CYANOTIC HEART DISEASE

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Fetal Circulation

- **Fetal circulation differs from new born circulation in many ways:**

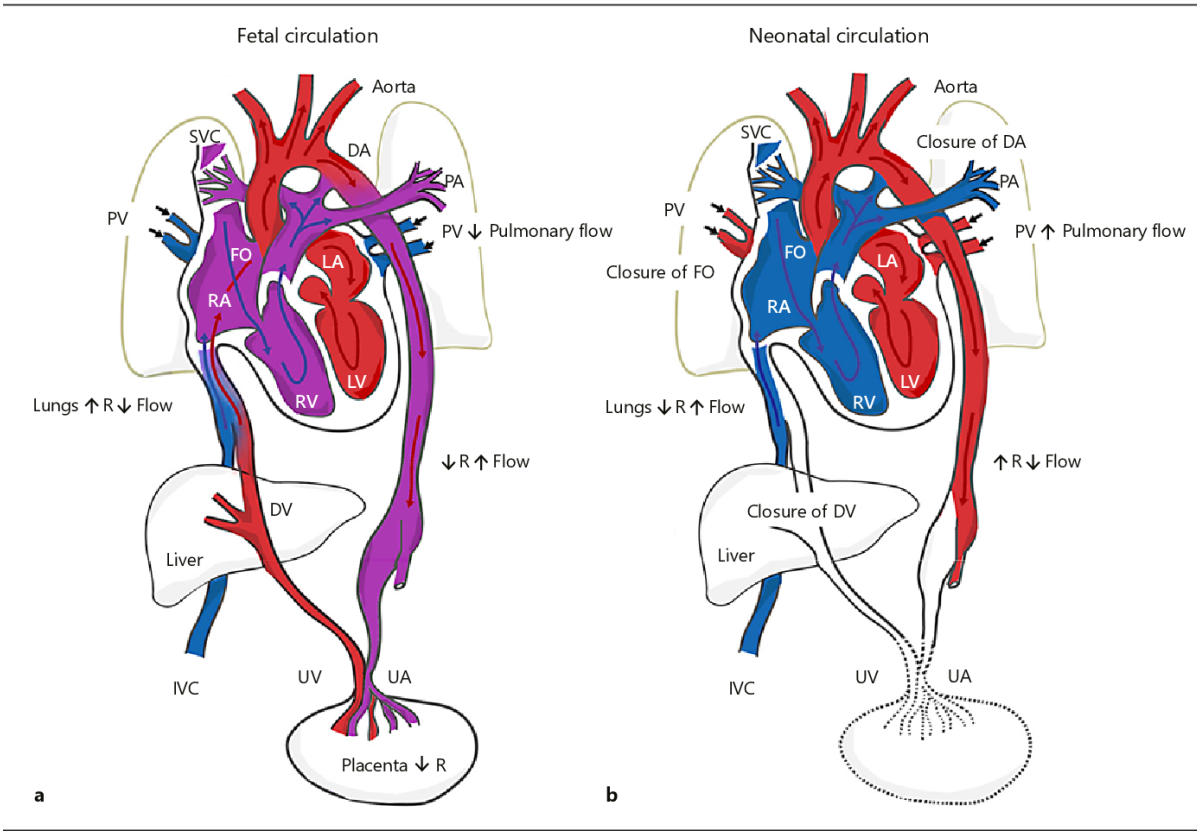
1. The cardiac output of the fetus is considered "total cardiac output"(CO) or "combined ventricular output" rather than the left ventricular (LV) and right ventricular (RV) output. This happens because the fetus does NOT use its lungs, as they are full of fluid. Instead of going to the lungs, the blood is directed across the patent ductus arteriosus to the descending aorta.
2. The organ responsible for oxygenation in fetal life is the placenta (NOT the lungs).
3. There are 3 communicating pathways that present in the fetal circulation and they close after birth:

Communication	Description	Function
Patent Ductus Arteriosus (PDA)	It is an opening which connects the pulmonary artery to the aorta	It directs blood away from the lungs because they are not used. So most of blood going to the pulmonary artery will cross the PDA to aorta. This means that the direction of flow across the PDA in the fetus is pulmonary to aorta (NOT aorta to pulmonary.
Patent Foramen Oval (PFO)	It is a foramen that presents between the left atrium (LA) and the right atrium (RA)	It directs blood from the right side of the circulation RA to LA. This blood is rich in oxygen because it comes mainly from the inferior vena cava (IVC) and from the placenta across the ductus venosus. In fact, it is the most oxygenated blood in the fetus (60-70% of blood is saturated), so it is directed to the most important organs in the body! Most of the blood crosses the PFO to the LA, then LV, then into the ascending aorta ending into the head & brain.
Ductus Venuses	It is a communication between the umbilical vein (which carries O2-rich blood from the placenta, O2 saturation ≈70%) across the hepatic area into the IVC	

- These 3 communications close after birth (they disappear), and the circulation divides into 2 separated parts:

1. The Pulmonary circulation: "blue" (deoxygenated or cyanotic) blood going to the lungs.
2. The Systemic circulation: "red" (oxygenated) blood going to the whole body.

***Remember that: CO in the newborn is LV output only & is no longer combined.**



Congenital Heart Disease (CHD)

- Are heart defects that present at birth and cause problems for newborns. The American Heart Association states that there are at least 35 distinct forms of CHD, categorized into either cyanotic or acyanotic lesions.
- Congenital heart defects can be classified into 2 types of lesions:

1. Shunt lesions: presence of a shunt (communication) between pulmonary and systemic circulations (direction of shunt either right to left or left to right). As a result, blood goes to the wrong circulation (the one that it's NOT supposed to go to) and there is mixing of the red and blue blood.
2. Non-shunt lesions: obstructive or regurgitant lesions (not included in the lecture).

Obstructive Lesion	Regurgitant Lesion
Congenital lesions	Acquired lesions
Cause pressure load	Cause volume overload on the heart
Examples: 1. Aortic stenosis (AS)-the most common-	Examples: 1. Aortic regurgitation 2. Mitral regurgitation 3. Tricuspid regurgitation

2. Supravalvar AS 3. Subaortic stenosis 4. Coarctation of Aorta 5. Mitral Stenosis 6. Pulmonary Stenosis	4. Pulmonary regurgitation

- The shunt lesions are divided into 2 major categories:

1. Right to Left shunt or “cyanotic heart disease”.
2. Left to Right shunt or “acyanotic heart disease”.

***NOTE: most patients with cyanotic heart disease have R to L shunt, but they may have a combined shunt (R to L &L to R) with complete mixing of blood in the heart.**

Cyanosis

- Cyanosis is a clinical sign where you see bluish discoloration of the body. It happens when there is clinically significant amount of deoxyhemoglobin.
- In order to see the cyanosis by your eyes there must be 3-4 gm/dL of deoxyHb OR the O2 saturation should be less than 70%.
- On the other hand, desaturation is not a clinical sign; cant be seen by your eyes but by the saturation that you measure, so not everyone who is hypoxic is cyanotic.

Causes of Cyanosis

- In **children** the majority of causes are pulmonary causes:

1. airway disease like pneumonia, asthma or foreign body.
2. Intrapulmonary shunting; the blue blood passes to the red blood area. Cardiac causes are always secondary to intracardiac shunting; the blue blood goes to the red blood area.
3. Other rare causes.

- In a **newborn** infant with respiratory distress (respiratory rate>60 breaths/min) may be due to:

1. cardiac disorders: cyanotic congenital heart disease.
2. Respiratory disorders: e.g. surfactant deficiency, meconium aspiration, pulmonary hypoplasia, etc.
3. Persistent pulmonary hypertension of the newborn (PPHN): failure of the pulmonary vascular resistance to fall after birth.
4. infection: septicemia, group B streptococcus and other organisms.
5. Inborn error of metabolism: metabolic acidosis and shock.
6. Polycythemia.

