valvular heart disease and chronic heart failure.
- Most individuals with structural heart disease can safely participate in prescribed physical activity.

# Exercise

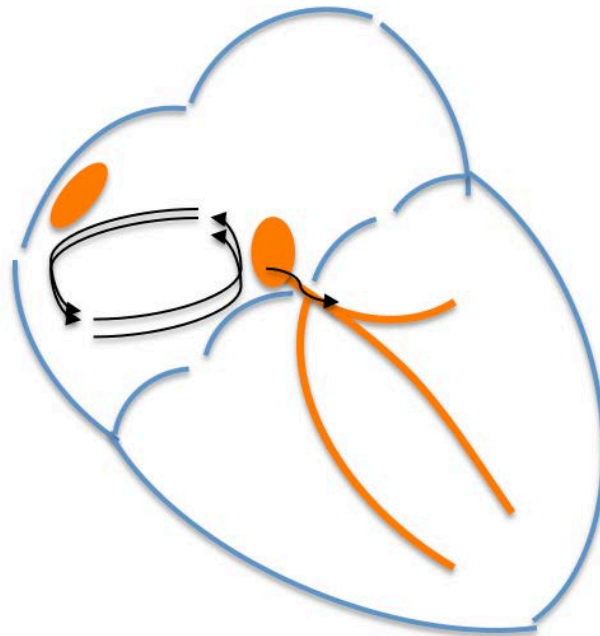
- “Progress gradually and pay attention to your symptoms.”
- Stop if chest discomfort, lightheaded, heart racing, or short of breath to point you can't talk.
- Goal 30+ minutes every day of moderate symptom limited exercise.
- Caveats for: Marfan's, cyanotic CHD, aortic stenosis, coarctation, devices. (Avoidance of isometric exercise, high intensity sports and contact sports).

# Arrhythmias

- Symptomatic arrhythmias are the most frequent reason for hospital admission in adults with CHD.
- Hemodynamic stress, structural abnormalities, scars, patches, and accessory pathways all contribute to the high incidence of tachy and brady arrhythmias.

