Non-Obstructive Congenital Heart Disease

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Objectives

1) Define and describe different forms of non-obstructive congenital heart disease that may impact management of the neonate.

2) Recognize key features of these diagnoses that will assist with both diagnosis and management of potential symptoms.

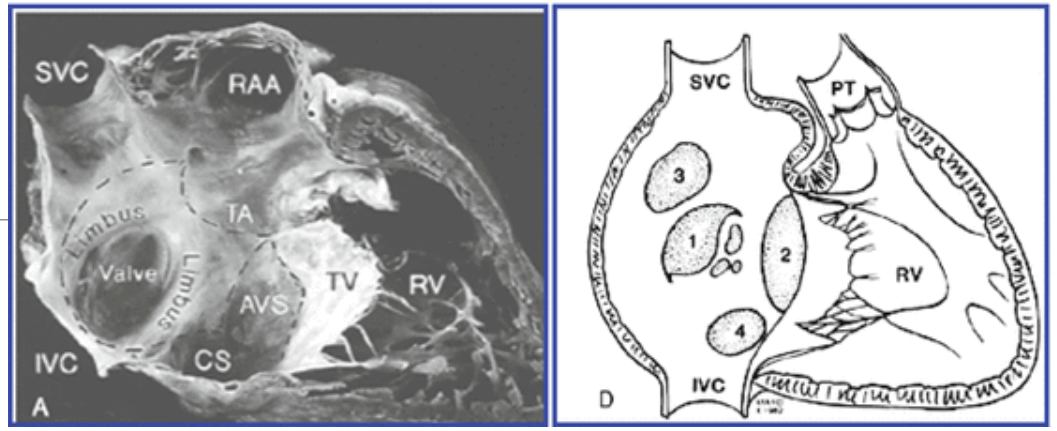
3) Choose appropriate testing to help with both diagnosis and surveillance monitoring of patient's with these diagnoses.

4) Choose management strategies that may assist with mitigating symptoms that can accompany these diagnoses.

5) Identify current indications for consideration of interventional or surgical management for these diagnoses.

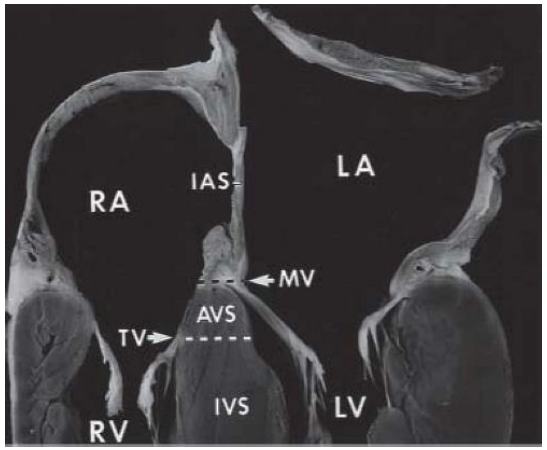
Intra-Cardiac Shunts

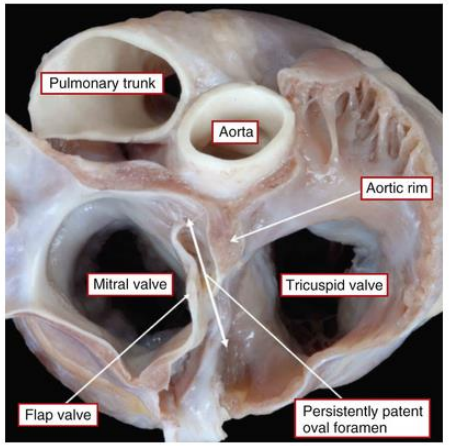
Atrial Septal Defects



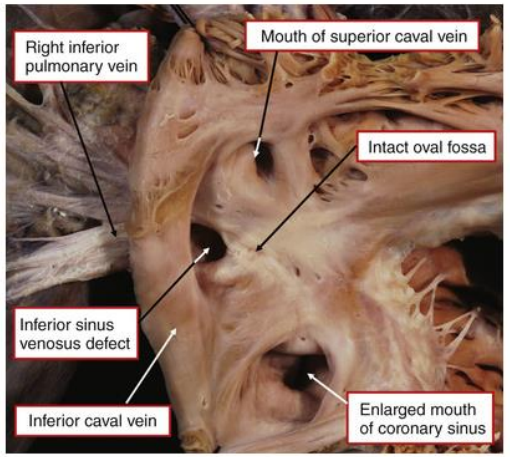
- 10% of all CHD in children (1/1500 live births)
- Multiple types of ASDs:
  - Secundum (75%)
  - Ostium Primum (20%)
  - Sinus venosus (5%)
  - Coronary sinus (<1%)
- Patent foramen ovale – can be found in up to 33% of hearts examined at autopsy
- Risk factors:
  - Genetics - Holt Oram syndrome, Noonan syndrome, Trisomy 21, Klinefelter syndrome, Williams syndromes, Kabuki syndrome, Goldenhar syndrome and Ellis van Creveld syndrome
  - Prenatal/maternal factors– Gestational DM, PKU, influenza infections, medication exposure (retinoids, NSAIDs, anticonvulsants, thalidomide), tobacco and EtOH exposure

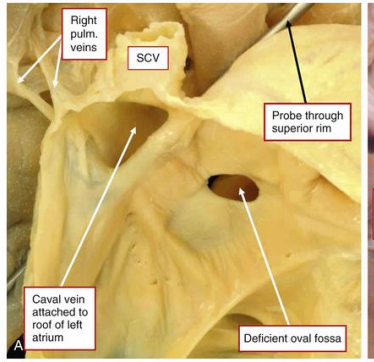
Patent foramen ovale

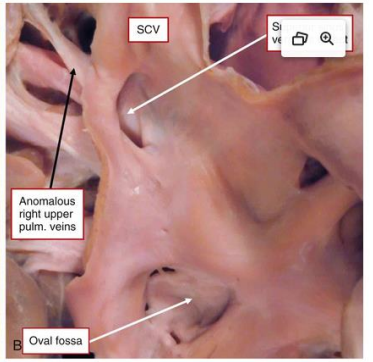


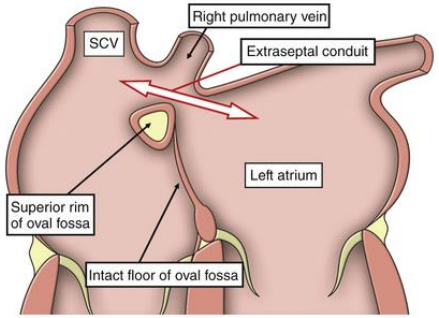


Sinus venosus defect

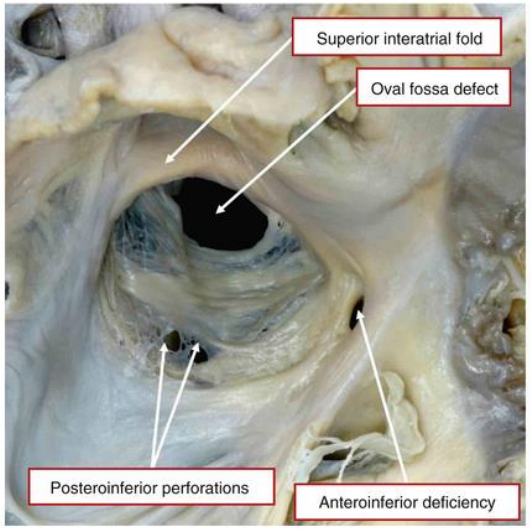


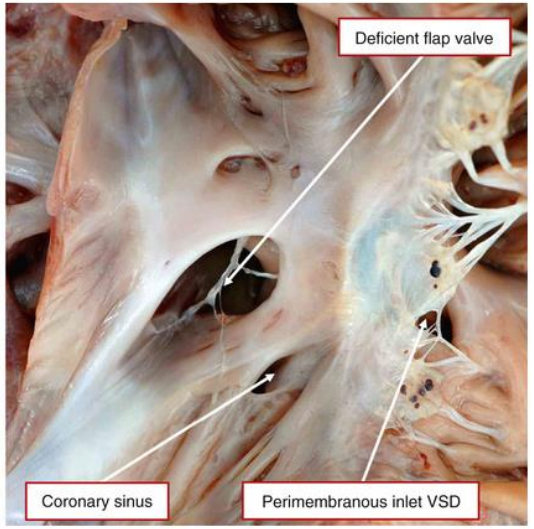




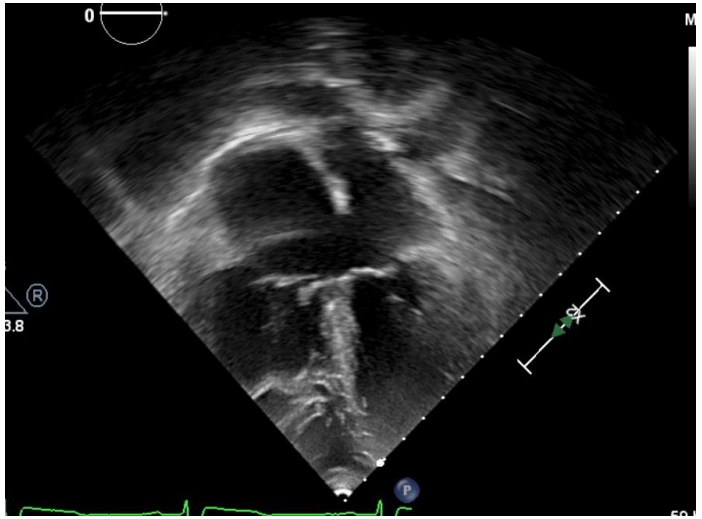


Secundum defect

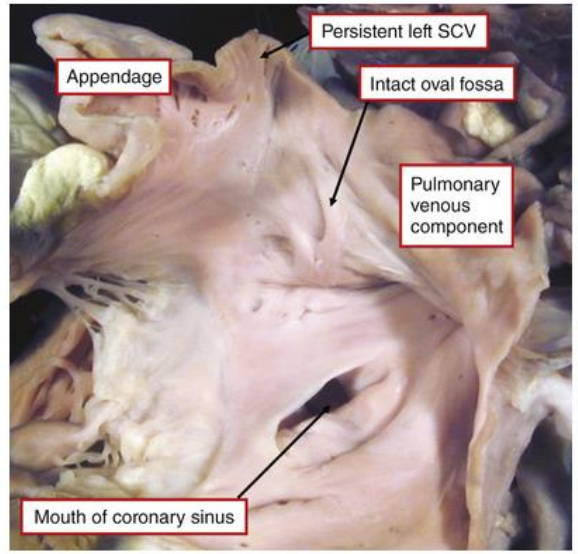




Ostium Primum defect



Coronary sinus defect



ASD Pathophysiology

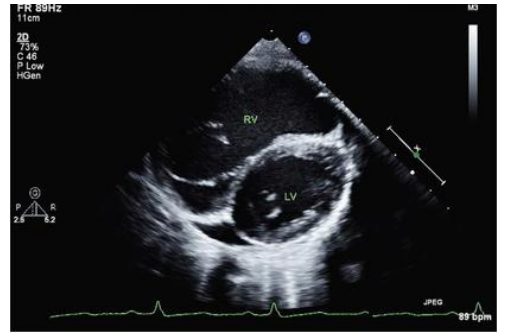
- Physiologic consequences of an ASD depends on the magnitude and duration of the shunt and its interaction with the pulmonary vascular bed
  - Primary factor that impacts this is the ventricular compliance
  - Maximal shunting occurs during diastole
- Hemodynamically significant left to right shunts (moderate to large shunts with estimated Qp:Qs ≥ 1.5:1) lead to right heart dilation secondary to volume overload
- With increased flow to the lungs, the pulmonary arteries, veins and capillaries become dilated and can develop flow related PAH
  - This over time can lead to medial hypertrophy of pulmonary arteries and arterioles resulting in pulmonary vascular obstructive disease
- Common early symptoms include → mild dyspnea on exertion and easy fatigability
- Physical examination → widely split or fixed splitting of S2 (may be less prominent in patients with PHTN), systolic ejection murmur at the LUSB (pulmonary flow murmur) and can also have a mid diastolic murmur at the LLSB secondary to increased flow across the tricuspid valve

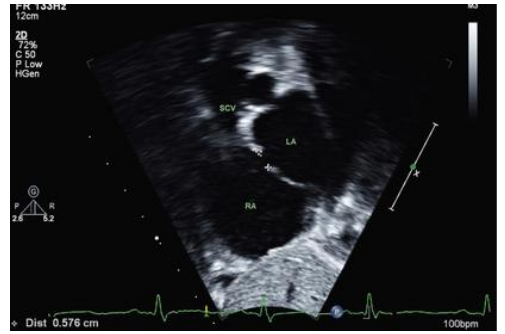
ASD Natural History

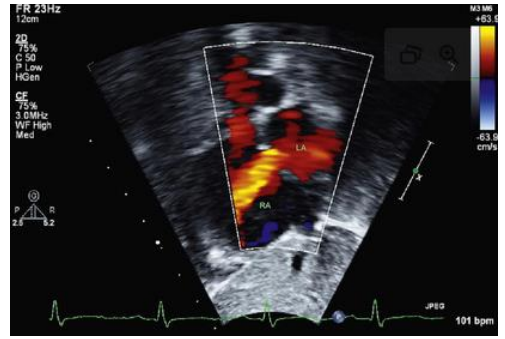
- Secundum defects are the only type of ASDs that can spontaneously close over time.
  - Frequency and timing of closure is inversely related to the diameter of the defect
- Most patients are typically asymptomatic until the 2nd to 3rd decade of life
  - Can be symptomatic sooner if have underlying pulmonary disease (ie CLD of prematurity), certain genetic syndromes or other congenital cardiac defects that impede left atrial emptying
- Mean age of death in unrepaired ASDs was 37.5 years +/- 4.5 years with 75% dying by 50 years and 90% dying by 60 years
- By 40 years of age, 90% of untreated patients have one or more of these symptoms
  - Early symptoms are predominantly dyspnea on exertion, fatigue, palpitations and sustained atrial arrhythmias

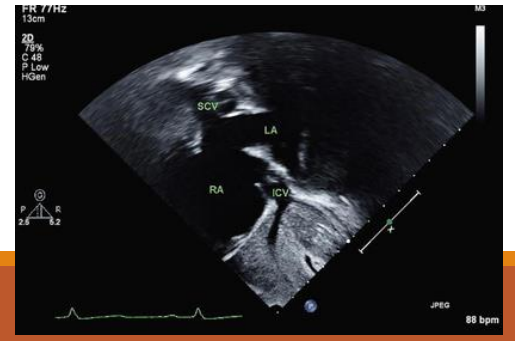
Diagnostic Tools for ASDs

- ECG changes: RAE, abnormalities of P wave axis (sinus venosus defects), RAD, RVH, iRBBB
  - Increased risk of atrial arrhythmia such as atrial flutter and fibrillation (typically > 40 yo)
- Chest x-ray findings: Cardiomegaly with prominent right heart and pulmonary trunk, increased pulmonary vascular markings
- Echocardiogram – hemodynamically significant shunts will have evidence of right heart dilation, helps to evaluate type of ASD
- Cardiac CTA – important for evaluation in patients with sinus venosus type defects due to the high association with PAPVR (90%)
- Cardiac catheterization – can consider if meets criteria for transcatheter device closure or if high risk for PAH for risk stratification (ie CLD, older age at time of diagnosis etc)









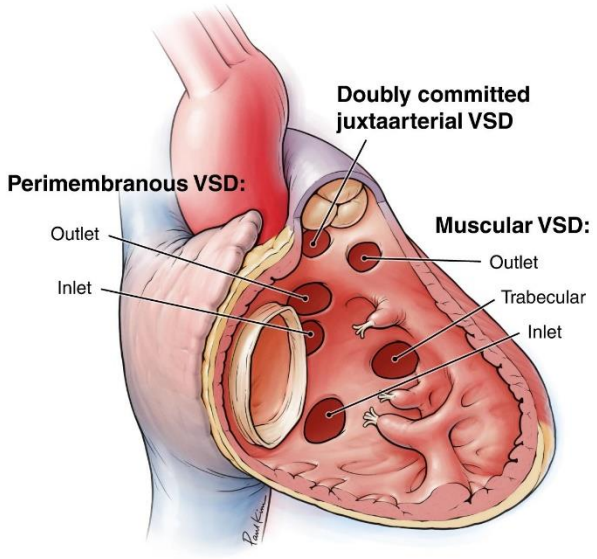
Indications for ASD Closure

- Hemodynamically significant shunts (estimated Qp:Qs ≥ 1.5:1)
- In asymptomatic patients with a moderate to large shunt- recommend elective closure between 2-5 years
  - Earlier repair may be indicated for symptomatic patients or patients with higher risk features (ex CLD, certain genetic syndromes etc) that may not tolerate increased pulmonary blood flow
- Life-long routine follow up is recommended for asymptomatic patients with small shunts
  - At risk for increased L-R shunting over time as LV compliance decreases secondary to CAD and systemic hypertension
- Decision for transcatheter device vs surgical closure
  - Secundum defects are the only defects that are routinely closed percutaneously

