ACYANOTIC HERAT DISEASE

Sarah Najim, MD

Introduction

- There are 3 major types of **Left** to **Right** shunts:

1. VSD (Ventricular Septal Defect)
2. ASD (Atrial Septal Defect)
3. PDA (Patent Ductus Arteriosus)

- It causes volume overload in the lungs.
- There is no cyanosis, because the systemic circulation blood is 100% saturated.
- Physiologic effect of the shunt depends on 3 factors:

1. Location of the shunt
2. Size of the defect
3. Relative pulmonary and systemic vascular resistance or ventricular compliance in case of atrial level shunt.

***Large shunts cause the pulmonary blood flow to increase and can be associated later with development of pulmonary arteriolar hypertrophy, increase in pulmonary resistance and pulmonary hypertension. Over time the elevated pulmonary resistance may force the direction of original shunt to reverse, causing R to L shunt and cyanosis.**

***The development of pulmonary vascular disease as a result of chronic left to right shunt is known as Eisenmenger Syndrome.**

VSD

- It is known that the most common CHD is VSD. However, recent studies show that bicuspid aortic valve is now #1.
- VSD is a communication between the LV and RV (ventricular level L to R shunt). So we have red blood going to the RV.
- The types of VSD differ in terms of management and prognosis, but they share the same physiology. **VSD is categorized into groups according to the site of the defect:**

1. Peri-membranous VSD (most common): co-ventricular defect around the membranous part of the ventricular septum (remember that the ventricular septum is mostly muscular, except for a small membranous part underneath the aortic valve). There is an incidence of aortic valve prolapse and aortic insufficiency.
2. Muscular VSD (2nd most common): one or more muscular defects in any part of the ventricular septum (anterior, apical, mid, posterior).
3. Inlet VSD or AV canal (more than a VSD): atrioventricular canal defect, also known as “endocardial cushion“ defect. This defect is associated with abnormal mitral and

tricuspid valves. Instead of being 2 valves, they are actually 1 valve (common junction). Complete AV canal is common in patients with trisomy 21 (Down syndrome).

4. Sub-pulmonary defect (conal septal hypoplasia) – NOT required.

Physiology

- A general rule: the heart will do whatever it can to make the CO normal. We will assume that the CO = 4 L/min. These 4 L will come back to the RA then to the RV, then another 4L will cross the VSD from the LV to the RV, then to the pulmonary artery, LA and LV.
- The ratio between pulmonary blood flow and systemic blood flow (Qp:Qs) is 2:1, volume overload.
- **The chambers that will be affected in VSD:**

1. LA dilation because it is receiving 8 L of blood.
2. LV dilation because it is receiving 8 L during diastole (it fills with 8 L).
3. RV hypertrophy due to pressure load (there is a hole between the 2 chambers, one is pumping at high resistance and the other is pumping at low resistance; so the pressure will be transmitted to RV thru systole. The RV will not be affected by any volume changes because the pulmonary valve is open, so all of the 4L will go directly to the lungs).
4. Pulmonary artery dilation because it is receiving more blood. NO changes in RA (at least early on) and CO well be maintained even in large VSDs.

***VSD shunt is systolic.**

- The hemodynamic changes that occur in VSD depend on the size of defect and relative resistance in pulmonary and systemic vasculature. In small VSD, the defect itself offers more resistance to flow than pulmonary or systemic vasculature, thus the magnitude of shunt depends on the hole. Conversely with larger defects, the volume of shunt depends on systemic and pulmonary resistances. Remember that in the perinatal period the pulmonary vascular resistance is approximately equal to the systemic resistance, so minimal shunting occurs between the ventricles. However, after birth the pulmonary resistance falls and the blood will be shunted to the right along the pressure gradient.