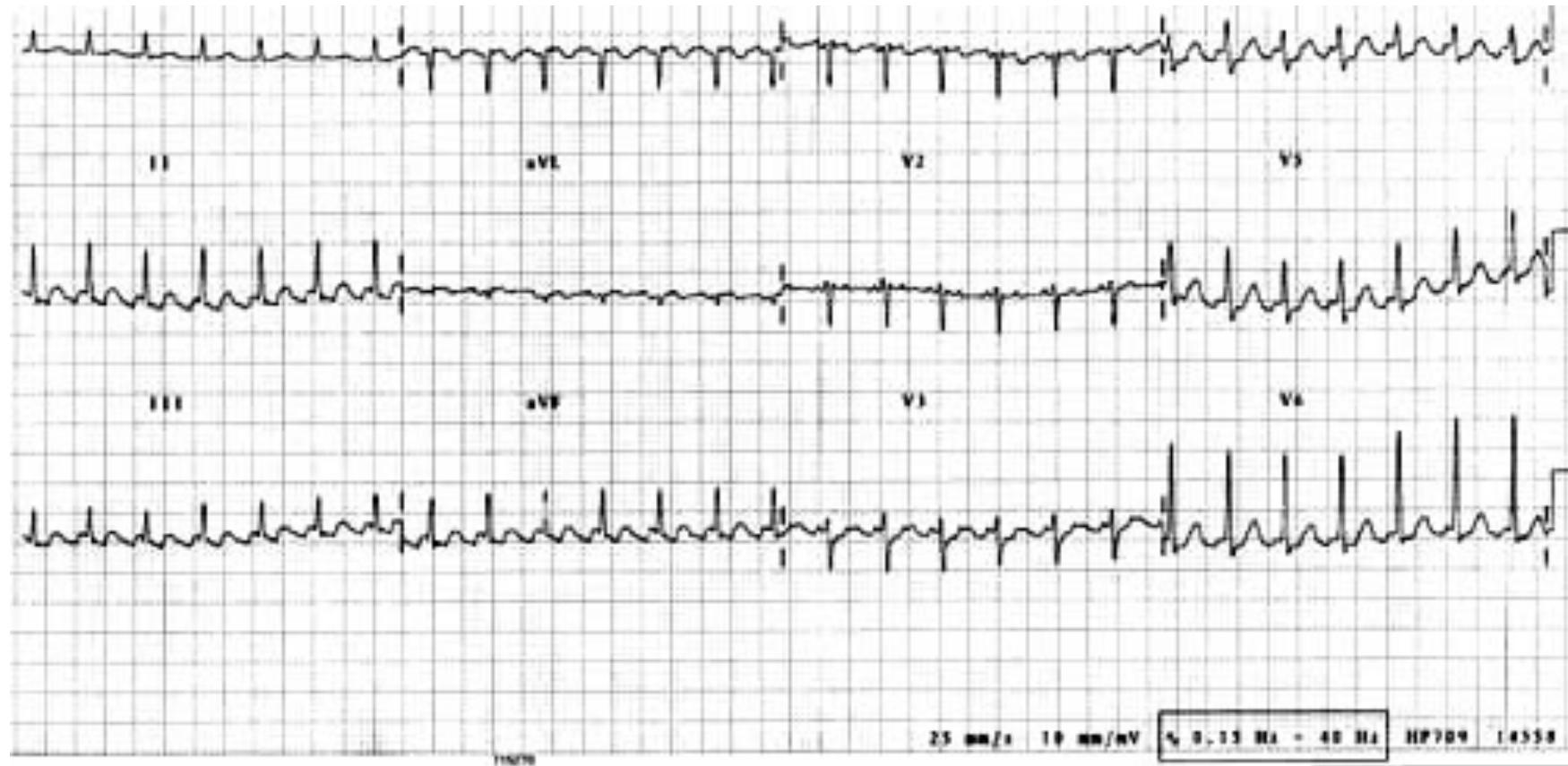
# Arrhythmias: IART

- Intra-Atrial Re-entrant Tachycardia (IART) is seen in up to 50% of patients in long term follow up after surgery involving the RA and/or LA due to macro reentrant circuits.
- 170-250 bpm (vs 300 bpm for typical atrial flutter). Can conduct 1:1 to ventricles and cause syncope or even SCD.
- Pharmacologic Rx disappointing. Consider ATP, atrial ICD, ablation.



# Typical Atrial Flutter with 2:1 AV Conduction

A rate = 300, V rate = 150



# IART (s/p atrial switch) with 1:1 AV Conduction

A rate = 190, V rate = 190

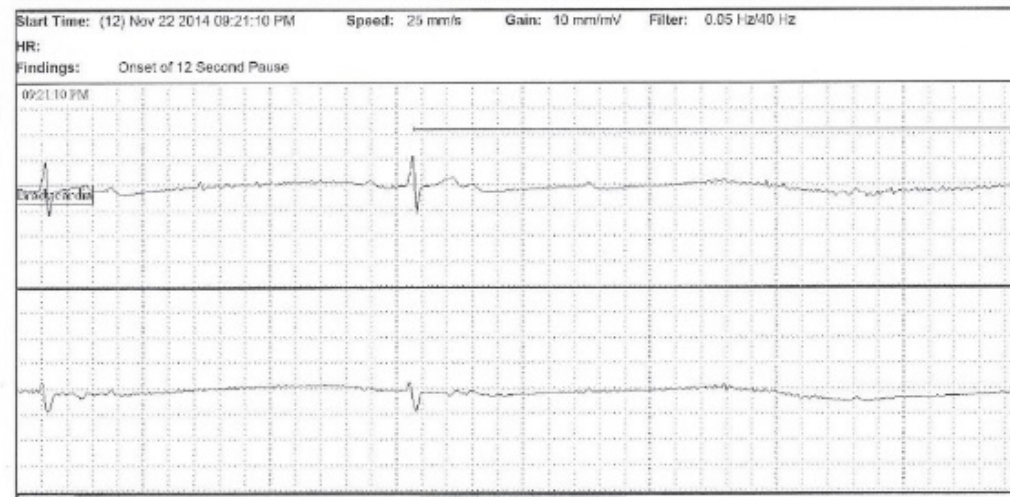




# Bradyarrhythmias in ACHD

- Sinus node damage after surgeries involving the atria.
- AV Block complicating surgery (VSD repair, LVOT repair, AVR).
- Congenital AV Block (CCTGA, AVSD).
- Pacemaker indications generally follow conventional guidelines.