Long Term Outcomes Following Procedural Closure

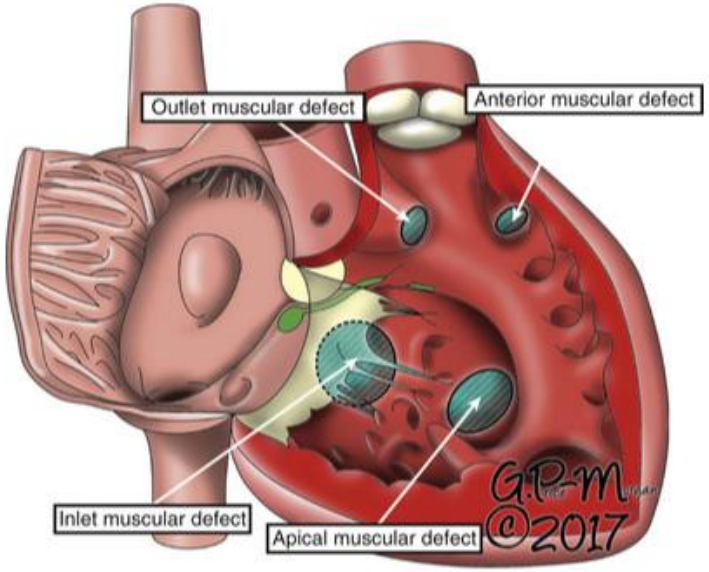
- Outcomes are related to age and presence of PAH
- Typically has excellent prognosis if operated on in childhood
- Short and long term management issues include:
  - Postpericardiotomy syndrome
  - Pulmonary hypertension – may progress in adult patients despit ASD closure
  - Atrial arrhythmias - more common in patients > 40 years (15% at 40, 61% at 60)
    - Superior sinus venosus defects – can be at increased risk for bradyarrhythmias
  - LV diastolic dysfunction - can be affected by long term RV volume overload
  - Mitral regurgitation - Can develop due to RV dilation and septal bowing and may persist after closure
  - Bacterial endocarditis risk - SBE prophylaxis recommended within the first 6 months following closure

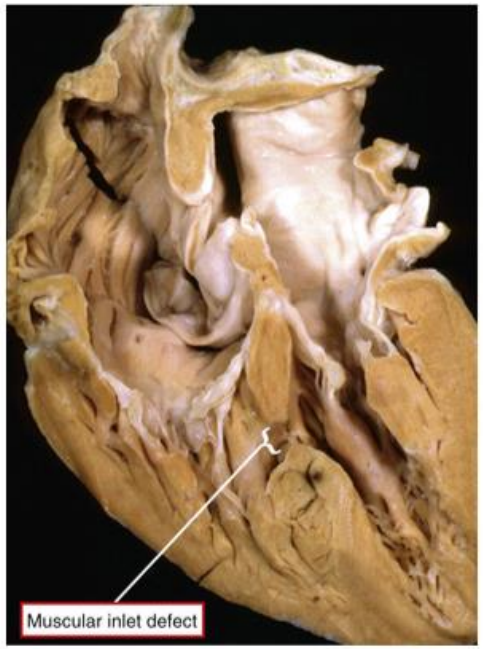
Ventricular Septal Defects

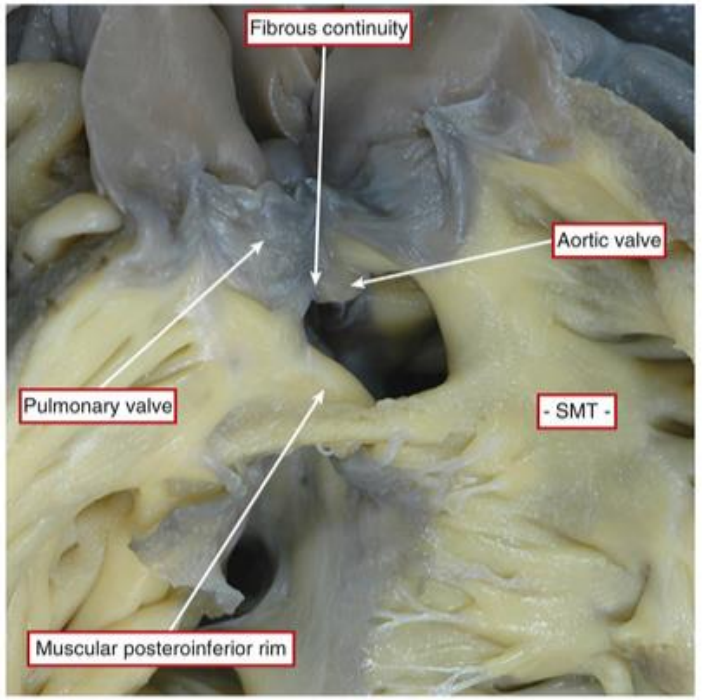


- Most common type of CHD (30-40%) or ~ 2/1000 live births
- Multiple types of VSDs:
  - Muscular
  - Perimembranous
  - Doubly committed and juxtaarterial defects
  - Septal Malalignment defects (ex TOF)
- Can be found in isolation but also commonly found in combination with other defects
  - TOF, DORV, common arterial trunk, TGA etc
- More common in premature infants and infants with low birth weight
- Can also be associated with certain genetic syndromes or environmental exposures
  - Genetics – Trisomies 13, 18 and 21, Holt-Oram syndrome etc
  - Environmental exposures – gestational DM, paternal drug use (MJ and cocaine) and paternal exposure to paint stripping

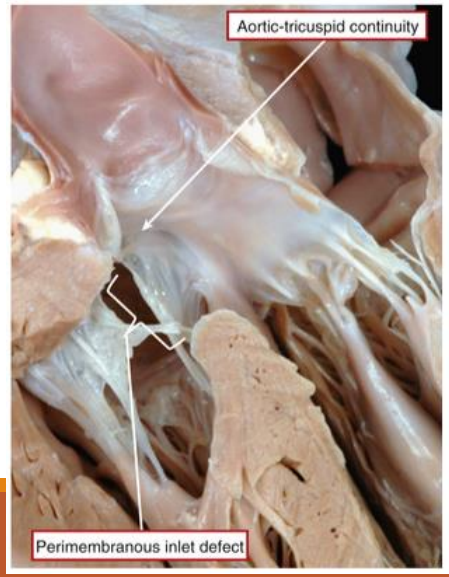
Muscular Defects







Doubly Committed and Juxtaarterial Defects



Perimembranous Defects

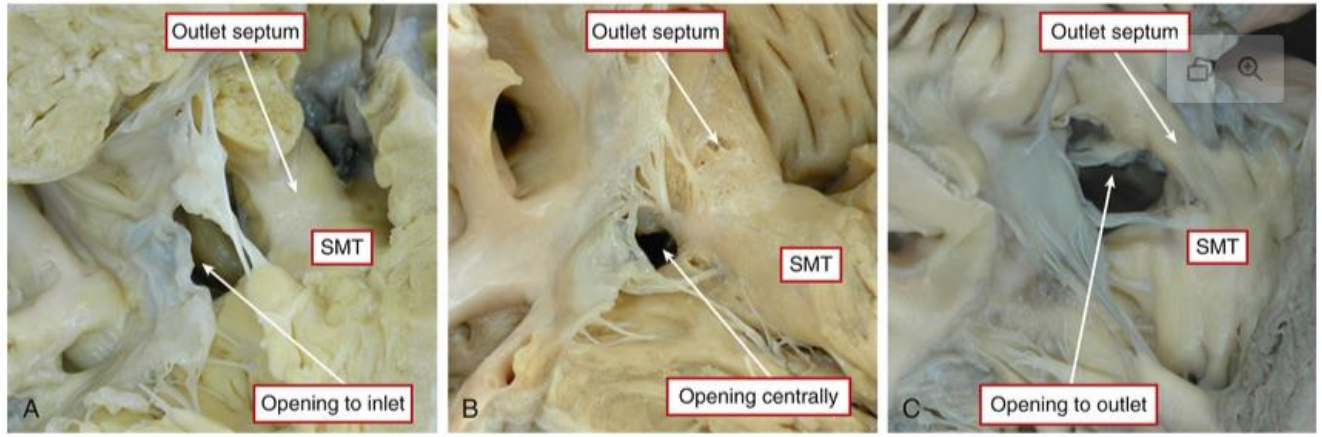


FIG. 32.8 Three defects that all have fibrous continuity in their posteroinferior margins between the leaflets of the aortic and tricuspid valves and hence are perimembranous. As can be seen, however, the defect shown in B opens centrally at the base of the right ventricle, whereas the defect shown in A opens to the right ventricular inlet, with the defect in C opening to the outlet. *SMT,* Septomarginal trabeculation.

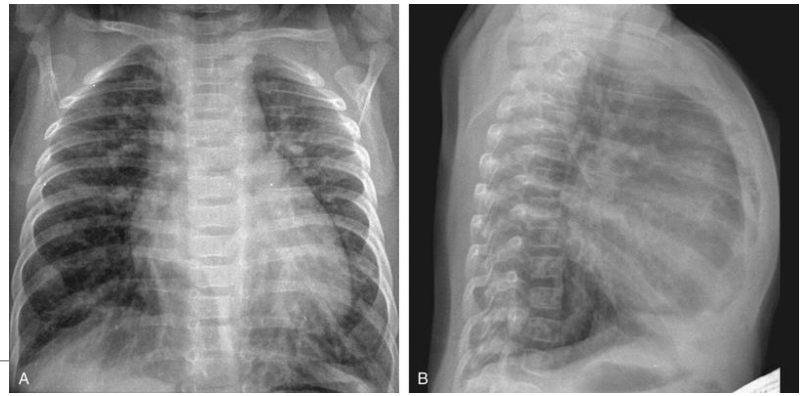
VSD Pathophysiology

- Primary factors include the size of the defect and the pulmonary vascular resistance
  - VSD size is inversely correlated to resistance of flow
  - PVR affects both the magnitude and the directionality of shunting
- Can also be effected by additional cardiac defects
  - Pulmonary stenosis will have a similar effect to elevation in PVR
- Patients with unrestrictive VSDs - the increase in Qp:Qs will lead to increase in left heart size
- The change in left heart volume/size will increase the work load of the LV causing an increase in LV mass as a compensatory mechanism
- Also with unrestricted VSDs there is an elevated in RV pressures leading to RV hypertrophy and can promote subpulmonary muscular obstruction

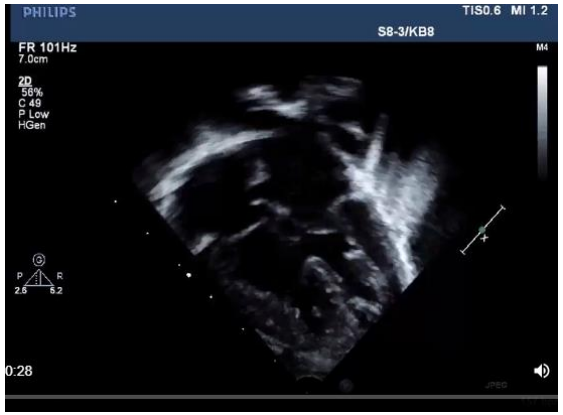
VSD Natural History

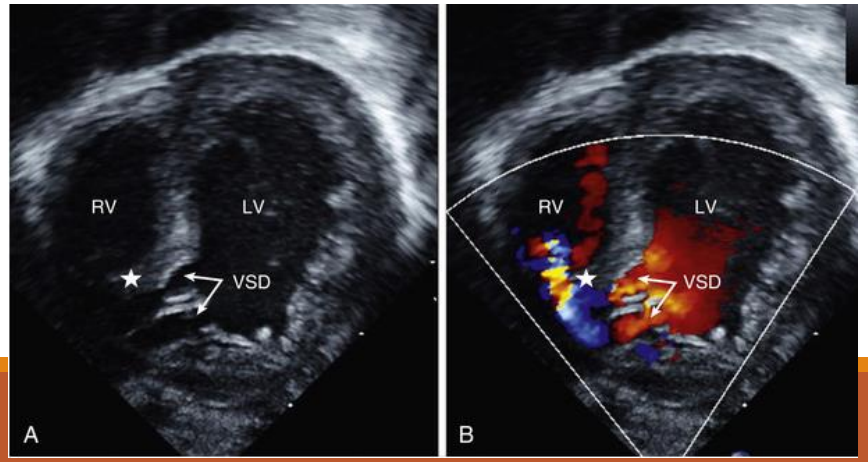
- Small restrictive VSDs
  - Most patients are typically asymptomatic and with muscular defects have a high rate of spontaneous closure within the first 6 years of life (90%).
  - Smaller defect = louder and higher pitched murmur
    - Small muscular defects can have a S1 coincident murmur (not holosystolic)
  - Survival in adults with small restrictive VSDs is comparable to that of the general population
- Hemodynamically significant VSDs
  - Typically present within the first 1-2 months of life as the PVR physiologically drops with neonatal transition
    - Infants can present with symptoms of tachypnea, difficulty with feeding and poor weight gain
  - High risk of pulmonary hypertension in patients with large unrestricted VSDs and this is often irreversible by 2 years of age
    - Death typically occurs < 40 years of age
  - Larger defects do have the possibility of becoming more restrictive but less likely in comparison to the rate of closure in small defects
    - Closure can occur in up to ¼ of this patient population but these defects tend to be small to moderate in size and muscular or perimembranous in location
- There are a subset of patients that my initially be asymptomatic but later on in life they develop significant subpulmonary muscular obstruction (10%) or progressive tricuspid or aortic valve disease (1-5%)

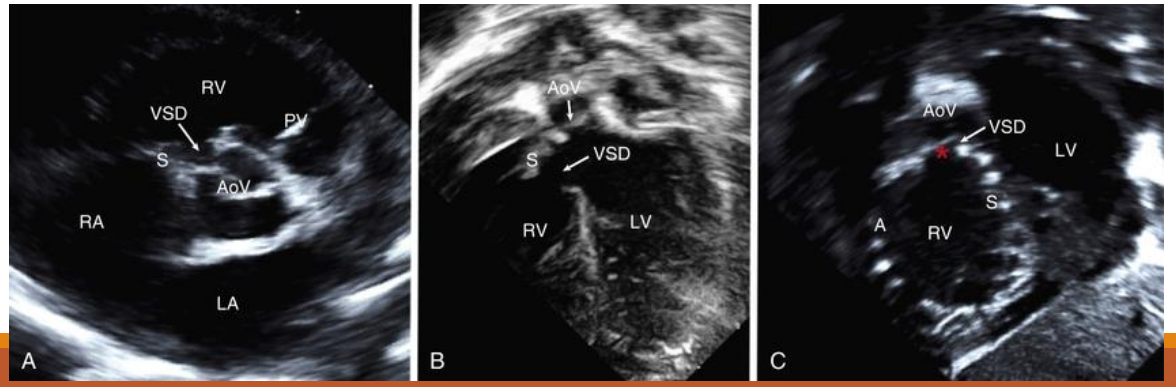
Diagnostic Tools for VSDs



- ECG evaluation: nonspecific findings pre-operatively but commonly have a RBBB following surgical VSD patch closure
- Chest xray findings: As the Qp:Qs increases for large defects within the first couple months of life the cardiac silhouette will increase and there will also be increased pulmonary vascularity
- Echocardiogram = gold standard of diagnosis
- Cardiac catheterization is typically only utilized for diagnostic evaluation if there is concern for severe PHTN







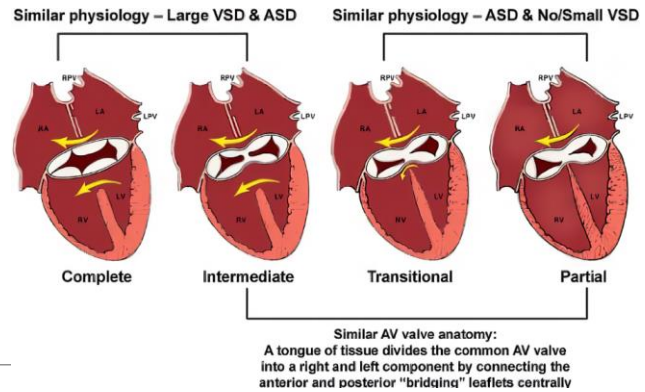
Surgical VSD Closure

- Indications:
  - Recommend intervention in the 1st year of life for patients with hemodynamically significant VSDs and congestive heart failure symptoms refractory to medical management
  - Aortic valve regurgitation secondary to valve/leaflet prolapse through the defect
  - Severe obstruction of the RV outflow tract (double chambered RV)
- Surgical approach:
  - Surgical patch closure
  - PA banding
    - High risk features = Small size, complex associated cardiac abnormalities, genetic syndromes, extracardiac co-morbities (ex pulmonary disease), multiple small muscular VSDs (“swiss cheese septum”)
    - Can lead to distortion of pulmonary trunk, distal migration of the band towards the branch PAs, pulmonary insufficiency, RV hypertrophy, development of supravalvular pulmonary obstruction, increased risk of subaortic obstruction