Symptoms

- Newborns with VSD are usually well (asymptomatic, especially if small VSD) because their pulmonary vascular resistance is high (right side of heart receives all systemic venous return, including blood from placenta). It takes about 4-8 wks for their pulmonary resistance to decrease to the normal adult level. So, the L to R shunt is minimal in the newborn period.
- With the first breaths the baby takes up on delivery, resistance to pulmonary blood flow falls and the volume of blood flowing through the lungs increases 6-fold. This results in a rise in the LA pressure. Meanwhile, the volume of blood returning to the RA falls as the placenta is excluded from the circulation. The change in the pressure difference causes the flap of the valve of the foramen oval to be closed. The ductus arteriosus also normally closes within the first few hours or days.
- Large VSD > 50% of the aortic width / Small VSD = 1-3 mm / Moderate VSD = in-between.
- Moderate to large VSD: LA/LV dilation due to increased pulmonary blood flow. Unlike ASD, congestive heart failure may follow (according to the Frank-Starling law, excessive

dilation of the LV will cause it to lose its contractility. So, there will be systolic dysfunction, the patient will become decompensated, and the CO will drop).

- Symptoms of heart failure in infants or babies are different than those seen in adults. Usually, infants with large VSD present with *respiratory symptoms due to fluid accumulation in the lungs (v.imp) For example: tachypnea, tachycardia, diaphoresis (sweating during feeding), decreased or difficulty feeding, resp.distress, failure to gain wt, recurrent LRTI, etc.
- FTT, usually due to large VSD.
- Compensated patients deteriorate rapidly with infection.

Examination

1. Newborns may NOT have heart murmur in the first day of life because the pressure in both ventricles is almost equal (high pulmonary vascular resistance). So, there is NO turbulence of blood across the VSD, meaning NO murmur may be heard until about 4-8 wks of life (musical grade 2 ejection systolic murmur).
2. Displaced apex beat because the LV is dilated.
3. Hyperdynamic precordium with large, visible pulsations.
4. Pan-systolic (holosystolic) murmur is common after the first day of life due to blood flow across the VSD.
5. Small muscular VSD may be associated with short systolic murmurs (These murmurs are actually pan-systolic, but we don't label them as such because they stop before S2. This occurs because the defect is in the muscular part of the ventricular septum; the VSD is initially open but then closes upon itself during ventricular contraction. This is a good sign because it means that the defect is getting smaller in size, so at one point the murmur will disappear)
6. Loud P2 of second heart sound (because there is pulmonary hypertension).
7. Diastolic rumble due to excessive flow across a normal mitral valve (diastolic murmur at the apex usually indicates a large VSD).
8. S3 gallop may be present in patients with heart failure.
9. Hepatomegaly due to liver congestion if the patient developed right heart failure.

Investigations

- **ECG:** (helpful only beyond infancy)

1. Signs of left axis deviation
2. Signs of LV hypertrophy (LV dilation is a more accurate description)
3. Signs of LA dilation
4. Northwest (superior) axis in AV canal defects

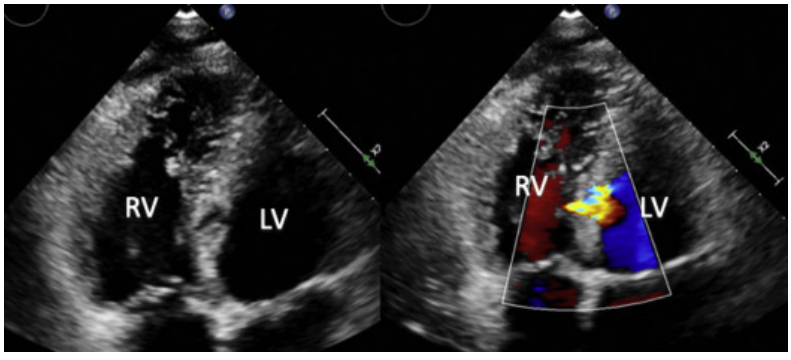
***NOTE: ECG does NOT differentiate between hypertrophy and dilation but it reflects the presence of a ventricular mass in general.**

- **CXR:**

1. Increased pulmonary flow
2. Increased vascular markings
3. Cardiomegaly (especially if large VSD)

- **Echocardiography:** (diagnostic)

☐ It diagnoses the presence and the type of VSD, as well as its effect on the other cardiac structures.