Mechanism of Cardiac Cyanosis

1. Pure right to left shunt; so a portion of the blue blood goes to the left side of circulation, this what happened in a patient with Tetralogy Of Fallot (TOF).

2. Mixing, which means that the blue blood & red blood are mixed together in a chamber & then it is distributed to the lungs & body.

***Note: the difference between them is that the 1st mechanism has only right to left shunt, while the 2nd one has right to left & left to right shunt.**

3.The Recirculation; the whole red blood will go to the wrong side of the circulation (to the pulmonary artery) & the whole blue blood will go to the body. That happens in transposition; where the left ventricle is pumping the blood into the lungs while the right ventricle is pumping to the body. So the pulmonary arterial saturation is 100%, and the systemic arterial saturation is 50%.

Approach to Cyanotic Baby

- Detailed History: age at presentation can give you a hint about what the pt have, ex. TGA may present in the 1st day of life while TOF appears in the 1st few months of life. Symptoms of presentation that might range from pure cyanosis with no respiratory symptoms to heart failure symptoms.
- physical examination might also help: General Exam, Vital signs, Lung exam, Cardiac exam.
- Testing: many tests can be done to a cyanotic baby: Chest radiography, Electrocardiogram, Hyperoxia test.

Cyanotic Heart Diseases

[5T's with 1-5 mnemonic]

Truncus arteriosus	Vessels join to make **1**
Transposition of great vessels	**2** major vessels switched
Tricuspid atresia	**3** (tricuspid)
Tetralogy of Fallot	**4** defects
Total anomalous pulmonary vascular return	**5** letters (TAPVR)

Truncus Arteriosis

- Is a common trunk arising from both ventricles so it implies that there is VSD.
- There is no truncus without VSD but we don't call it VSD or truncus-VSD. This common trunk is divided into the pulmonary artery & the Aorta, it differs from the others because the red & the blue blood are mixed together. It will be divided into 2 circulations both have the same color; same O2 saturation so it's complete mixing.
- Then the blood will pass according to the resistance; pulmonary vascular resistance (PVR) is very low (large way) BUT the systemic vascular resistance is very high (small way) so more blood will go to the lungs.

- Pts of TA won't present with cyanosis BUT they will present with Heart Failure! **why?**
- [ ] Because there is more blood in the lungs; the ratio between pul. & systemic blood flow is more than (5 : 1) so there will be dilation of the LA & LV & symptoms of Heart failure will appear. But with time they become cyanotic because of PVR rising as in Pulmonary Hypertension.

Physical Exam

- Single S2
- Ejection click of the abnormal truncal valve
- Systolic murmur of truncal valve stenosis if present
- Diastolic murmur of truncal valve insufficiency
- Gallop

Investigations

- **CXR**: Cardiomegaly, increased pulmonary circulation.

Management

- Basically the management is **surgery**, we close the VSD and separate the Aorta and the Pulmonary artery.

Transposition of Great Arteries (TGA)

- The pulmonary artery comes from the LV while the aortic artery from RV, so the blue blood will go to the systemic circulation & the red blood to the pulmonary circulation.
- We should have some mixing in order for pt to survive; because he won't survive more than few minutes if no communication. The communication should be at the atria level (PFO or ASD).
- 50 % of the pts have VSD; usually small.
- Usually TGA babies presents within the 1st day/hrs of life with cyanosis.
- More common in boys.
- pts present with profound cyanosis at birth; we should do number of things medically before sending to surgery; one is to insure that there is mixing at the atrial level & to insure that blue blood is going to lungs so what we do is to keep the PDA open because it will allow the blood to go from aorta to pulmonary artery which is blue as it comes from Aorta that is connected to RV.
- Blue blood to lungs = effective pulmonary circulation, so more blood going to lungs, more blood(saturated) to the LA, so more pressure in the LA than RA pressure, so the ASD will function as the effective systemic opening. Blood will cross from LA to RA which is the red blood that goes to the body. So by opening the PDA you assign it to give you effective pulmonary blood flow and you assign the ASD to deliver oxygenated blood to the body. But if you open the PDA and there's no ASD **nothing will happen**.