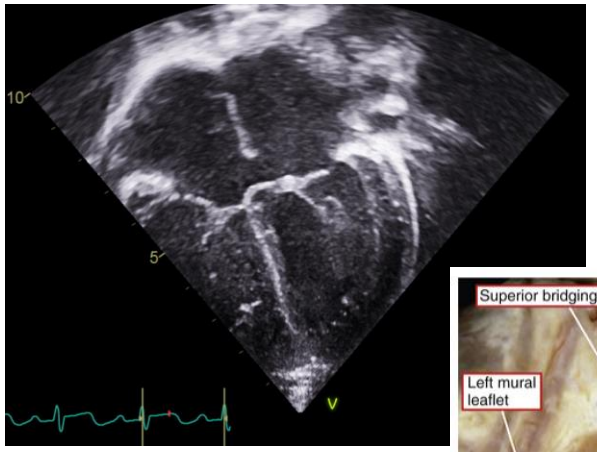
Long Term Outcomes Following VSD Surgery

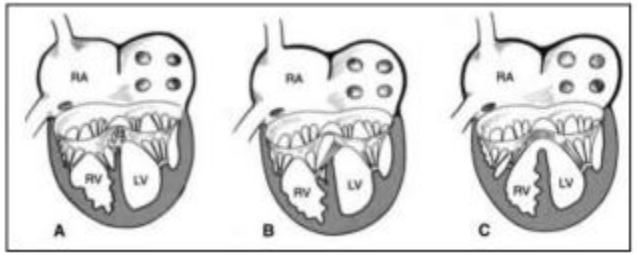
- Generally good long term prognosis in patients repaired early in life
- STS Congenital Heart Surgery Database data from Jan 2013-2016 (7322 operations):
  - Isolated VSD – mean operative mortality 0.6%, mechanical circulatory support 0.2%, permanent pacemaker 0.9%
  - Risk of post-operative complications higher in patients with multiple defects – operative mortality 2.6%, mechanical circulatory support 2.6%, permanent pacemaker 3.4%
- Long term risks:
  - Diminished exercise capacity
    - Thought to be secondary to abnormal RV structure/function, RBBB and its possible correlation to chronotropic impairment and pulmonary vascular disease
  - Rhythm related abnormalities (5%)
    - 1% risk of permanent complete heart block
    - RBBB
    - Atrial fibrillation
  - May have residual shunting
  - Progressive RV outflow tract obstruction
  - Can have progressive aortic valve disease or the developmental of a sinus of Valsalva aneurysm

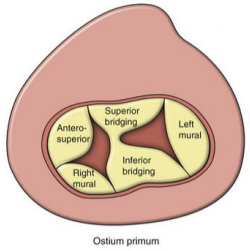
Atrioventricular Septal Defects

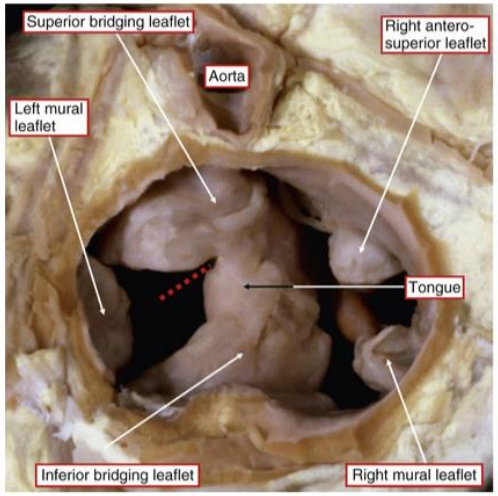


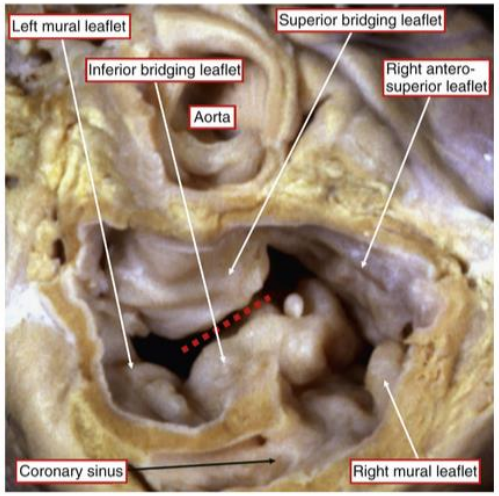
- Abnormal development of the atrioventricular junction – including the AV component of the membranous septum and the surrounding myocardial separating structures
- Different types:
  - Partial AVSD
  - Intermediate and Transitional AVSDs
  - Complete AVSD
- Estimated to occur in 0.19-0.35/1000 live births and 3-5% of CHD
- Strong association of atrioventricular septal defects with Trisomy 21 (~30-45%)
- Ellis van Creveld syndrome – associated with common atrium and a common AV valve with no ventricular component

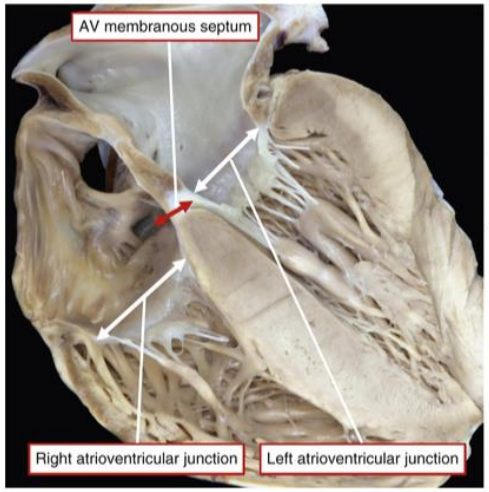


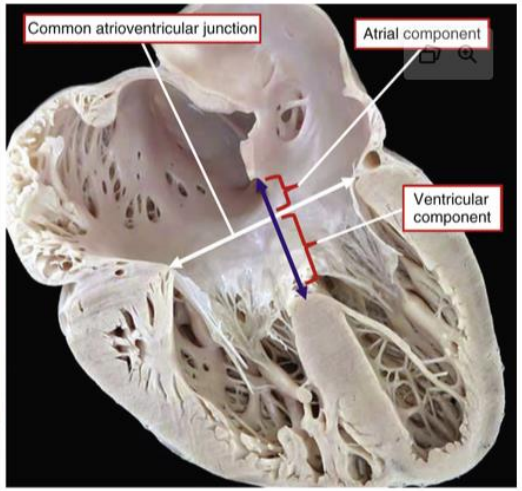

RA
RV
LV
A
B
C

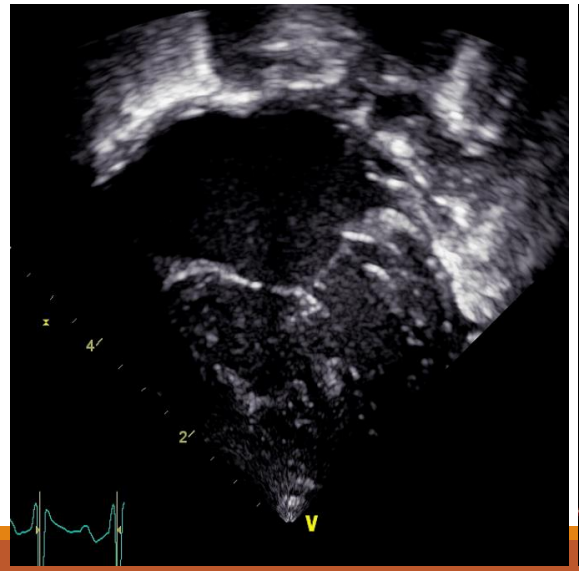

Superior bridging
Antero-superior
Left mural
Inferior bridging
Right mural
Ostium primum

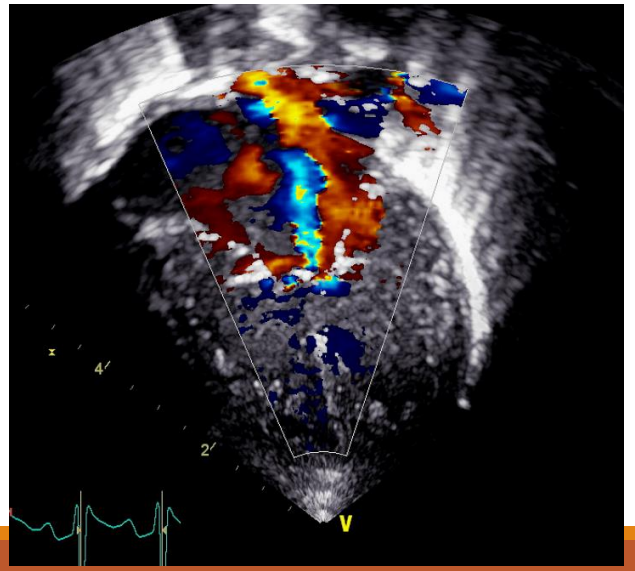

Superior bridging leaflet
Right antero-superior leaflet
Aorta
Left mural leaflet
Tongue
Inferior bridging leaflet
Right mural leaflet

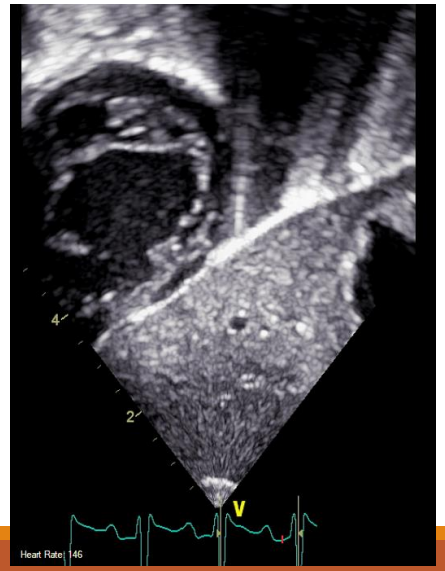

Left mural leaflet
Superior bridging leaflet
Inferior bridging leaflet
Right antero-superior leaflet
Aorta
Coronary sinus
Right mural leaflet

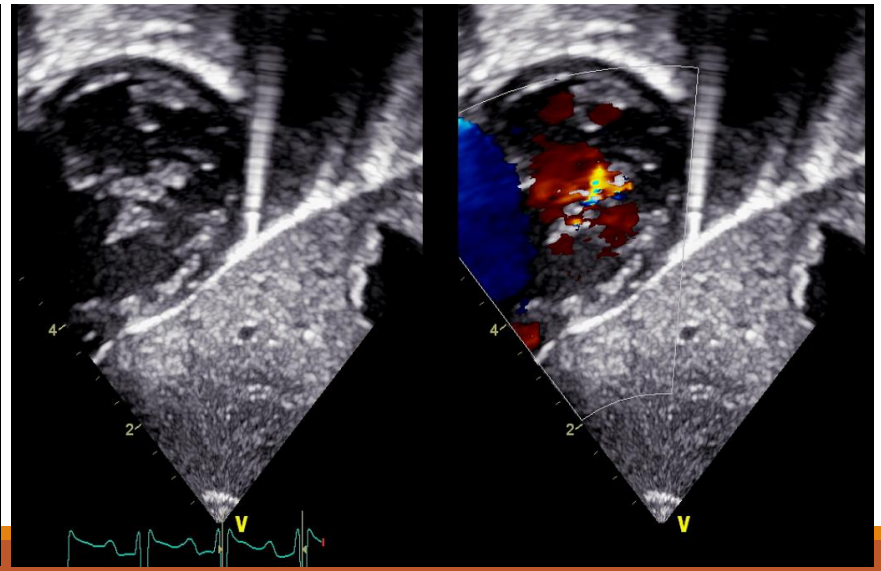

AV membranous septum
Right atrioventricular junction
Left atrioventricular junction


Common atrioventricular junction
Atrial component
Ventricular component






Heart Rate 146



AVSD Pathophysiology

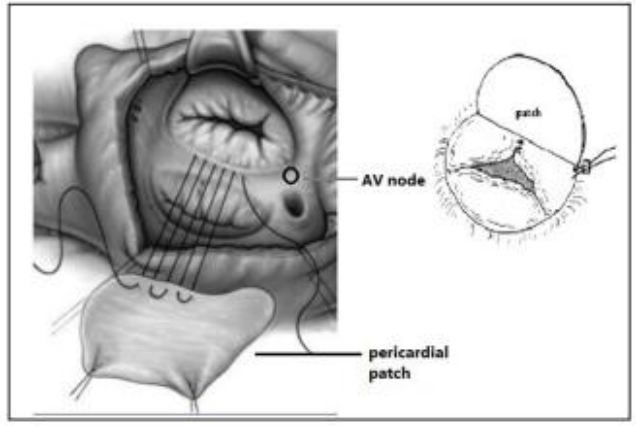
- Clinical presentation depends on the following factors:
  - Size of ventricular and atrial septal shunts
    - Depends on the relationship of the bridging leaflets and the connecting tongue of tissue to the leading edge of the atrial septum and the crest of the scooped out ventricular septum
  - Functionality of the AV valve
  - Associated cardiac findings
    - Tetralogy of Fallot, subaortic obstruction, aortic arch hypoplasia, ventricular dominance (unbalanced type defects), atrial isomerism, heterotaxy syndrome etc

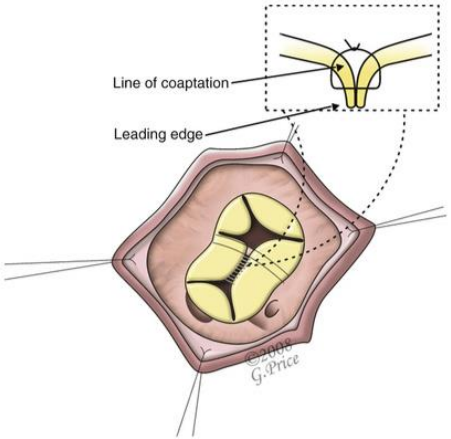
Diagnostic Tools for AVSDs

- ECG – Northwest axis deviation
- Chest Xray – cardiomegaly secondary to volume overload secondary to intracardiac shunts and/or significant AV valve regurgitation, prominent pulmonary trunk, increased pulmonary vascular markings
- Echocardiogram
- Cardiac catheterization – typically only used if concern for significant pulmonary hypertension

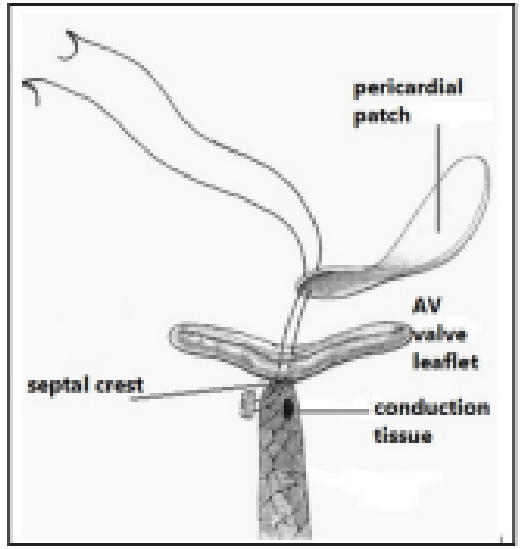
Surgical AVSD Repair

- Indications:
  - Hemodynamically significant intracardiac shunts (both atrial and ventricular)
  - Hemodynamically significant AV valvular disease
  - Typically involves a combination of septal defect closure and AV valve repair
- Different surgical techniques include single patch, modified single patch (Austrailian), two patch techniques and surgical left AV valve repair