Management

- **Asymptomatic patients** do NOT need any treatment (we just wait for the VSD to close spontaneously. Spontaneous closure is common in small and moderate perimembranous and muscular defects.
- **Symptomatic patients** may need medications to alleviate symptoms.
- NO restriction from activity.
- NO SBE prophylaxis (according to the new guidelines). However, some people still give SBE prophylaxis while others do NOT.
- Surgical treatment is the standard treatment for symptomatic VSDs like the AV canal type VSDs because they don't close spontaneously.
- If Eisenmenger syndrome occurs, then surgery is **contraindicated**.
- Transcatheter closure of certain types of VSD can be done.

ASD

- ASD is an atrial level shunt (persistent opening of interatrial septum after birth that allows direct communication between the LA and RA).
- The defect can occur anywhere along the septum and produce different types of ASD that share the same physiology (ex. secundum ASD, primum ASD – NOT required).

Physiology

- The CO in patients with ASD is normal, which means that the heart will pump the same volume of blood to the body as a normal heart (normal CO= 3-5 L/min).
- Lets assume that the CO in a patient = 4 L (volume of blood leaving the heart) and the O2 saturation = 100%. It will return to the systemic veins as 4 L.
- Then Let's assume that the ASD (between the LA and RA) will allow another 4 L of blood to cross per minute (from high pressure of LA to low pressure of RA). So, we will have 8 L in the RA and RV. This volume will go to the lungs and will come back to the LA as 8L. Then it will be divided into 4 L across the ASD and 4 L into the LV (which is the normal CO). So at

the equilibrium state, there is compensated CO in patients with ASD. This means L to R shunt does NOT compromise the systemic blood flow, but it increases the pulmonary blood flow.

***In the RA 1/2 of blood is arterial with 100% O2 sat and 1/2 of blood is venous with 70% O2 sat. They will mix and the RV out put will be 8L blood with 85% O2 saturation.**

- Based on the above, changes in heart chambers will be:

1. LA dilation (it becomes bigger): because it is receiving 8 L of blood instead of 4 L (twice the normal amount).
2. RA dilation
3. RV dilation
4. Pulmonary artery dilation

- These areas are receiving more blood than usual, so they are subjected to volume overload. Volume overload gives dilation in chambers and stenosis on valves (tricuspid and pulmonary valves = stenotic due to chronic shunting and volume overload).
- The majority of shunts occur during diastole (ventricular filling, ventricular diastole = atrial systole).
- The LV in patients with ASD is NOT affected, so there is NO LV dilation. This means that the patient will NOT develop any sign of heart failure or decreased tissue perfusion.
- Also the patient will NOT have any type of holosystolic murmur, because the flow across the ASD is at low pressure.

Symptoms

- Usually there are NO symptoms (**asymptomatic**).
- Some patients who present with FTT (failure to thrive) are incidentally found to have ASD, but we should look for other causes of FTT.
- ASD **aggravates** the symptoms of pulmonary diseases (ex. asthma, pneumonia, etc) but does NOT cause them. So, children with underlying pulmonary disease may be more symptomatic with ASD due to increased pulmon. blood flow.
- There is NO congestive heart failure (very rare).
- Older children may complain of dyspnea on exertion only due to increased pulmonary blood flow (asymptomatic at rest).
- Pulmonary hypertension almost never happens in young patients with ASD because it is a low pressure shunt (less than 10% risk).
- Older patients may present with arrhythmias like SVT (supraventricular tachycardia) or atrial/vent. tachy due to RA dilation.
- Paradoxical emboli may occur in older patients (rare in children).

***A paradoxical embolus is a venous embolus that by passes the lung due to R-L shunt through ASD, and becomes arterial embolus. It occurs with reversal of the shunt in EISENMENGER syndrome.**

Examination

1. Normal in young infants
2. Prominent RV heave due to RV dilation

3. Wide, fixed split of S2 because the RV is pumping twice as much blood to the lungs across the pulmonary valve. So, systole will occur over a longer period of time and P2 will happen later than A2 of second heart sound.
4. Ejection systolic murmur of the pulmonary valve because there is more blood across a normal valve (relative pulmonary stenosis).
5. Diastolic murmur (rumble) of the tricuspid valve (if the ASD is very large, due to increased blood flow thru the tricuspid valve).