# Transient Complete Heart Block (Septum Primum Atrial Septal Defect)



# Arrhythmias: VT

- 35% of repaired TOF patients have PVC's or NSVT. Approximate 6% risk of sustained VT or late SCD during long term follow up.
- Clinical predictors (imperfect) after repaired TOF: RV dilatation, QRS 180ms or greater, ventricular ectopy on Holter monitor, PES.
- No generally accepted scheme for rhythm surveillance in asymptomatic patients.
- Symptoms should prompt a thorough investigation.

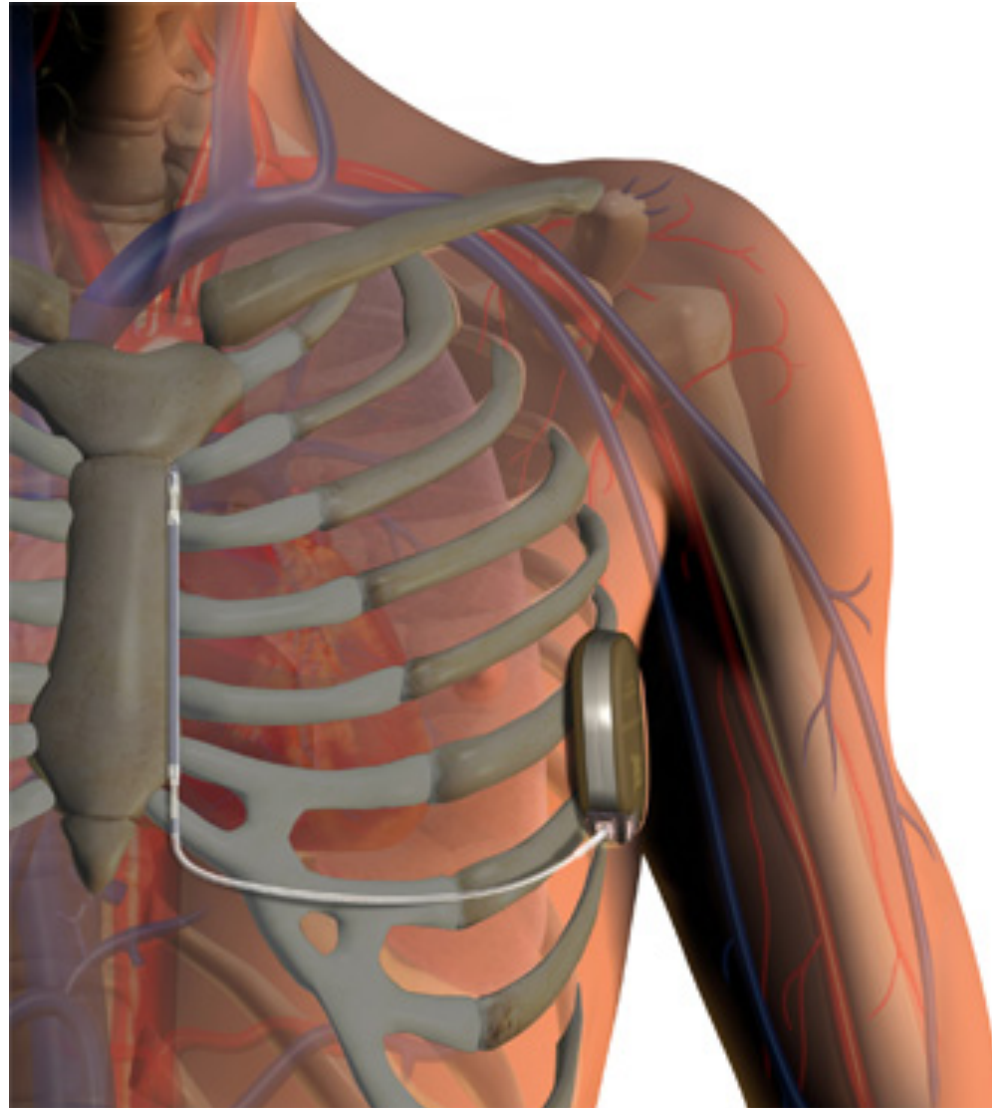
# Arrhythmias: Sustained VT, SCD

- Echo, cath, EPS. If surgery indicated for structural heart indication then consider intra-operative VT mapping and ablation.
- If no surgery indicated then consider catheter ablation of VT (recurrence may be 20% or greater in long term follow up).
- Cardiac arrest, hemodynamically significant VT and sustained VT are Class IIa indications for ICD placement.
- Optimal timing of ICD placement represents a crucial research gap.