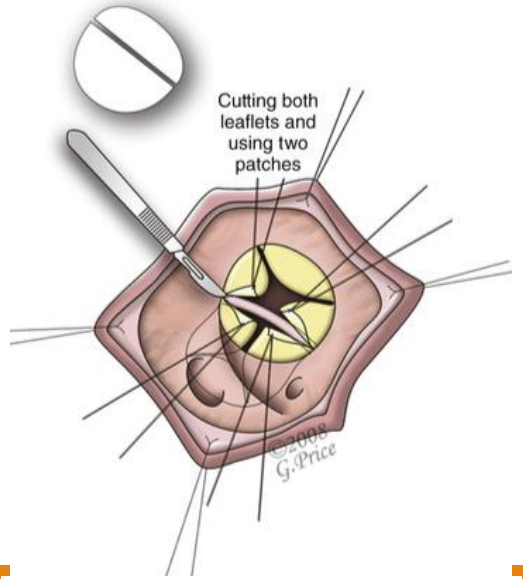




Single Patch technique



Modified Single Patch Technique



Double Patch Technique

Long Term Outcomes Following Surgery

- Perioperative mortality and the risk for heart block is relatively low in partial AV septal repairs
  - Data from the Pediatric Heart Network (PHN) → overall perioperative mortality ~3% and 20 year survival rate of 95%
- Residual left AV valve disease has significant impacts on both short and long term outcomes following intervention
  - Most common indication for reoperation is left AV valve regurgitation (~15-20%)
- Monitor for evidence of progressive LV outflow tract obstruction (~5%)
  - Multiple etiologies → discrete membrane, diffuse tunnel like stenosis, accessory AV valve tissue, aneurysmal AV valve tissue, asymmetric ventricular hypertrophy
  - More common in partial AV canal defects

	Total Number of Pts (N)	Early Mortality (%)	Late Mortality (%)	3rd Degree Block (%)	Late MV Reop (%)
CSP	350	4.8	4	9.7	2.3
MSP	200	2	0	2	0.5
DP	889	3.5	2.1	7.2	2

Medical Management for Intra-cardiac Shunts

- Patients with significant signs of pulmonary overcirculation:
  - Nutritional optimization and consideration of enteral tube placement
  - Optimizing hematocrit to optimize oxygen carrying capacity
  - Diuretics to help with volume loading effect on the heart
  - May benefit from maneuvers that increase PVR:
    - Minimization FiO2
    - Ventilatory strategies – increase PEEP, targeting slightly higher PaCO2, targeting slightly lower pH
  - If have significant concern for low effective systemic cardiac output can consider inotropic support with agents such as epinephrine
- Patients with significant AV valve disease:
  - Consider afterload reduction with agents such as ACEi

Mixing Lesions

Truncus Arteriosus

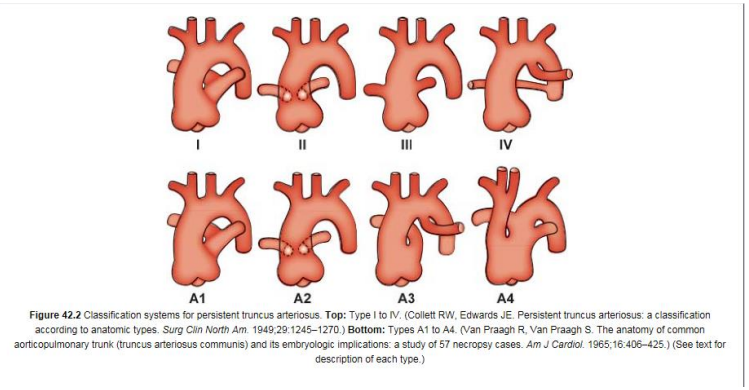
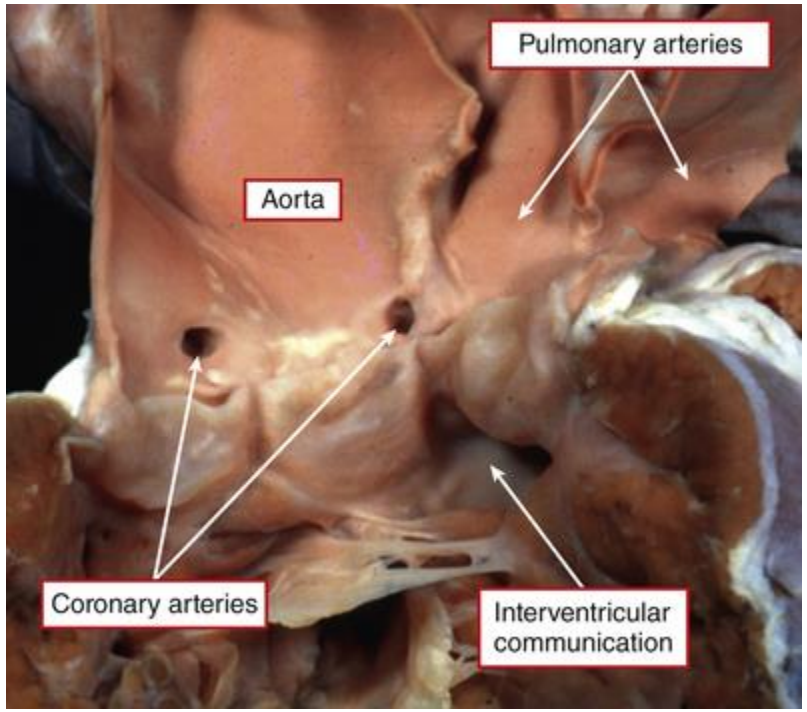
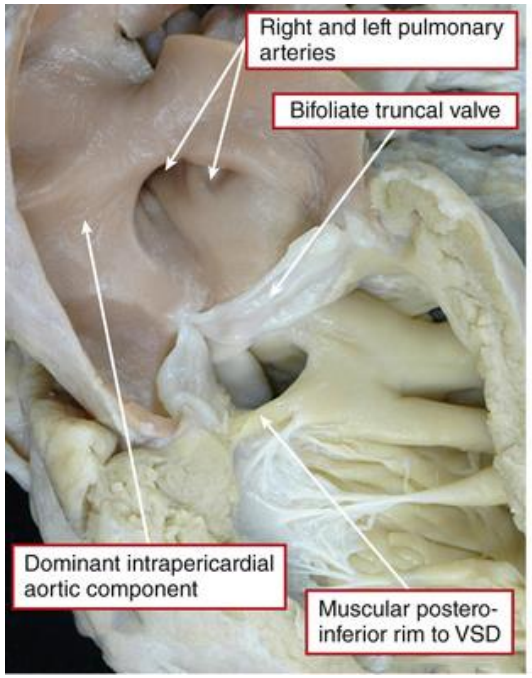
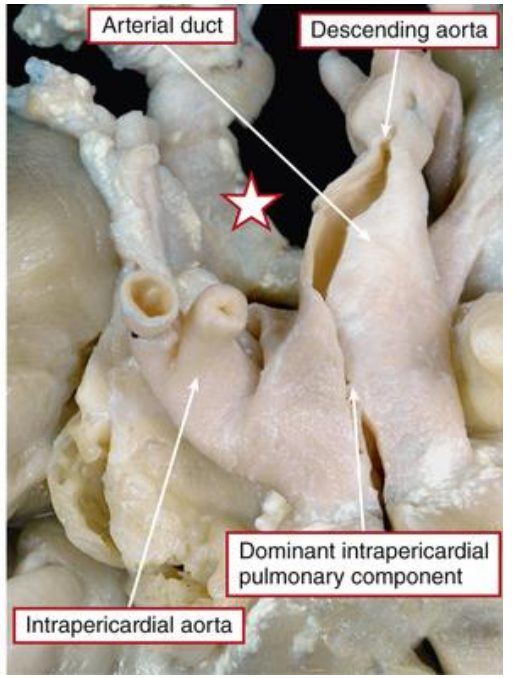


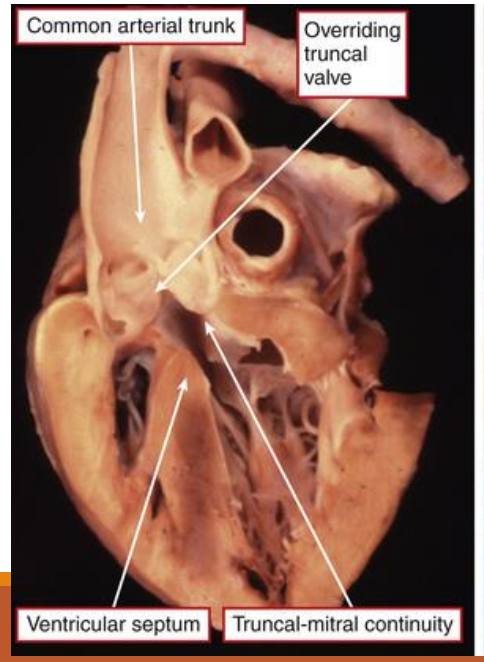
Figure 42.2 Classification systems for persistent truncus arteriosus. **Top:** Type I to IV. (Collett RW, Edwards JE. Persistent truncus arteriosus: a classification according to anatomic types. *Surg Clin North Am.* 1949;29:1245–1270.) **Bottom:** Types A1 to A4. (Van Praagh R, Van Praagh S. The anatomy of common aorticopulmonary trunk (truncus arteriosus communis) and its embryologic implications: a study of 57 necropsy cases. *Am J Cardiol.* 1965;16:406–425.) (See text for description of each type.)

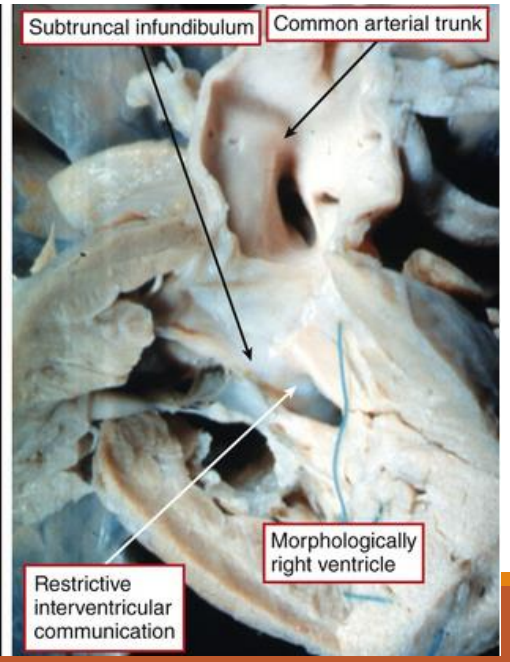
- Common arterial trunk that gives rise to systemic, pulmonary and coronary circulations
- Occurs in 6-10/100,000 live births
- Accounts for 0.7% of all CHD and 4% of critical congenital heart disease
- Truncal valve dysplasia can account for significant morbidity
  - ~50% have truncal insufficiency with 50% of these patients having moderate to severe insufficiency
- Can be commonly associated with other forms of CHD
  - Coronary anomalies, aortic arch abnormalities, pulmonary artery stenosis
- Commonly have associated extracardiac or genetic abnormalities
  - 20-30% of patients have Digeorge syndrome (22q11)
  - Extracardiac anomalies include skeletal deformities, hydroureter, bowel rotation and multiple complex anomalies (21-30%)
  - Maternal DM and AMA have been implicated as a risk factor











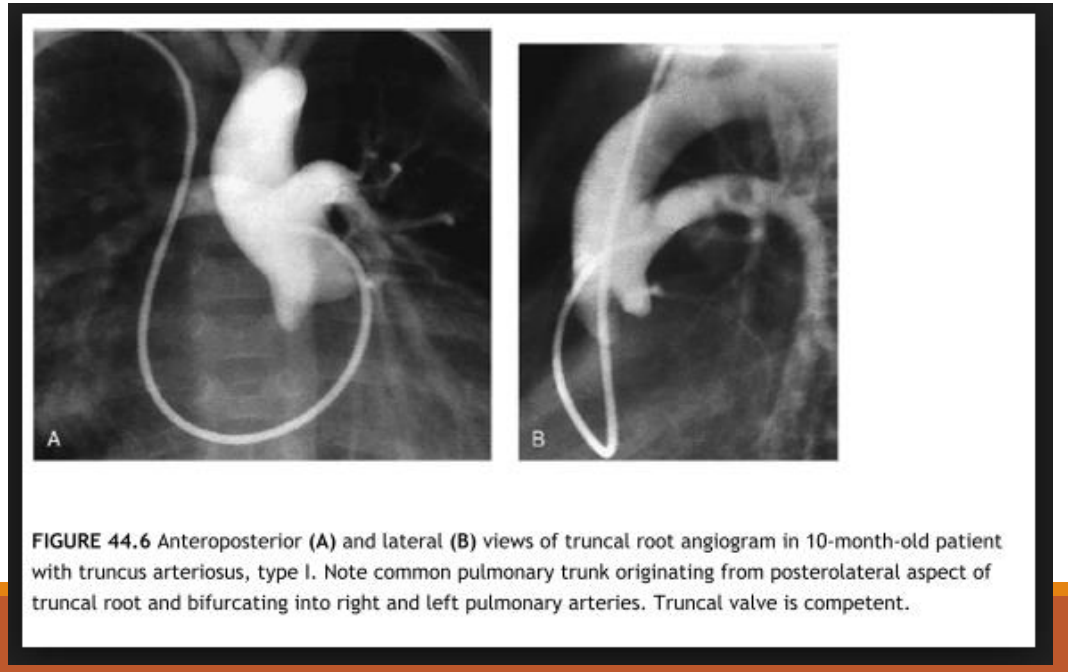


FIGURE 44.6 Anteroposterior **(A)** and lateral **(B)** views of truncal root angiogram in 10-month-old patient with truncus arteriosus, type I. Note common pulmonary trunk originating from posterolateral aspect of truncal root and bifurcating into right and left pulmonary arteries. Truncal valve is competent.

Pathophysiology of Truncus Arteriosus

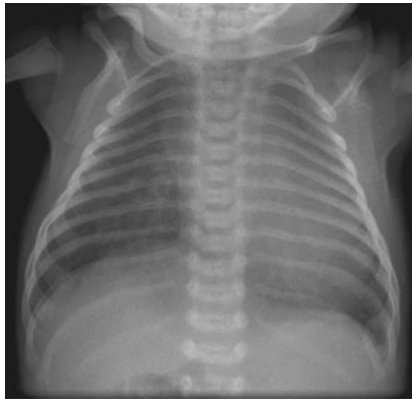
- In the present era is commonly diagnosed prenatally
  - However if is missed antenatally, patients with typically present early in infancy
- As PVR decreases in the neonatal period , you start to see signs of pulmonary overcirculation
  - Tachypnea, tachycardia, excessive sweating, poor feeding
  - While they may have signs of mild cyanosis, this is not a common initial feature unless there is significant pulmonary obstruction or pulmonary hypertension
- As the Qp:Qs starts to increase they are at significant risk for systemic and coronary steal
- If they have significant truncal insufficiency, this can increase their risk of heart failure, low cardiac output and coronary insufficiency.
- Eventually if left unrepaired for too long they will develop hypertensive pulmonary vascular disease (plexogenic pulmonary arteriopathy) due to chronic exposure to systemic arterial pressures
- They may also develop pulmonary venous hypertension if exposed to chronic truncal insufficiency (secondary to left atrial hypertension)

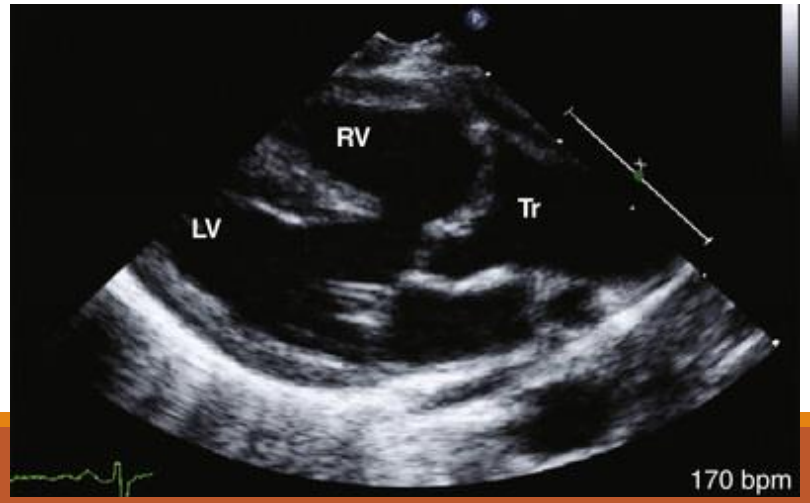
Natural History of Truncus Arteriosus

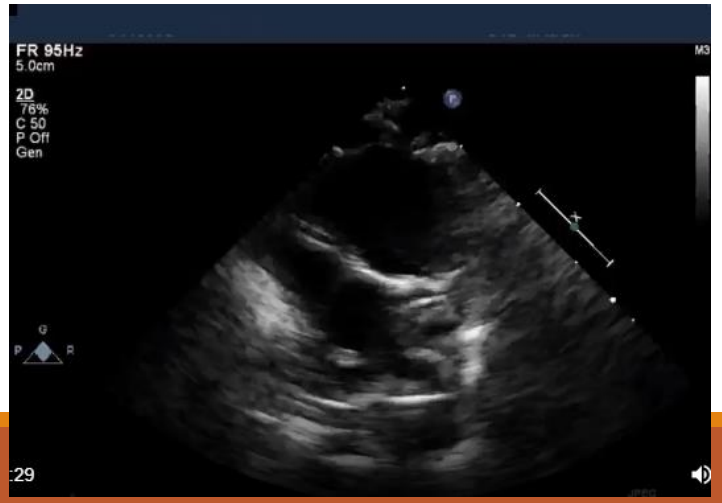
- Without treatment, most patients die within the first 6-12 months
- Survival past infancy was 15% without treatment
  - Most commonly caused by heart failure
  - Past 1 year of age, death is most likely to be due to complications from hypertensive pulmonary vascular disease and infective endocarditis
- Repair between 6-12 months is associated with 2x the mortality risk than for those repaired between 6 weeks to 6 months