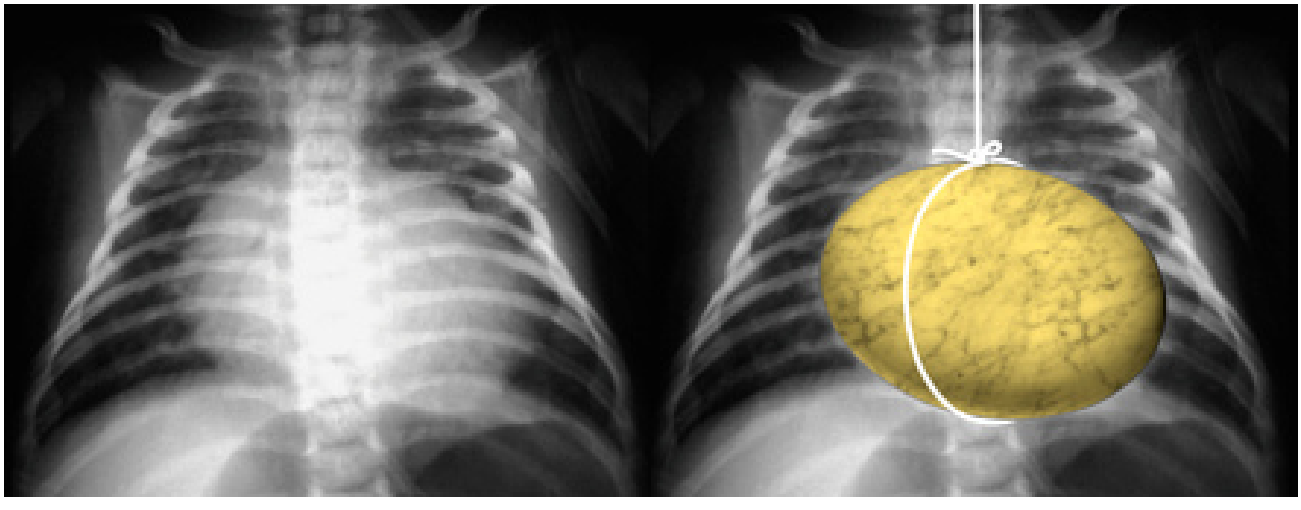
***Note: you will find two types of TGA; d-TGA (d for dextro) which is the one we're discussing here, and l-TGA (l for levo) which is not a cyanotic heart disease, it is a physiologically corrected transposition and it is not our topic. It accounts for about 5-7% of CHDs. On Exam: they are usually healthy, crying and with good birth weight, but cyanosed. There might be loud S2 as the aorta is right under the sternum.**

Investigations

- If left untreated then the CXR finding will be prominent:

1. Egg on string appearance
2. Narrow mediastinum; because the pul. Artery & aorta are front & back NOT side by side.

***Note: you can't find that in newborn period but later on.**



Management

1. You have to keep the PDA open by using PGE-1 infusion, if it doesn't work so you have to consider point 2.
2. Opening up the atrial septum by a balloon (Balloon atrial septostomy).
3. Surgery by switching arteries back where they should be, and moving coronary arteries from pulmonary artery to the new aorta because coronary arteries are always attached to aorta, this is the difficult part.
4. Atrial switch.

Tricuspid Atresia

- Atresia means that the valve didn't form so there is NO communication between RA & RV.
- Requires both ASD & VSD for viability.
- Symptoms will depend on the amount of the Pulmonary Blood Flow:

☐ If the VSD is small there will be cyanosis.
☐ If the VSD is large there will be Heart Failure because more blood will go to the lungs than the body, and the cyanosis will be minimal.