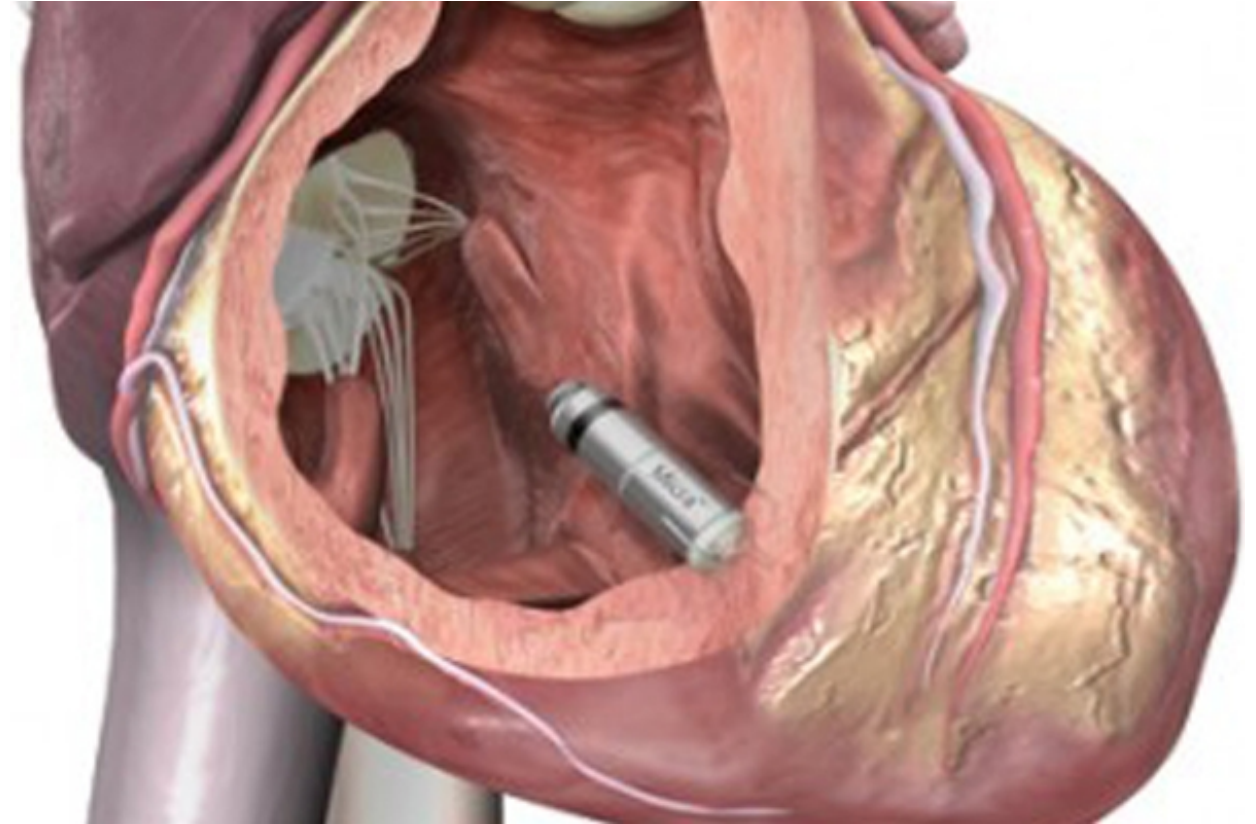
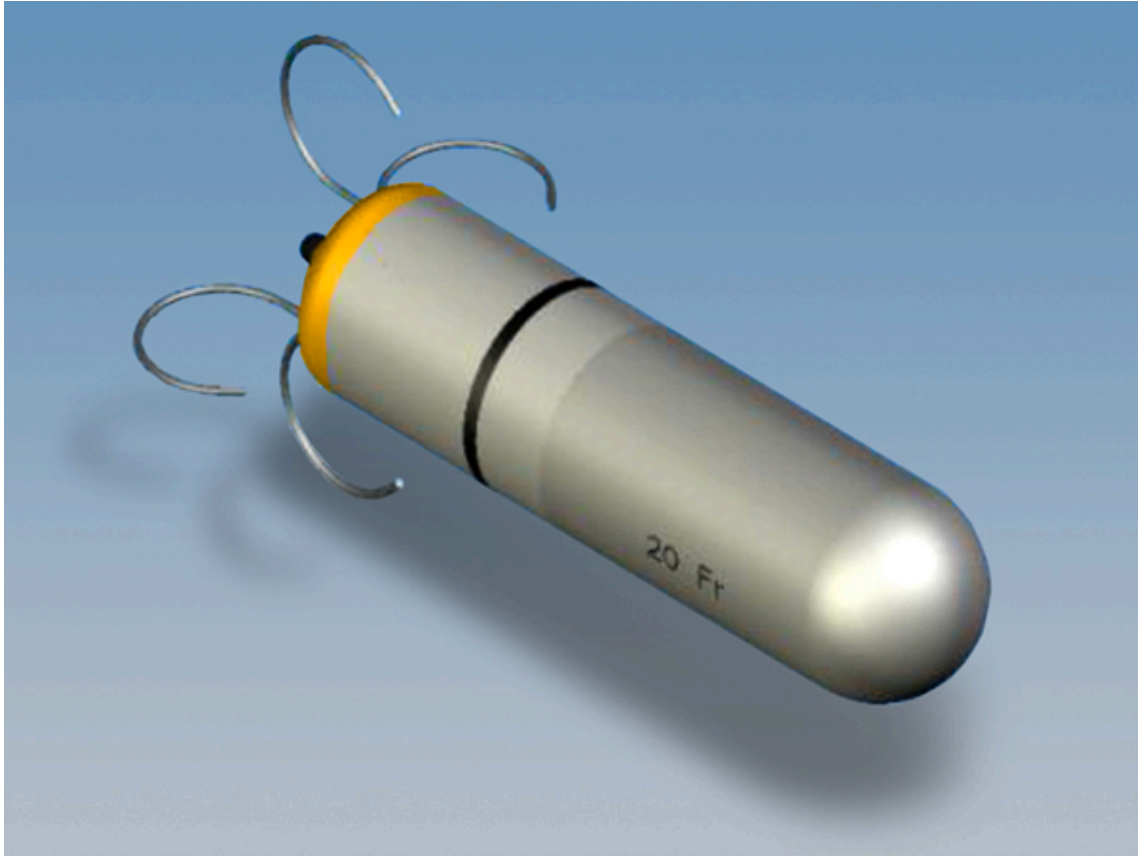
# Arrhythmias: Device Concerns

- Venous return to heart often abnormal.
- In cyanotic patients with R to L shunt there is a risk of lead-related systemic embolism.
- Consider epicardial leads or subcutaneous defibrillator.
- Abdominal or submuscular generator placement an option for cosmetic purposes.

# Subcutaneous ICD



# Leadless Pacemaker





# Adult Congenital Heart Disease

- Growing population.
- Evolving New Specialty.
- Unique concerns.