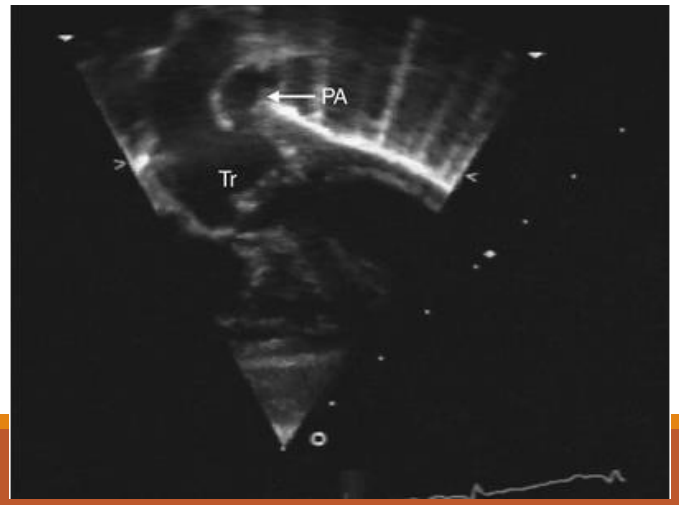
Diagnostic tools for Truncus Arteriosus

- ECG – generally nonspecific but may see evidence of ST segment changes if there is coronary insufficiency
- Chest Xray – typically shows cardiomegaly with increased pulmonary vascular markings (unless there is unilateral or bilateral pulmonary obstruction)
- Echocardiogram
- Cardiac CTA – helpful tool for surgical planning to define pulmonary arterial anatomy and aortic arch anatomy (if interrupted)

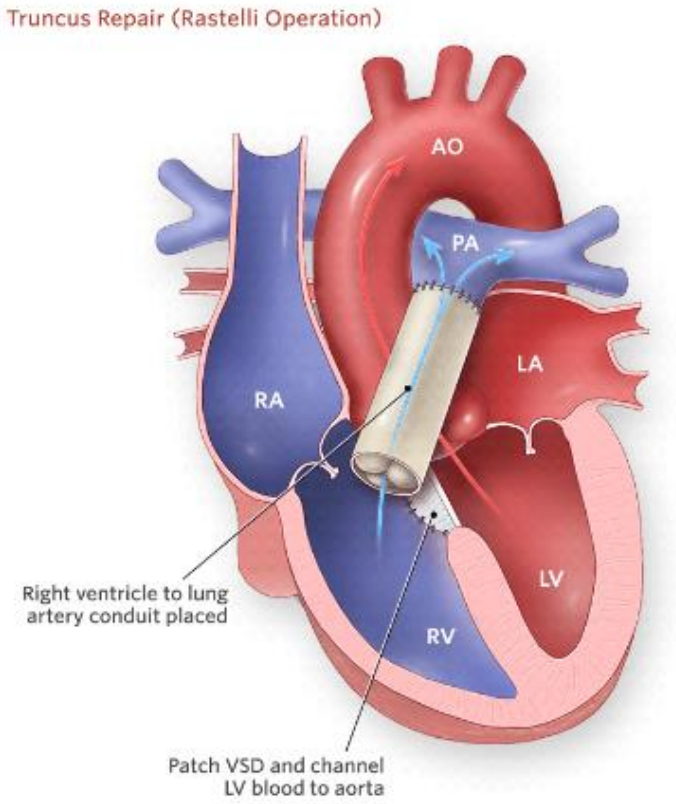




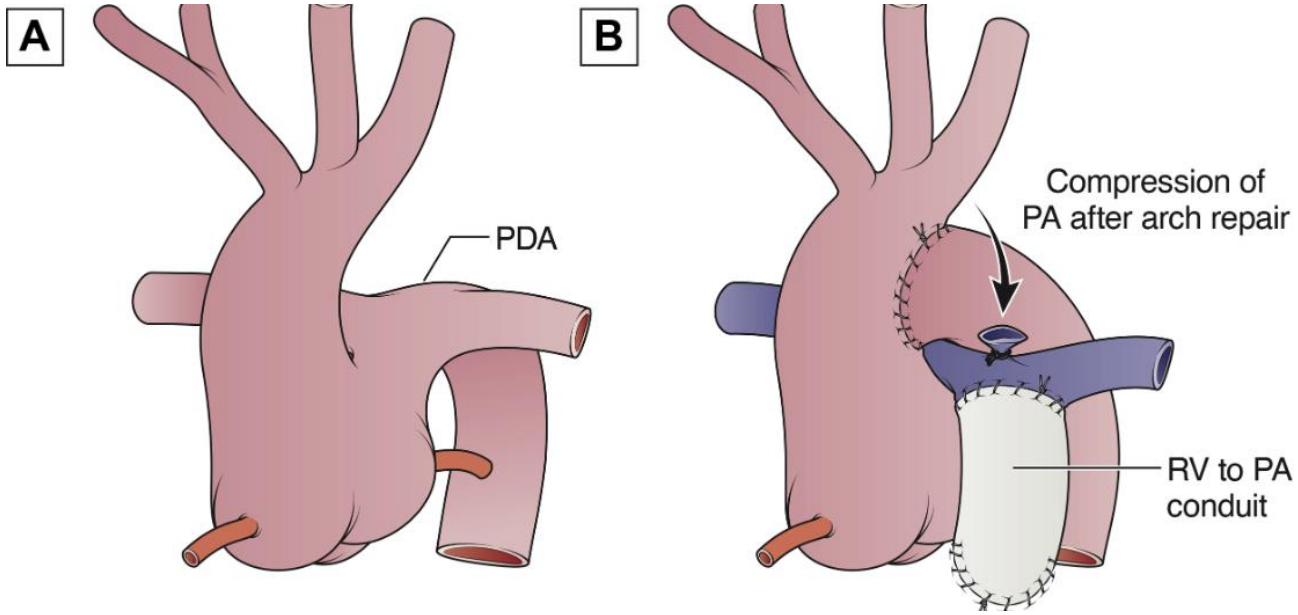




Surgical Truncus Arteriosus Repair



Rastelli operation – VSD closure and RV-PA conduit placement



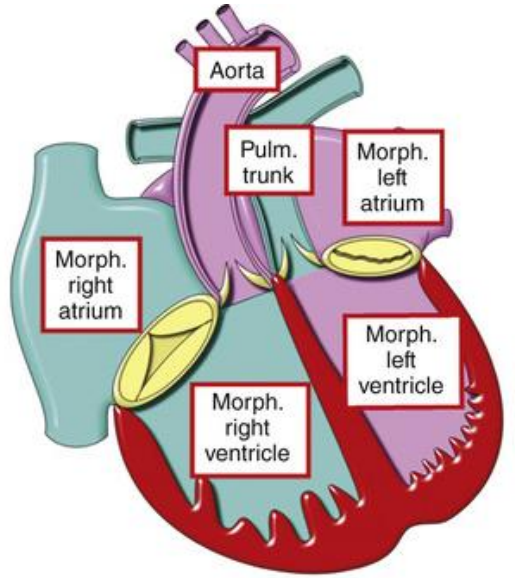
Rastelli operation with aortic arch reconstruction

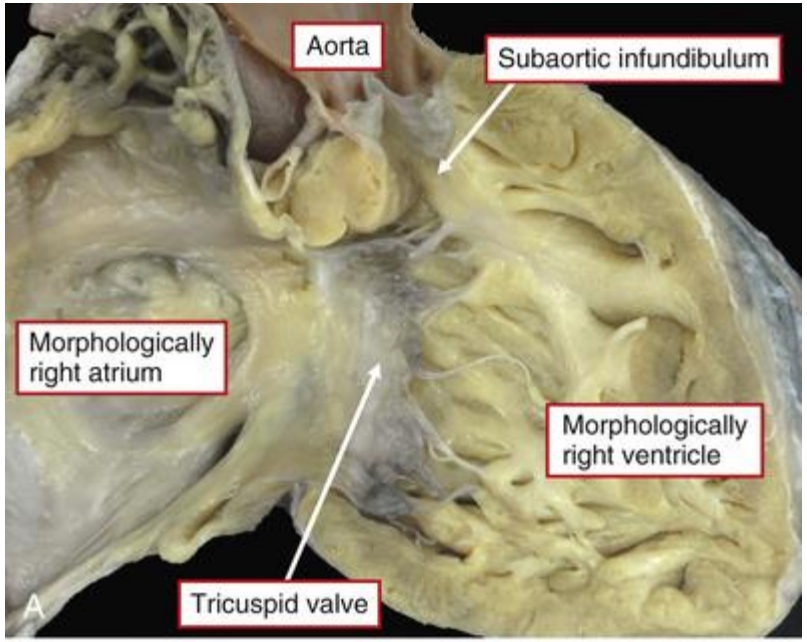
Long Term Prognosis for Truncus Arteriosus

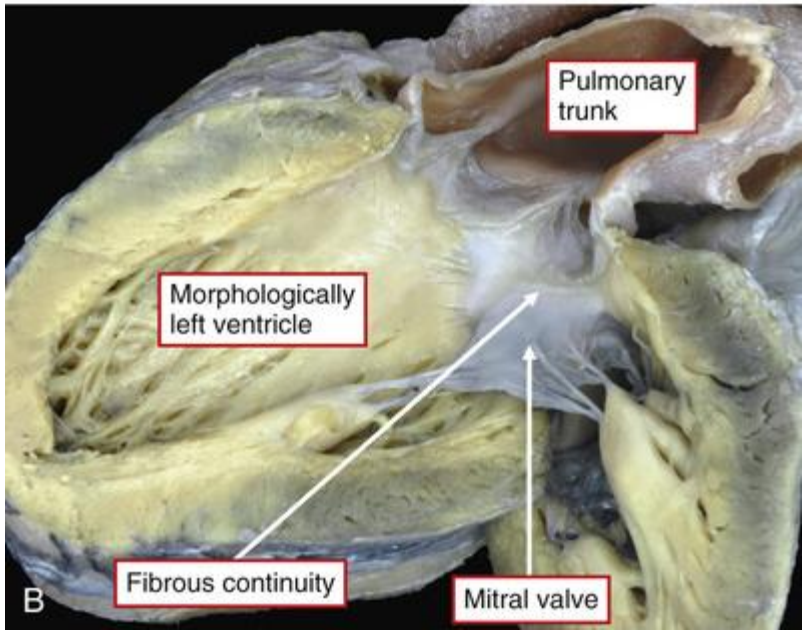
- Operative mortality is 9.4%
  - Risk factors: significant truncal valve disease, interrupted aortic arch, need for post-operative ECMO, low weight, coronary abnormalities
- Long term prognosis following repair is largely determined by the degree of truncal valve insufficiency and the need for conduit replacement
  - Mean time to conduit replacement is 5.5 years
  - 90% survival at 5 years, 85% at 10 years, 83% at 15 years

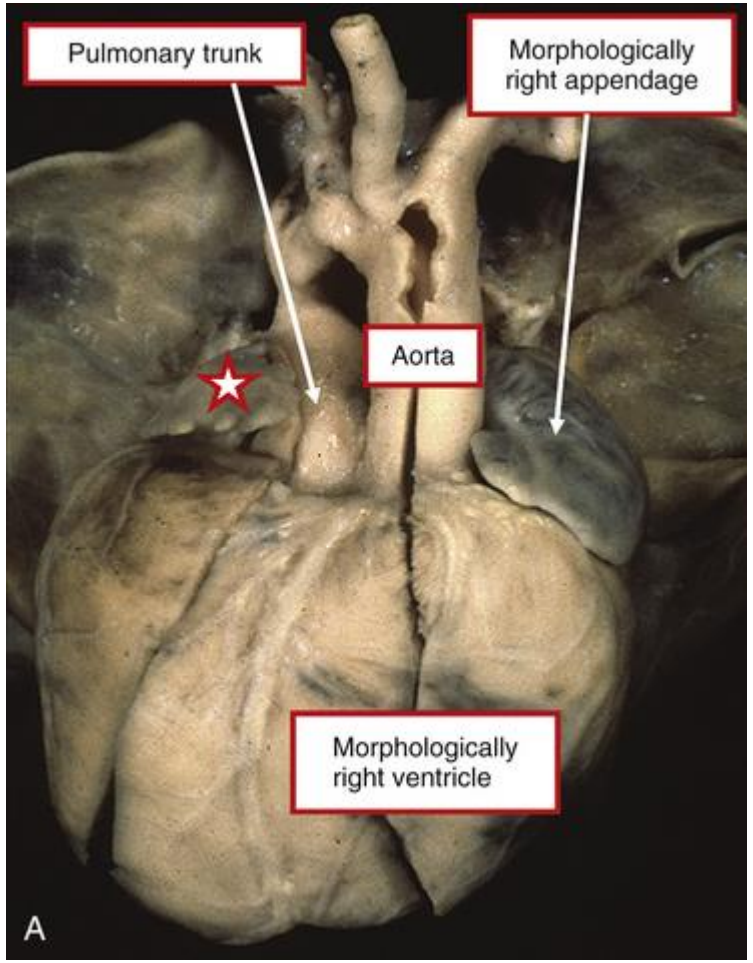
Transposition of the Great Arteries

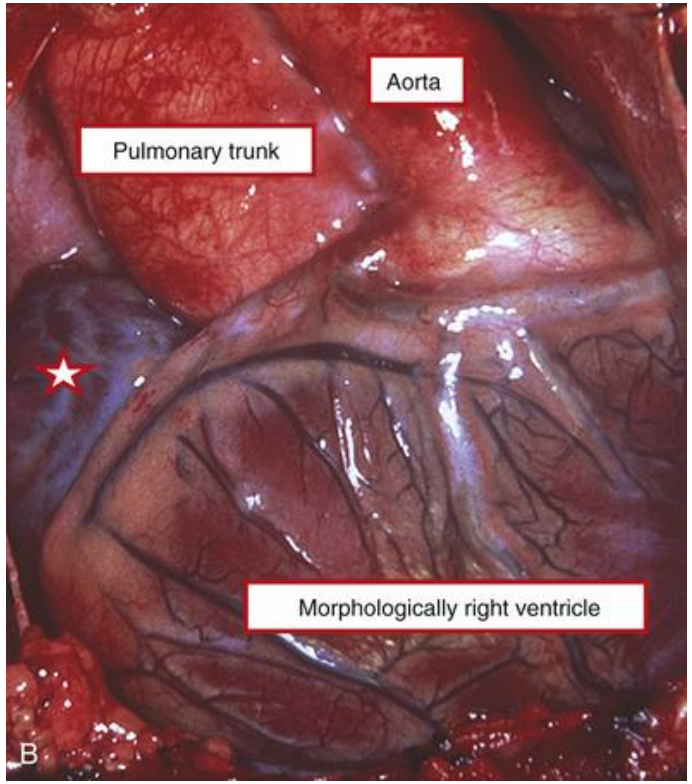
- D-Transposition = atrioventricular concordance with ventriculoarterial discordance
  - Most common form of cyanotic CHD in the neonatal period and accounts for 5% of all types of CHD
  - Increased risk with infants of diabetic mothers, maternal etOH ingestion or poor nutrition during pregnancy
  - Predominately seen with males (2-3x)
  - Can be associated with other forms of CHD – VSD, PS, coarctation of the aorta, coronary anomalies, dextrocardia, heterotaxy syndrome (in particular RAI)