Investigations

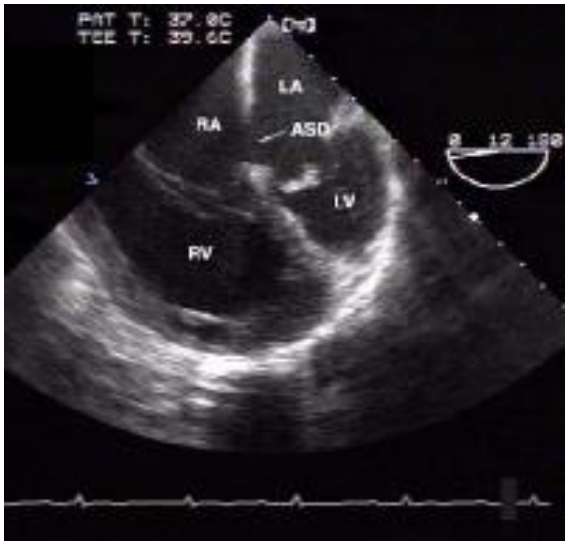
- **ECG:** (beyond infancy - findings reflect the physiology)

1. Signs of right axis deviation
2. Signs of RA dilation or hypertrophy
3. Signs of RV increased mass (the ventricular mass is increased, NOT necessarily hypertrophic)
4. Atrial arrhythmias + other things in specific types (pattern of incomplete right bundle branch block, peaked P waves in lead II, sinus node dysfunction in sinus venosus defects, northwest/superior QRS axis is typical of Primum ASD& AV canal defect).

- **CXR:**

1. Normal (most of the time)
2. Dilated RA, RV & pulmonary artery: RA or right border of the heart is dilated and extends more to the right (NO shifting of apex beat)
3. RV against sternum because it's dilated (lateral CXR)
4. Increased pulmonary blood flow (plethoric lungs)
5. Increased vascular markings (blood vessels are a little larger than normal. The lungs are NOT opaque and there is NO pulmon. edema. Actually, the lungs are "**plethoric**" (increased pulmonary blood flow)
6. Cardiomegaly (occasionally, because there is mainly RA dilation NOT LV dilation)

- **Echocardiograph:** the Diagnostic method.



Management

- The only method is the closure of the defect, if it is indicated (by surgery or by cathether). ASD: transcatheter closure is amenable in most cases. Sinus venosus, primum ASD and extremely large or deficient rim secundum ASD's require surgical closure.
- NO medications should be given.
- NO restriction from activity
- Spontaneous closure may occur in small or med. sized secundum ASD.

***There is an increased risk of subacute bacterial endocarditis (SBE), because with time the shunt damages the epithelium (breaks it down), which creates a good media for bacterial growth.**

PDA

Physiology

- PDA is a L to R shunt between the aorta and pulmon. artery.
- If we apply same numbers used in the above examples to PDA, we will find that it is very similar to VSD. It causes the same chamber changes, but it differs in that it also causes wide pulse pressure.

Symptoms

- Usually present in first 2 wks of life, when the PDA starts to constrict.
- Signs and symptoms of decreased systemic perfusion.
- Lethargy, and signs of CHF.
- Metabolic acidosis and shock develops quickly.
- Should be in the differential diagnosis of neonates with R/O sepsis.

Examination

- Continuous murmur beneath the left clavicle.
- Poor pulses – collapsing or bounding (wide pulse pressure).
- Poor capillary refill.
- May not detect radio femoral delay or BP gradient in patients with COA (coarctation of aorta).
- Ejection systolic murmur may indicate aortic stenosis.
- Tachycardia and gallop.
- Tachypnea.
- Hepatomegaly.