***Note: both Tricuspid Atresia + Truncus Arteriosus will have HF symptoms.**

Tetralogy of Fallot

1. Pulmonary stenosis or right ventricular outflow tract (RVOT)obstruction.
2. VSD.
3. Overriding of the aorta; looks like arising from both RV & LV.
4. RV hypertrophy because VSD is large so the RV has the same pressure as the LV.

***TOF is the most common cyanotic heart disease & the 3rd most common congenital heart disease = 10% of CHD.**

- There is no known etiology, but it is more common in syndromes like 22q 11 (DiGeorge) syndrome and Down syndrome patients who usually have AV canal but may have TOF.
- TOF is seen with malformations: VACTERL, CHARGE, Velo-cardio-facial.
- Decreased pulmonary blood flow, in X-ray you will see oligemic lungs or more black lungs.
- To differentiate between:

☐ **Oligemic lungs**: normal # of ribs + black lung
☐ **Hyperinflated lungs**: more ribs + black lung

- Pulmonary stenosis differs from pt to another & it's progressive with time. At 1 month of age it is moderate, but at 6 months it is sever & more cyanotic with time.
- The flow depends on the difference between systemic & pulmonic outflow resistance.
- Normally, PVR (pulmonary vascular resistance) is 1/10th of the systemic resistance (10 times lower), so in case of large VSD, 10 times the amount of blood will go to the lungs compared to the body. This of course means an overload on the lungs' vascularity.
- Cyanosis will be accompanied by:

☐ Hyperpnea: Increased rate and depth of respirations
☐ Increased fussiness progressing to decreased level of consciousness
☐ Increasing acidosis, can be fatal

- **Theories:**

☐ Primary infundibular spasm
☐ Hyperpnea as a primary cause
☐ Circulating catecholamines

Hypercyanotic Spell

- Mainly here we are talking about SQUATTING; the pt squats (knee-chest position) when he feels cyanotic. It is seen in pts beyond the age of 1 year, they learn how to do this when they have excessive/sever cyanosis which is a life threatening precipitated by activity or fright, BUT it could be spontaneous.
- Hypercyanosis happens because there is more RV outflow obstructions at times when they become profoundly cyanotic, the mechanism is some where around the obstruction; where they have hypovolemia.
- Tachycardia will impair ventricular filling; because it happens at the expense of DIASTOLE not systole so impairment in the RV filling so the obstruction to the RVOT increases (becomes more profound) because of its dynamics just under the valve, just like pts of

Hypertrophic obstructive cardiomyopathy >> they have less volume in the ventricle so the obstruction becomes sever when they stand up the murmur becomes louder because the filling drops. So the same thing with the RV because of depressed/decreased RV filling there will be more obstruction to the pulmonary outflow.

- the blood of right ventricle will cross the VSD to the LV so more Right to left shunt during those times. Cyanosis will increase the agitation, so it becomes like a cycle: the pt either dies or loses consciousness.
- Spells are an indication for need of surgical intervention.

Treatment of Spells

- Reverse elements of the cycle:

- [ ] If cyanosed, give O2 it will improve PO2 in the pulmonary venous blood.
- [ ] knee-chest position, **why?**

1. Because squatting will squeeze the liver so this will increase the venous return, filling the RV, decreasing the obstruction.
2. Squatting kinks the femoral arteries so this increases the resistance in the systemic circulation so blood would rather go to the lungs than the aorta.

- [ ] Give Morphine to calm the pt down.
- [ ] Give fluids to open up RV to fill it.
- [ ] Finally give systemic vasoconstrictor (phenylephrine) which works as the same mechanism of kinking femoral artery; increasing SVR.
- [ ] Beta blockers if surgery is not available to prevent these events.