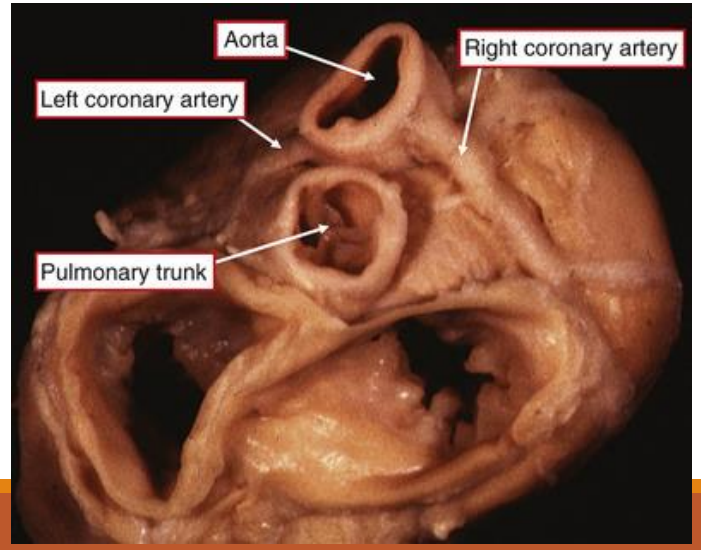



Aorta
Subaortic infundibulum
Morphologically right atrium
Morphologically right ventricle
Tricuspid valve
A


Pulmonary trunk
Morphologically left ventricle
Fibrous continuity
Mitral valve
B


Pulmonary trunk
Morphologically right appendage
Aorta
Morphologically right ventricle
A


Aorta
Pulmonary trunk
Morphologically right ventricle
B

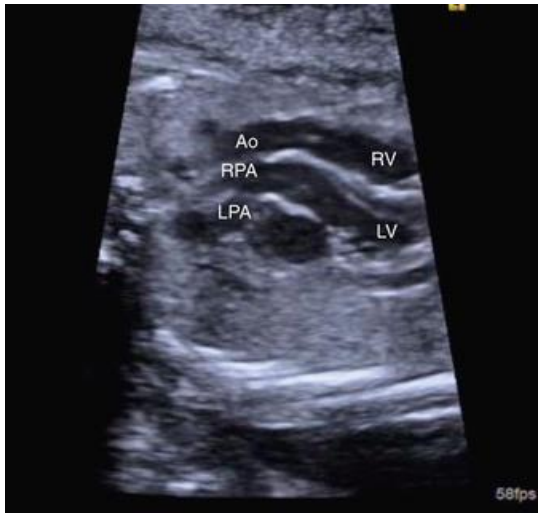

Aorta
Right coronary artery
Left coronary artery
Pulmonary trunk

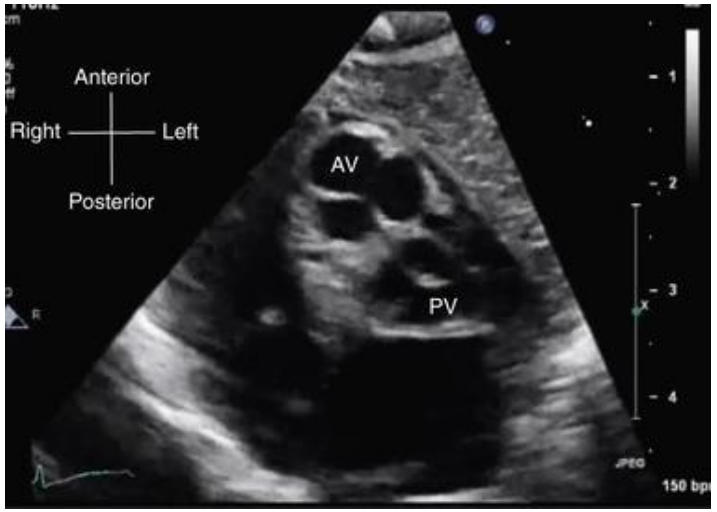
D-TGA Physiology

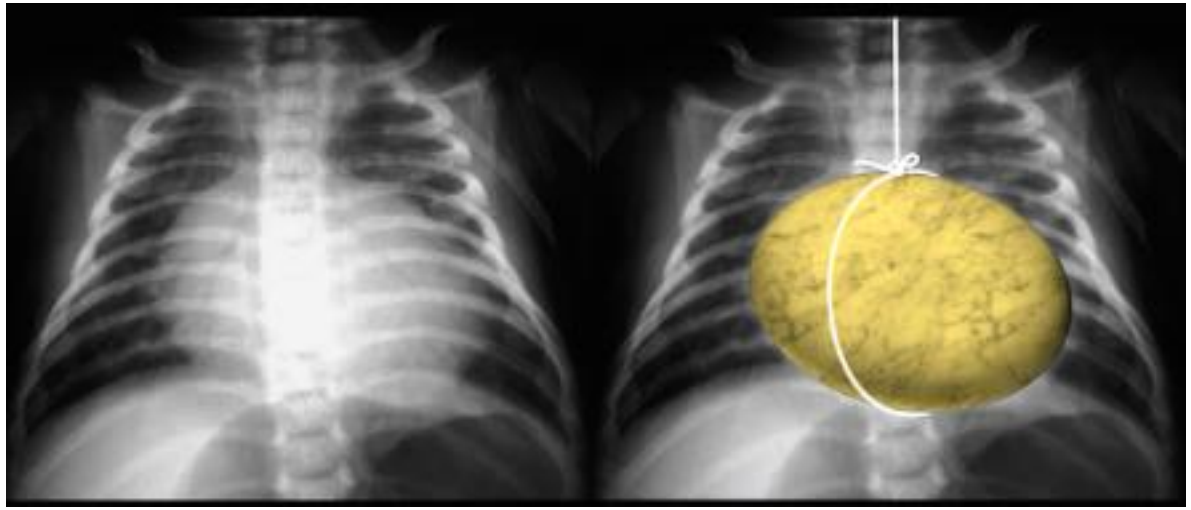
- Separate parallel circulations
  - Systemic venous return → RV → Aorta
  - Pulmonary venous return → LV → Pulmonary artery
- Without adequate intra-cardiac mixing, there can be severe systemic arterial desaturation following birth
  - PDA
    - Promotes mixing at the ductal arterial level but also promotes intra-atrial mixing by increasing left atrial return which increases left atrial pressures
  - PFO
    - Usually not alone adequate to promote intra-atrial mixing – so will require transcatheter balloon atrial septostomy
    - Up to 12% cases will require urgent balloon atrial septostomy following delivery due to severe acidosis and hypoxemia from inadequate mixing
- Can have different physiological presentation if have associated VSDs and/or outflow tract obstruction
- Prenatal diagnosis and initial post-natal management is crucial in minimizing preoperative mortality (3-10%)
  - Can be profoundly cyanotic following delivery if there is not adequate intra-cardiac mixing → will cause significant metabolic acidosis causing them to have decreased global perfusion, cool extremities and weak pulses
- In the neonatal period while the PVR is still physiologically high, LV work load remains elevated to help maintain normal LV mass
- However as the PVR starts to physiologically drop, there is a progressive reduction in LV pressure and work load resulting in the LV myocardial mass to progressively decrease with time
- Timing of neonatal surgical intervention is crucial given this physiological progression
  - High likelihood of post-operative LV failure if the LV is no longer trained following this transition period

Diagnostic Tools for D-TGA

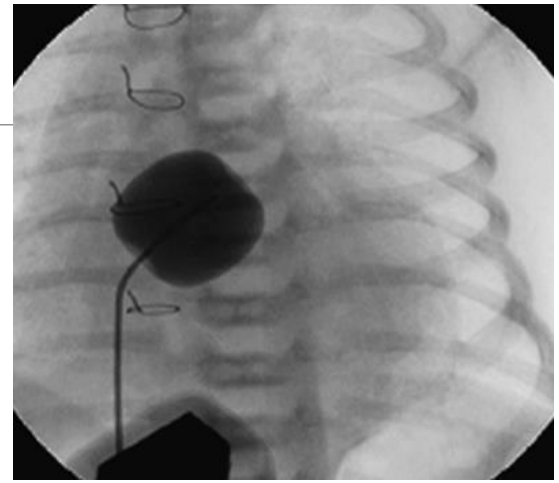
- ECG – typically normal following delivery
- Chest Xray – cardiac size is often normal, mediastinum may appear narrow due to the anteroposterior relationship of the arterial trunks (“egg on a string”)
- Echocardiogram



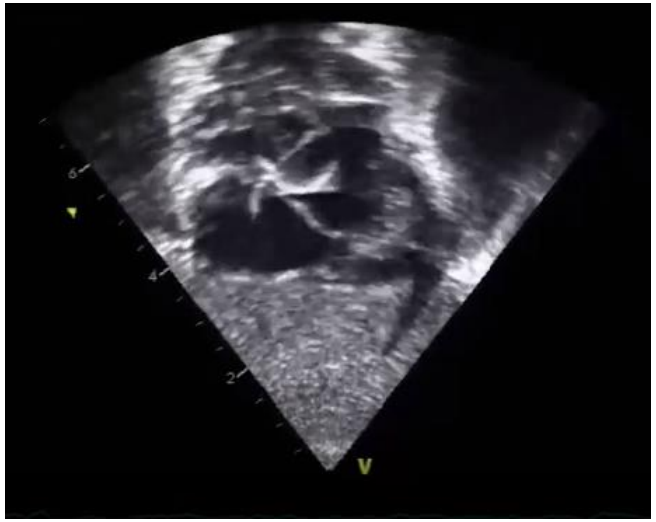




Medical Management for D-TGA

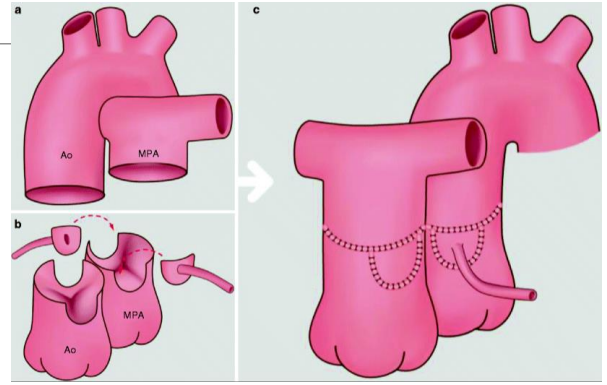


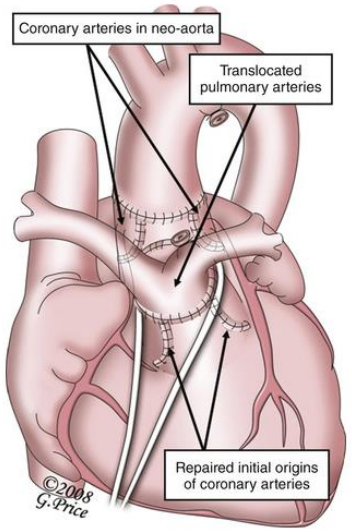
- Post-natal stabilization is crucial!
  - Venous access should be obtained quickly so that PGE can be initiated
  - If significantly cyanotic → consider intubation with mechanical ventilation
  - Increasing FiO2 may improve oxygen saturations by increasing pulmonary blood flow but typically has poor response to increasing the effective Qs
    - Important to pay attention to your PaO2 (not just your oxygen saturation)
  - Treatment of metabolic acidosis and consideration of inotropic initiation if in significant distress
  - Optimize oxygen carrying capacity by optimizing hematocrit
- Balloon atrial septostomy
  - Emergent (12%) vs elective timing



Surgical Management for D-TGA

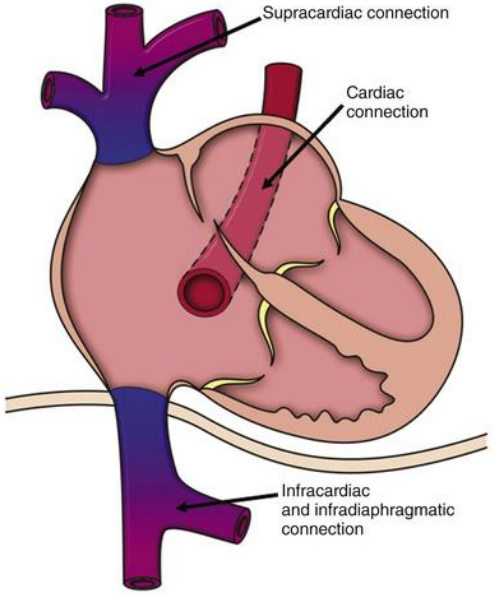
- Surgical intervention
  - Arterial Switch operation
    - Typical timing targeted for the first week of life
    - Targeting timing 3-4 days of life shows most favorable outcomes
    - Low operative mortality (Society of Thoracic Surgeons Congenital Heart Surgery Report)
      - 2.7% with intact septum
      - 5.3% with VSD
    - Factors that are associated with increased mortality risk → VSD, prolonged cardiopulmonary bypass time, early gestational age, institutional volume and certain coronary anomalies
    - Excellent long term survival outcomes → 25 year survival rate 97%
    - Long term complications: Reduced exercise capacity (82%), poorer neurodevelopmental outcomes (65% receiving remedial academic or behavioral services), coronary arterial obstruction (10%), neo-aortic dilation, neo-aortic valve insufficiency, branch PA stenosis (most common indication for reintervention)
  - Other potential surgical interventions if have additional cardiac findings (ie PS) – Rastelli operation, Nikadoh operation and Reparation a l'Etage Ventriculaire (REV) Procedure





Anomalous Pulmonary Venous Return

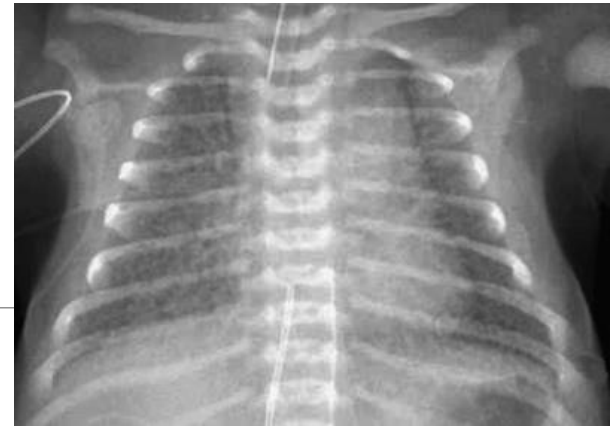
- A pulmonary vein is connected anomalously when it is attached to a site other than the morphological left atrium
- Total vs Partial
- Different types of TAPVR depending on the site of pulmonary venous drainage:
  - Supracardiac
  - Intracardiac
  - Infracardiac
  - Mixed
- Incidence of TAPVR accounts for ~1.5% of all CHD (1/14,700 live births)
- Can be associated with other forms of CHD – including VSDs and heterotaxy
- Can be associated with certain genetic syndromes: Holt-Oram, Klippel-Feil, phocomelia and Schachenmann syndromes



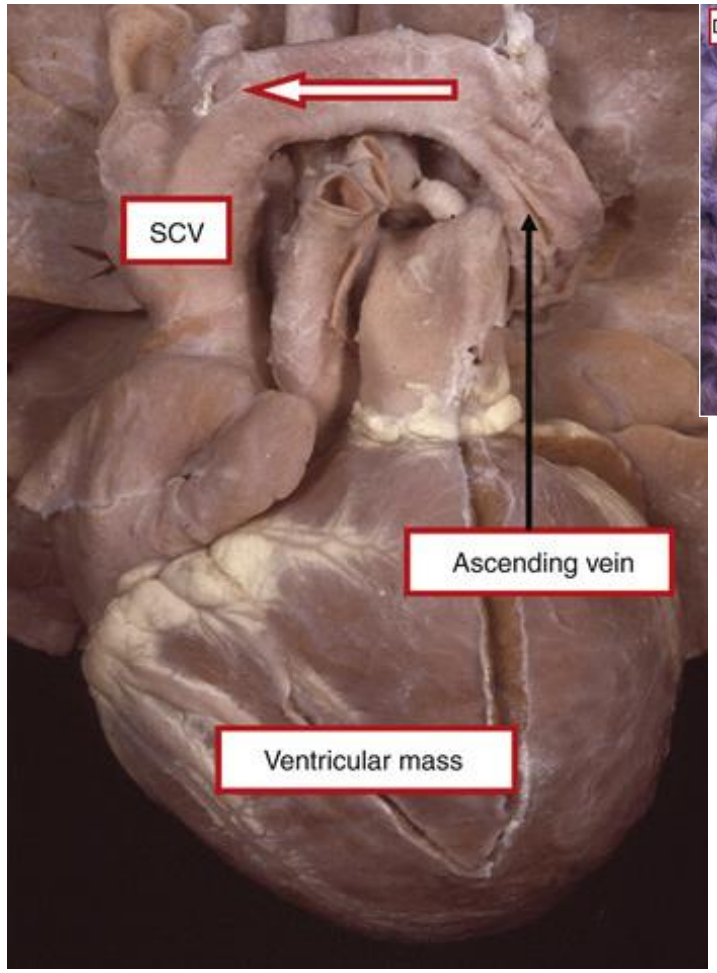
TAPVR Pathophysiology

- Because this is an obligatory left to right shunt, systemic cardiac output can only be maintained if there is a right to left shunt (ie atrial or ventricular septal level or ductus arteriosus).
- Can have associated pulmonary venous obstruction
  - Can be unmasked over time as PVR physiologically drops
- Can also be at risk for pulmonary hypertension secondary to pulmonary vascular changes that can begin during fetal life
- Survival rate past one year of age without surgical intervention is very poor (mortality rate of 92-96%)
- Presentation can be variable depending on the degree of obstruction:
  - Without obstruction – can present in heart failure within the first 2-3 months of life (similar to VSD physiology)
    - Tachypneic with retractions, poor weight gain, hepatomegaly, cyanosis may not be quite as evidence with an adequate right to left shunt, may have wide fixed splitting of S2 with a pulmonary flow murmur or a mid-diastolic murmur secondary to increased flow across both the tricuspid and pulmonary valves
  - With significant obstruction – cyanotic with potential signs of low cardiac output; present earlier in life (typically within the first 1-2 weeks)
    - Severely cyanotic, skin mottling, poor peripheral pulses, may hear a venous hum in the region of pulmonary venous obstruction, may be tachypneic but with quiet respiration