Investigations

- **ECG:** RV hypertrophy
- **Echocardiography**: Final diagnosis
- **CXR:** (like VSD)

1. Cardiomegaly 2. Pulmonary edema

Management

- In infants with asymptomatic PDA, closure at about 1 yr of age is recommended to abolish the lifelong risk of SBE.

Non-Shunting HERAT DISEASE

Coarctation of the Aorta

Physiology

- Coarctation is a non-shunt, obstructive lesion (coarctation= constriction or pinching).
- There is obstruction in the distal part of aortic arch (just distal to subclavian artery), which will cause the heart to pump at a higher pressure in order to perfuse the lower extremities.
- Eventually, there will be collateral blood flow across area of obstruction.

Symptoms

- Symptoms usually do not show at birth, they begin to emerge after 1 wk.
- Hypertension in the upper extremities (above the constriction).
- Normal or low blood pressure in the lower extremities.

Examination

1. LV hypertrophy with dilation of apex beat.
2. Difference in blood pressure between upper and lower limbs.
3. Normal or ejection systolic murmur between shoulder blades.

Investigations

- **ECG:** LV hypertrophy
- **CXR:** 1) Usually normal 2) Rib notching from development of collaterals

Management

- A child with severe coarctation should have surgery in early childhood, after which long-term follow up is necessary.
- Important note in patient with coaractaiton of aorta **Neonates or infants** with severe coarctation may depend on a right-to-left shunt through the PDA for perfusion of the lower thoracic and descending aorta. Such infants may be minimally symptomatic initially, but symptoms of CHF develop and progress as the PDA closes ,Blood pressure may be elevated in the upper extremities and low in the lower extremities before the onset of CHF. Once the infant or neonate develops CHF, pulses in all four extremities are poor, any murmur is absent, and hypotension may develop.
- **Older children or adolescents** may have no symptoms and may have only hypertension or a heart murmur,(radio-femoral delay).
- Blood pressure and pulse findings may be less prominent if collateral vessels (intercostal arteries) develop that allow the ascending aortic pressure and flow to circumvent the coarctation.
- Bicuspid aortic valve or aortic stenosis is present in 50% of patients. If either of these conditions is present, a systolic murmur of aortic stenosis may be heard Bruit of turbulence through the coarctation may be audible at the left upper back near the scapula.

Pulmonary Stenosis

Etiology

- Pulmonary stenosis accounts for approximately 10% of all congenital heart disease and can be valvular, subvalvular, or supravalvular in nature.
- Pulmonary stenosis results from the failure of the development, in early gestation, of the three leaflets of the valve, insufficient resorption of infundibular tissue, or insufficient canalization of the peripheral pulmonary arteries.

Symptoms & Examination

- Symptoms depend on the degree of obstruction present.
- Mild pulmonary stenosis is asymptomatic.
- Moderate to severe stenosis results in exertional dyspnea and easy fatigability.
- Newborns with severe stenosis may be more symptomatic and even cyanotic because of right-to-left shunting at the atrial level.
- Pulmonary stenosis causes a systolic ejection murmur at the second left intercostal space which radiates to the back.
- A thrill may be present.
- S2 may be widely split with a quiet pulmonary component.
- With more severe pulmonary stenosis, an impulse at the lower left sternal border results from right ventricular hypertrophy.
- Valvular stenosis may result in a click that varies with respiration.
- Worsening stenosis causes an increase in the duration of the murmur and a higher frequency of the sound.

- The systolic ejection murmurs of peripheral pulmonary stenosis are heard distal to the site of obstruction in the pulmonary circulation, including radiation to the back.

Investigations

- ECG and chest x-ray findings are normal in mild stenosis.
- Moderate to severe stenosis results in right axis deviation and right ventricular hypertrophy.
- The heart size is usually normal on chest x-ray, although dilation of the main pulmonary artery may be seen.
- Echocardiography provides assessment of the site of stenosis, degree of hypertrophy, and valve morphology, as well as an estimate of the pressure gradient.