Symptoms of TOF

- Extreme variability of presentations, Related to the degree of RVOT obstruction
- Consistency of symptoms relates to the degree of shunting between right and left:

- [ ] Cyanosis might be present in the neonatal period, it is always present in patients with PA (pulmonary atresia) or severe obstruction. If you listen to them you might not here a murmur, especially if the PDA is large (the PDA would be the only route of blood to the lungs).
- [ ] Cyanosis might be minimal if the obstruction is mild or not severe
- [ ] Some patients would be asymptomatic but with a murmur. The murmur of TOF can't be missed as the pulmonary valve is right under the sternum and it is a loud murmur.
- [ ] Cyanosis: Typically appears after 1 month as the obstruction becomes more prominent (typically appears between 6wks and 6 months in the unrepaired infant). Parents complain from cyanosis of nail beds and mucous membranes. May be present at rest or only with agitation/exercise.
- [ ] Persistent cyanosis in childhood and clubbing if not repaired.

Investigations

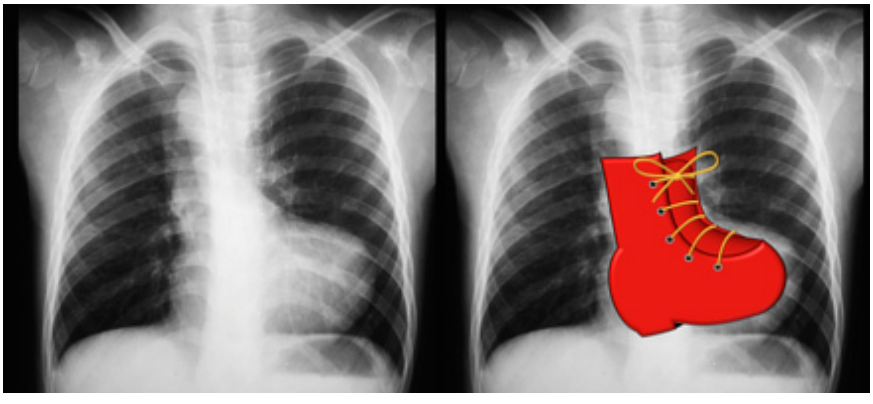
- **CXR >>**

- [ ] normal size BUT abnormal shape (boot-shaped heart)
- [ ] Upturning of the apex

- [ ] Concavity on the area of pulmonary artery, instead of convexity there is concavity because the pulmonary artery is small.
- [ ] Lungs are oligemic.

- **EKG >>**

- [ ] RV Hypertrophy is the characteristic finding.
- [ ] RA Dilatation.
- [ ] Right axis deviation.



*On examination you can hear ejection systolic Murmur because LV & RV both pump to aorta so the heard sound is associated with pulmonary stenosis (inc. intensity, inc. severity of pul.stenosis).

Treatment

- Polycythemia: most pts of TOF are polycythemic as a secondary mechanism to the Hypoxia. Hematocrit will be > 60% so might have symptoms of hyperviscosity syndrome, Consider phlebotomy pre-operatively
- Infection: R→L shunt direct route to body Bacterial, endocarditis and Brain abscess.
- TET spells As mentioned above.
- **Surgical management:**

1. VSD closure.

- [ ] trans atrial access if possible
- [ ] Infundibular resection for visualization
- [ ] Patch closure

2. Alleviation of RVOT obstruction done by surgery:

- [ ] Infundibular resection vs. transannular patch

TOTAL ANOMALOUS PULMONARY VENOUS RETURN (TAPVR)

- It is the rarest cyanotic heart disease. Instead of pulmonary veins coming to LA, they miss the LA but will come to the RA through a communication either through an abnormal vein (vertical vein or directly to RA so the pulmonary vein which has red blood is coming to the SVC then to the RA or to INC to RA so all the blood will be mixed in the RA (complete mixing), the blood can't go to the LV except through ASD or PDA.
- The blood will be distributed to both circulations through ASD & the pt is cyanotic.

- **Types of (TAPVR):**

1. Supracardiac through the SVC
2. Infracardiac through the IVC
3. Mixed type.

*In a radiograph of a Pt with supracardiac TAPVR, you will see a dilated superior vena cava, and it would show a characteristic finding. While in the infracardiac type you would see pulmonary edema without cardiomegaly, unlike in CHF where the heart would be large.