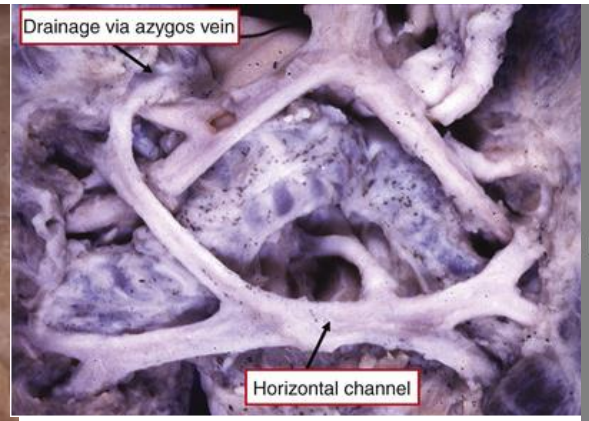
Diagnostic Tools for TAPVR

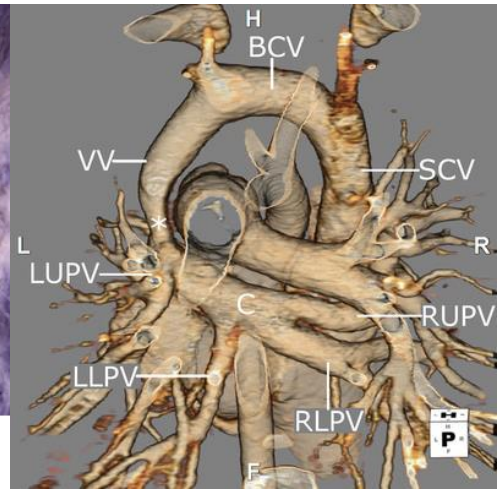


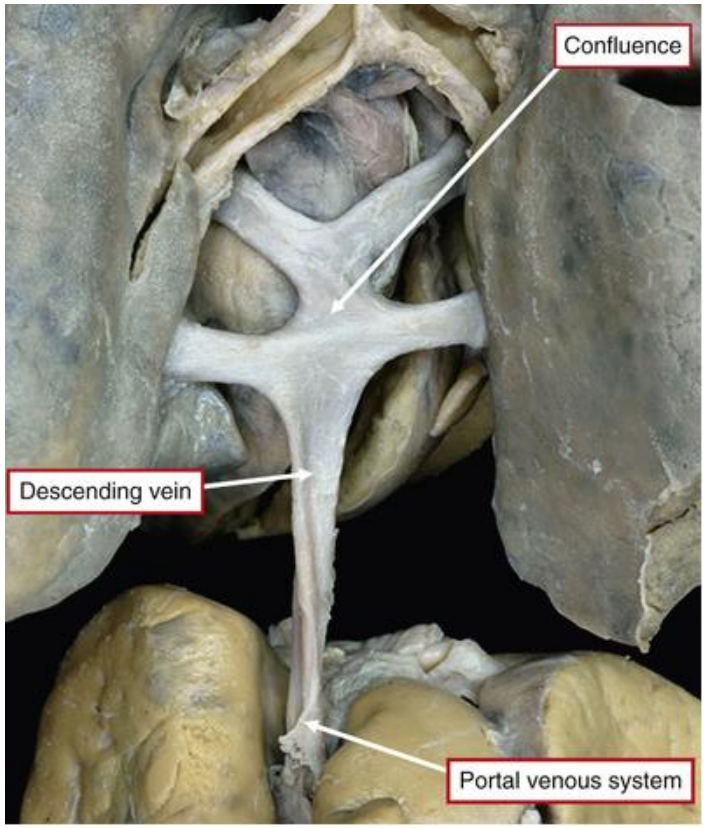
- Chest Xray findings
  - If unobstructed – heart appears enlarged due to RV volume overload
    - With supracardiac TAPVR – can see “snowman” appearance from the dilated left vertical vein and innominate vein
  - If obstructed – heart may appear small or normally sized framed by ground glass lung fields
    - Ways to differentiate from RDS - with TAPVR typically it’s rare to see air bronchograms and also tend to have a uniform distribution of changes
- Echocardiography is key in the diagnosis and also in the evaluation of obstruction
- Cardiac CTA is also a helpful tool in establishing the diagnosis and assisting in surgical planning

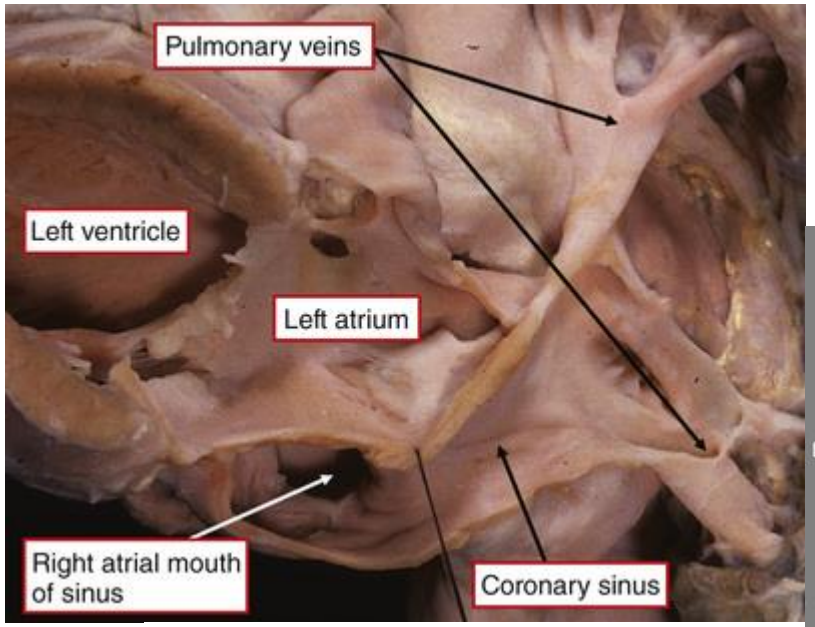


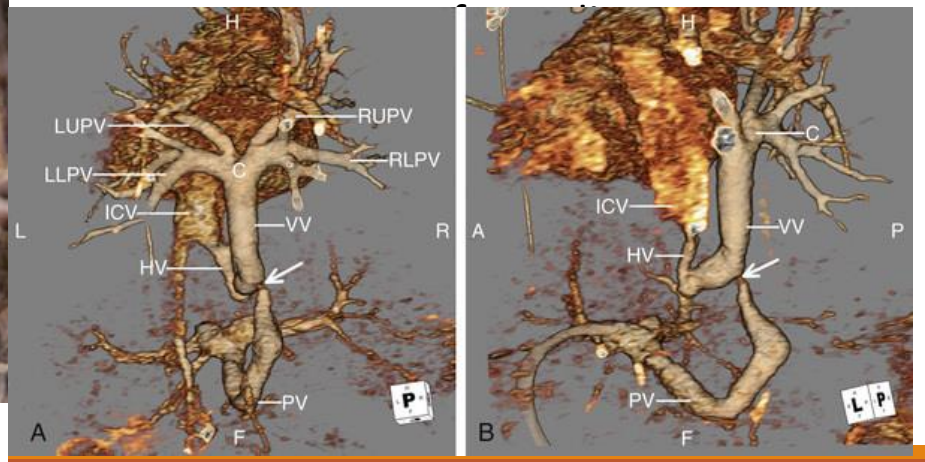
Supracardiac TAPVR











Intracardiac TAPVR

Medical Management for TAPVR

- Medical management consists of supportive measures in preparation for surgical intervention
- Unobstructed TAPVR – aimed at optimizing right heart failure and hypoxia
- Obstructed TAPVR – aimed at correcting hemodynamic compromise and metabolic acidosis
- Management considerations:
  - Intubation and ventilation
  - If has significant venous obstruction try to minimize medications that optimize pulmonary blood flow (ie iNO)
  - Consideration of PGE initiation – have to be cautious though, may improve systemic cardiac output at the expense of pulmonary blood flow
  - Can consider utilization of diuretics but need to be cautious if venous obstruction is present – a noncompliant RV may be reliant on preload to maintain contractility
  - Balloon atrial septostomy has not been shown to have a significant impact on hemodynamic improvement
  - Can consider endovascular stenting in obstructed vertical veins as a palliative bridge to surgery in high risk neonates
  - Can consider ECMO as a potential bridge to surgery in patients presenting with obstructed TAPVR and cardiogenic shock

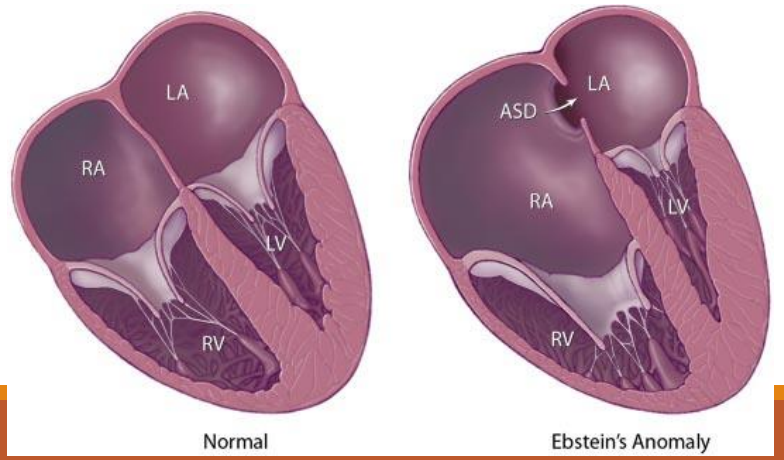
Surgical Management for TAPVR

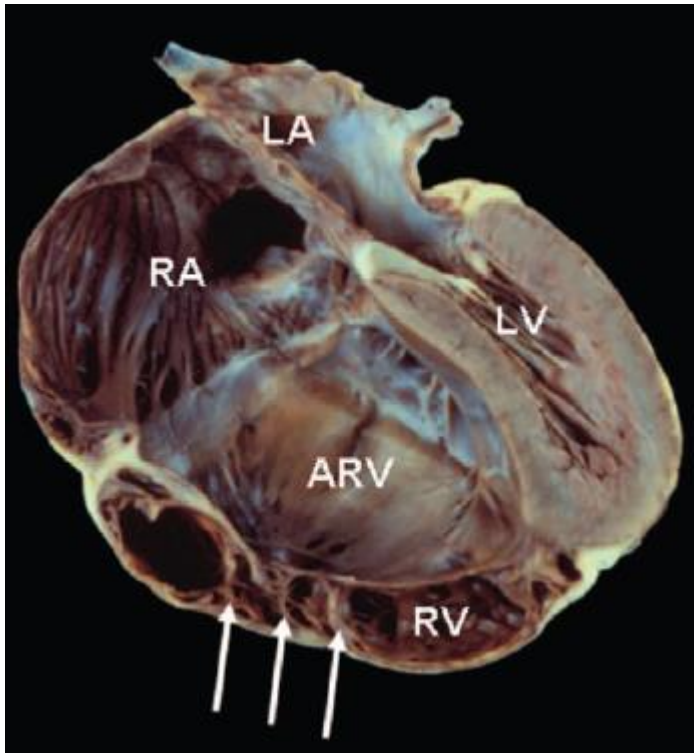
- Timing for surgery is influenced by the clinical status of the child
  - As outcomes for neonatal surgical intervention have improved, many have started advocating for early elective neonatal repair in unobstructed cases.
- Pre-operative mortality risk factors:
  - Preoperative pulmonary venous obstruction, single ventricle physiology, high PA systolic pressures, preoperative need for inotropic agents, post-operative events secondary to PHTN, infradiaphragmatic and mixed types of TAPVR
- ~8% of children undergoing surgical repair for TAPVR require ECMO post-operatively
- Surgical mortality in the current era can be less than 5% in patients with isolated disease
- Post-operative pulmonary vein stenosis (PVS) can occur in 10% of patients
  - Typically occurs within the first year following surgical repair
- Long term survival rate – 84% 17 year survival rate
  - Most deaths occurring within the first 24 months following surgical intervention

Valvular Abnormalities

Ebstein Anomaly

- Rare congenital cardiac malformation in which the tricuspid valve leaflets fail to properly delaminate from the ventricular wall
- Most common congenital tricuspid valve malformation
- Occurs in about 0.005% of live births and less than 1% of CHD
- Most cases are sporadic but there are certain risk factors that can be associated with the diagnosis:
  - Twin gestation, family h/o CHD, certain genetic mutations (cardiac tf NKX2.5 mutations, 10p13-p14 deletions and 1p34.3-p36.11 deletions), maternal medication exposures (ie lithium)
- Can be associated with other forms of CHD:
  - ASD/PFO (80-94%), VSD, pulmonary valve disease, aortic valve disease, coarctation of the aorta, CC-TGA, LVNC cardiomyopathy
- High risk for arrhythmias
  - High prevalence of atrial and accessory pathway mediated tachyarrhythmias
    - 6-36% of patients have > 1 accessory pathways
    - 25% have WPW
  - Higher risk of sudden cardiac death