Management

- Valvular pulmonary stenosis usually does not progress, especially if it is mild.
- Balloon valvuloplasty is usually successful in reducing the gradient to acceptable levels for more significant or symptomatic stenosis.
- Surgical repair is required if balloon valvuloplasty is unsuccessful or when subvalvular (muscular) stenosis is present.

Aortic Stenosis

Etiology

- Valvular, subvalvular, or supravalvular aortic stenosis represents approximately 5% of all congenital heart disease.
- Lesions result from failure of development of the three leaflets or failure of resorption of tissue around the valve.

Symptoms & Examination

- Mild to moderate obstructions cause no symptoms.
- More severe stenosis results in easy fatigability, exertional chest pain, and syncope.
- Infants with critical aortic stenosis may present with symptoms of heart failure.
- A **systolic ejection murmur** is heard at the right second intercostal space along the sternum and radiating into the neck. The murmur increases in length and becomes higher in frequency as the degree of stenosis increases.
- With valvular stenosis, a **systolic ejection click** often is heard, and a **thrill** may be present at the right upper sternal border or in the suprasternal notch.
- The aortic component of S2 may be decreased in intensity.

Investigations

- ECG and chest x-ray findings are normal with mild degrees of stenosis.

- Left ventricular hypertrophy develops with moderate to severe stenosis and is detected on the ECG and chest x-ray.
- Dilation of the ascending aorta or aortic knob due to an intrinsic aortopathy may be seen on chest radiographs.
- Echocardiography shows the site of stenosis, valve morphology, and the presence of left ventricular hypertrophy, and it allows an estimate of the pressure gradient.

Management

- The degree of aortic stenosis frequently progresses with growth and age. Aortic insufficiency often develops or progresses. Serial follow-up with echocardiography is indicated.
- **Balloon valvuloplasty** is usually the first interventional procedure for significant stenosis. It is not as successful as pulmonary balloon valvuloplasty and has a higher risk of significant valvular insufficiency.
- **Surgical management** is necessary when balloon valvuloplasty is unsuccessful or significant valve insufficiency develops.

Important Notes About the Lecture

- Tricuspid Atresia:
- ☐ The absence of the tricuspid valve results in a hypoplastic right ventricle.
- ☐ All systemic venous return must cross the atrial septum into the left atrium. A PDA or VSD is necessary for pulmonary blood flow and survival.
- ☐ Clinical Manifestations in Infants with tricuspid atresia are usually severely cyanotic and have a single S2.
- ☐ If a VSD is present, there may be a murmur (VSD murmur).
- ☐ ECG shows right atrial enlargement, left axis deviation (LAD), and left ventricular hypertrophy (LVH).
- ☐ Tricuspid atresia is the only cause of cyanosis in the newborn period that results in LAD and LVH.

Q: You are called to the nursery because of a neonate with deep central cyanosis, upon examination you find holosystolic murmurs at the left sternal border, ECG shows a left axis deviation, what is your diagnosis?

a. Tricuspid Atresia

- Oligemic lung field (decreased vascular markings): occur in diseases that reduce pulmonary blood flow (pulmonary/ tricuspid atresia , TOF , pulmonary hypertension).
- Plethoric lung fields (increase pulmonary blood flow): seen in Left to right shunt – ASD, VSD, PDA, Coronary artery fistula into right heart, Aortopulmonary window, Transposition of great arteries with ASD or VSD, Truncus arteriosus, Total anomalous pulmonary venous connection.
- In Turner syndrome; The most commonly observed are congenital obstructive lesions of the left side of the heart, leading to reduced flow on this side of the heart. This includes bicuspid aortic valve and coarctation (narrowing) of the aorta. More than 50% of the

cardiovascular malformations of individuals with Turner syndrome in one study were bicuspid aortic valves or coarctation of the aorta, alone or in combination.

- Other congenital cardiovascular malformations, such as partial anomalous venous drainage and aortic valve stenosis or aortic regurgitation, are also more common in Turner syndrome than in the general population. Hypoplastic left heart syndrome represents the most severe reduction in left-sided structures.