LA
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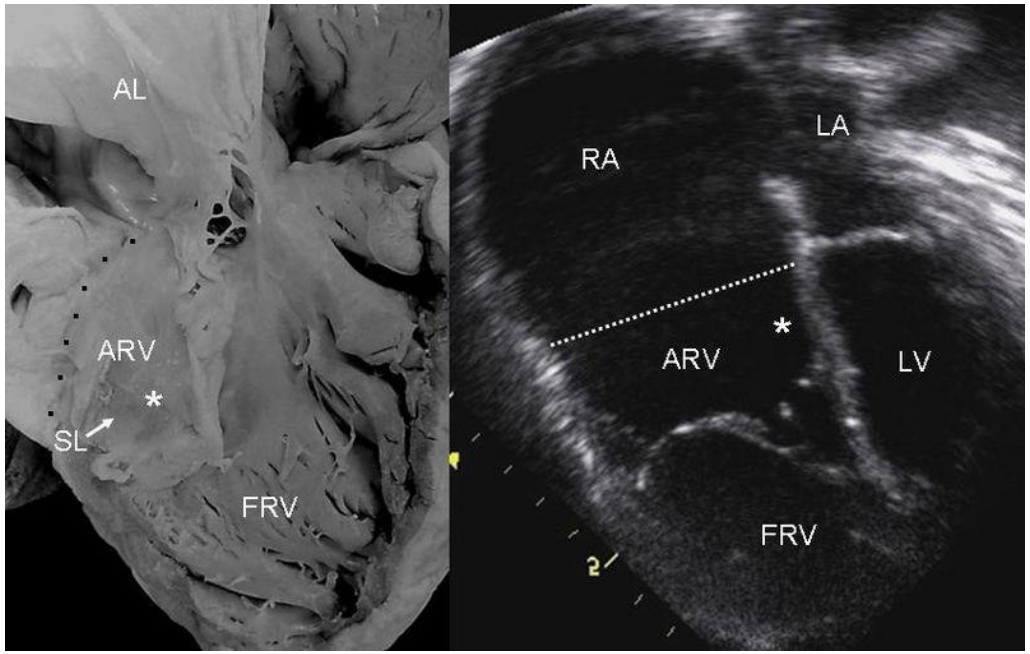
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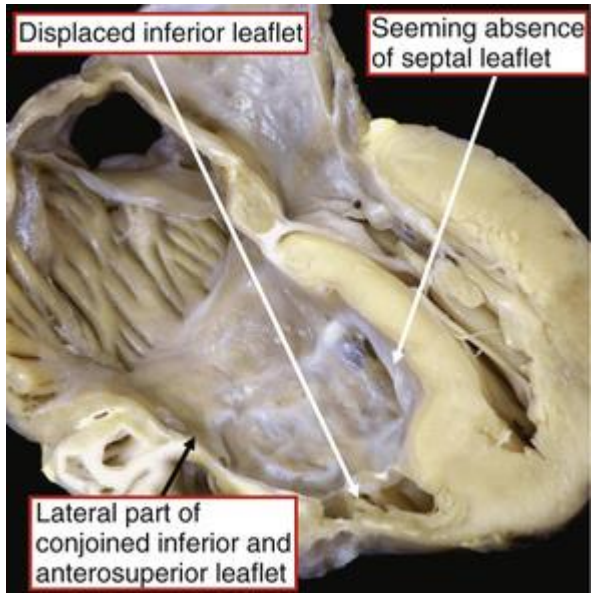
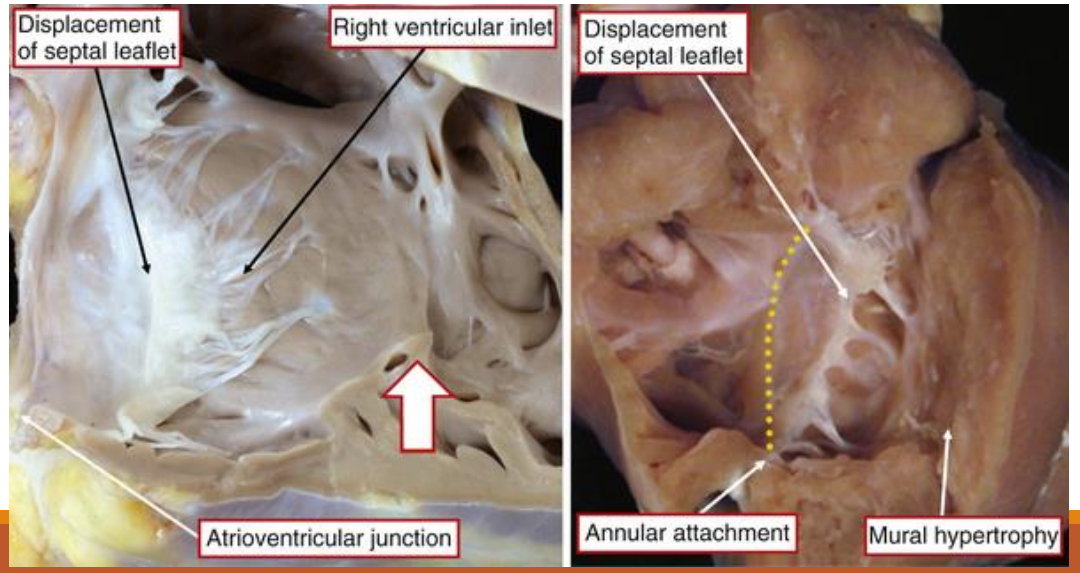
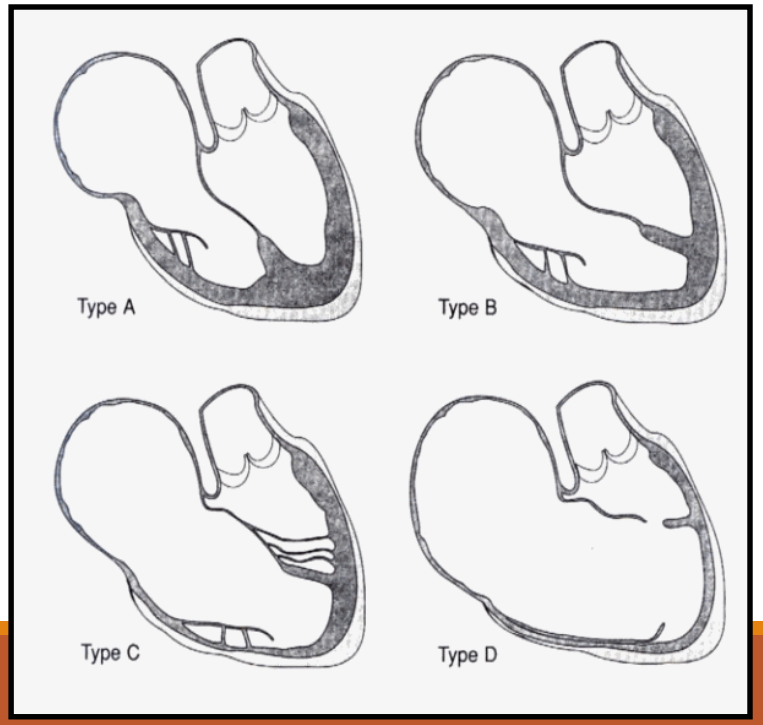

Displaced inferior leaflet
Seeming absence of septal leaflet
Lateral part of conjoined inferior and anterosuperior leaflet


Displacement of septal leaflet
Right ventricular inlet
Atrioventricular junction
Displacement of septal leaflet
Annular attachment
Mural hypertrophy

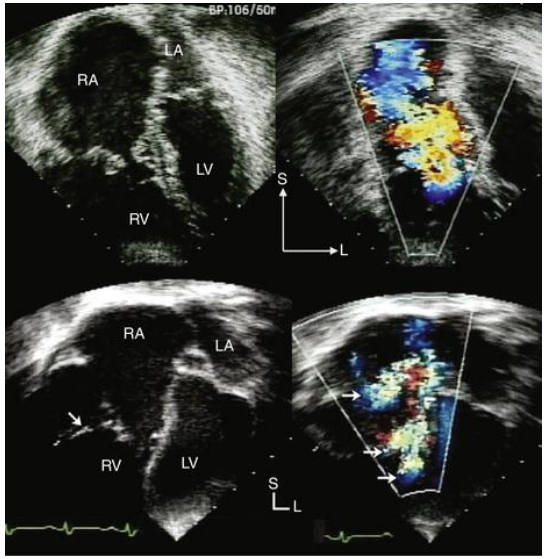

Type A
Type B
Type C
Type D

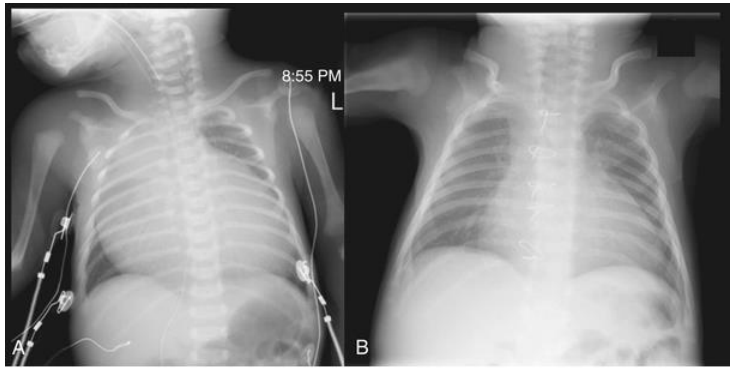
Pathophysiology of Ebstein Anomaly

- The initial presentation can be variable and depends on the severity of TV dysfunction and impacts on ventricular function
- Severe cases typically present prenatally or in the neonatal period
  - Can show fetal evidence of significant cardiomegaly secondary to severe TR and decreased ventricular function → can lead to hydrops and pulmonary parenchymal hypoplasia
  - Can also have functional pulmonary atresia → if develops pulmonary insufficiency can lead to circular shunt
  - Newborns often present with cyanosis
  - Slightly older infants may present with a combination of desaturation and symptoms of cardiac failure

Diagnostic Tools for Ebstein Anomaly

- ECG – this is an important tool due to the increased risk of atrial arrhythmias in this patient population
- Chest Xray – In severe cases will see significant cardiomegaly with hypoplastic lung fields (“wall to wall” heart), pulmonary vascular markings will also be decreased in severe cases
- Echocardiogram
- Cardiac MRI





Natural History of Ebsteins Anomaly

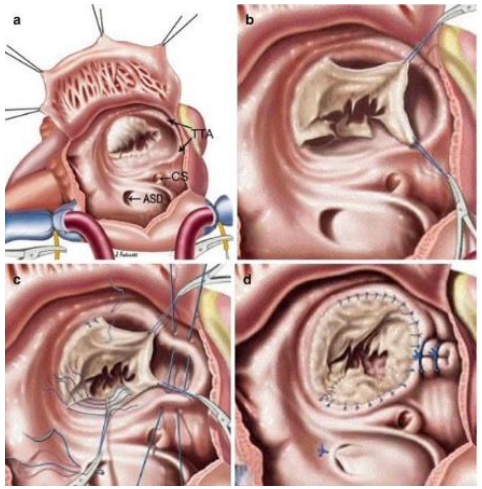
- Of all neonates diagnosed with Ebstein Anomaly, 20-40% of them do not survive past 1 month and ~ 50% survive to 5 years.
  - In cases of severe Ebstein Anomaly – neonatal mortality has been reported as high as 95% in the first month of life
- Factors that are associated with poorer outcomes:
  - A ratio of combined right atrial and atrialized ventricular area in comparison to functional right ventricle and left ventricle > 1
  - Larger atrial septal defects
  - Functional or anatomical pulmonary atresia
  - Left ventricular systolic dysfunction

Medical Treatment for Ebstein Anomaly

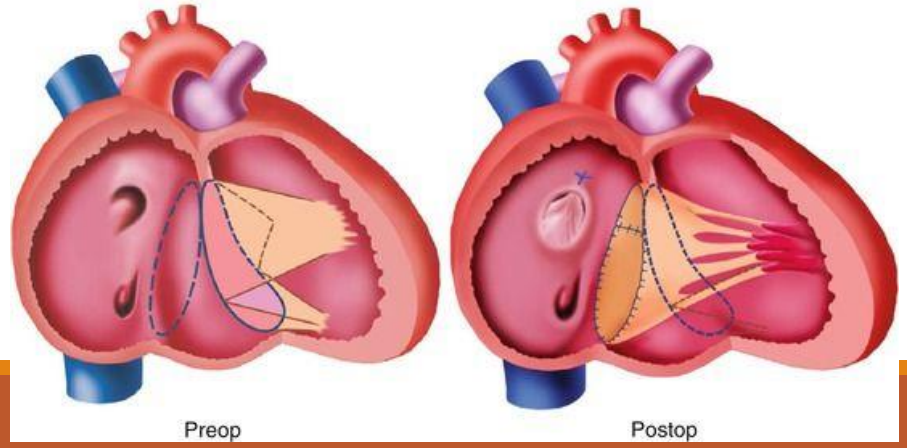
- Will be variable based on presentation
- Severe cases in the neonatal period – treatment often focuses on PVR reduction strategies and providing inotropic support
  - Consideration of PGE initation to maintain ductal patency to provide pulmonary blood flow
    - Have to be careful if significant pulmonary valve insufficiency – can lead to circular shunt
  - Pulmonary vasodilators such as FiO2 and iNO
  - Intubation and mechanical ventilation strageties
  - Providing inotropic support – consideration for epinephrine, milrinone
  - Managing potential arrhythmias
- Symptoms may improve with natural physiological transition/decrease in PVR
- May consider ductal stenting in certain cases with associated pulmonary atresia (either functional or anatomical)

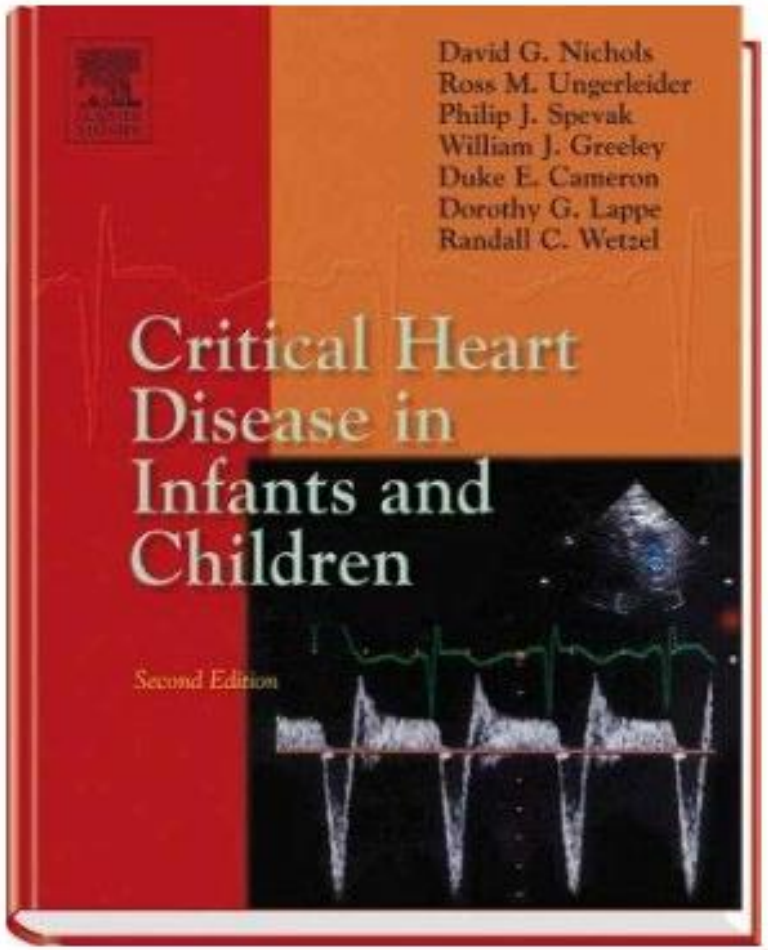
Surgical Interventions for Ebstein Anomaly

- Indications for surgery:
  - Significant symptoms → fatigue, cyanosis, decreased exercise tolerance, poor growth, the presence of an ASD, paradoxical embolism, increasing cardiomegaly, and onset/progression of atrial arrhythmias
  - Asymptomatic patients → at least moderate valvular distortion and cardiac enlargement



- Surgical options:
  - Neonates or infants with persistent cyanosis – weigh the risk of biventricular vs univentricular repair
    - Biventricular repair → tricuspid valve repair with subtotal ASD closure
    - Univentricular repair options:
      - Starnes procedure with closure of tricuspid valve with a fenestrated patch, atrial septectomy and placement of a surgical systemic to pulmonary arterial shunt
      - Bidirectional Glenn anastomosis ~3-6 months of age
  - Tricuspid valve repair or replacement
  - Selective plication of the atrialized RV
  - Reduction atrioplasty
  - Closure of atrial level shunt
  - Arrhythmia surgical management – MAZE procedure

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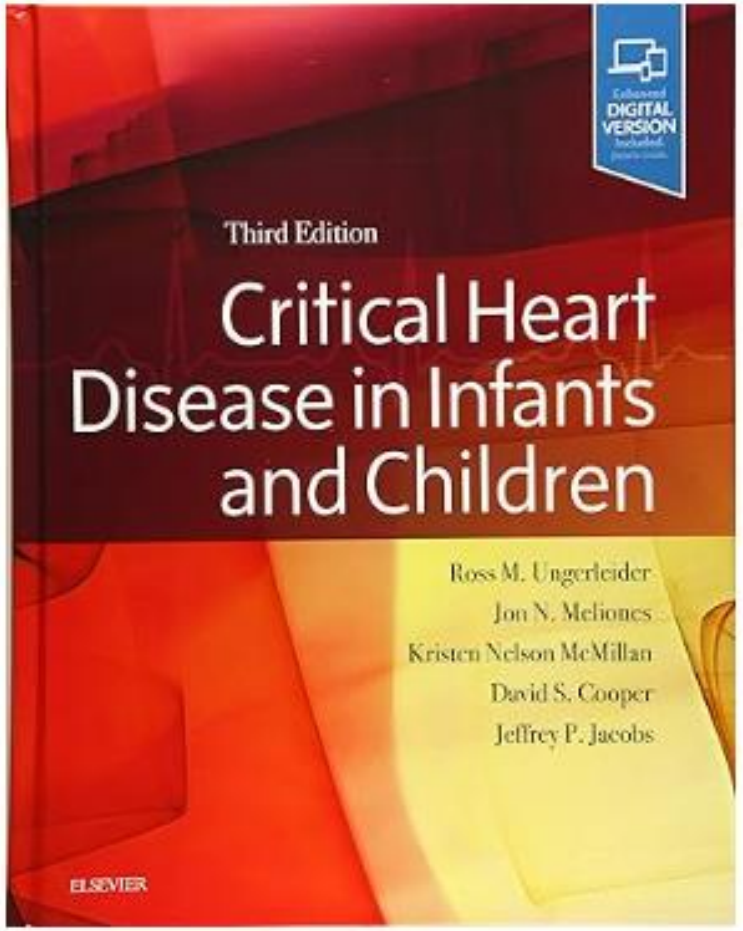
Second Edition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Questions?