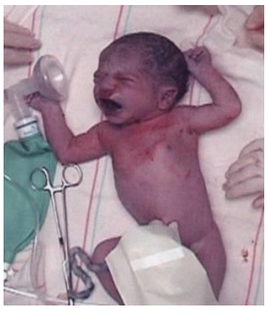
CASE TWO

Short case number: 3_23_02
Category: Children and Young People
Discipline: Paediatrics_Medicine / Surgery
Setting: Urban_Hospital
Topic: Congenital heart disease-cyanotic

CASE



You are the O&G intern called to the labour floor to assist at the LSCS of Harry Ingham, who is a term infant being delivered by LSCS for foetal distress.

On delivery Harry is crying and breathing well, but is cyanosed. His colour does not change with the delivery of oxygen via mask.

The paediatric registrar is concerned that Harry has a heart problem.

QUESTIONS

1. Review the normal foetal circulation and explain the types of problems that may result in a cyanosed infant at birth.
2. Briefly outline the three main subgroups of cyanotic heart disease.
3. Subsequent investigations reveal that Harry has transposition of the great arteries, outline the key features and pathophysiology of this condition.
4. Following Harry's delivery you recall a condition called Tetralogy of Fallot that you covered during one of your PBL cases, you decide to revise these conditions as your next rotation is Paediatrics. Briefly outline the key features and underlying pathophysiology of the following conditions tetralogy of fallot, tricuspid atresia and pulmonary atresia.

Resources

- South M, Isaacs D editors. Practical Paediatrics. 7th edition. Edinburgh: Churchill Livingstone; 2012.

1. **Review normal foetal circulation & what types of issues could result in cyanosed infant at birth:**

Placental blood to foetus through umbilical vein
To foetal liver and to RA
2/3 blood RA to LA via Foramen Ovale
1/3 blood RA > RV > Pulmonary Artery > Ductus Arteriosus > Aorta
(no need to oxygenate blood as has allready been done at placenta)

- RDS - ventilation issue
- Patent Foramen Ovale (ASD - R to L Shunting)
- Patent Ductus Arteriosus (not enough pressure in pulmonary circulation
- Big VSD R to L Shunting
- ToF (R to L shunt)

2. Main Subgroup Cyanotic Heart Disease - Cyanotic (Right to Left) - *Most Start with letter "T"*

- ToF
- Transposition of Great Arteries
- Tricuspid Atreasia

3. Transposition of Great Arteries

Cyanotic Congenital Heart Abnormality
Due to abnormal development of fetal heart in first 8 weeks of pregancy
Aorta Connected to RV and Pulmonary Artery connected to LV
Survival depends on if the pt has a PDA, PFO, VSD - allowing mixing of blood

Infants will only survive a few days to a month - even if they have a communication between blodd supplies

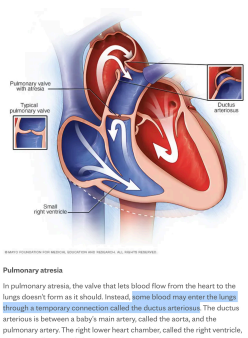
4. Explain ToF, Tricuspid Atresia, Pulmonary Atresia

ToF - 1) Overriding Aorta, 2) VSD, 3) Pulmonary valve stenosis, 4) RV Hypertrophy
Tet Spells (increased intrathoracic pressure from crying, stress worsens the R-L shunt and causes a cyanotic episode)

Tricuspid Atresia - Tricuspid valve completely blocked (only way for blood to get to lungs is via a PFO AND a VSD)

Pulmonary Atresia - Complete Occlusion of POulmonary valve

- Need patent PDA to survive
- Wil require e,ergency surgery & medical management



Pulmonary atresia

In pulmonary atresia, the valve that lets blood flow from the heart to the lungs doesn't form as it should. Instead, some blood may enter the lungs through a temporary connection called the ductus arteriosus. The ductus arteriosus is between a baby's main artery, called the aorta, and the pulmonary artery. The right lower heart chamber, called the right ventricle, may be small in some babies with pulmonary atresia.

ANSWERS

Question 1
Review the normal foetal circulation and explain the types of problems that may result in a cyanosed infant at birth.

Blood from the placenta enter and returns to the foetus through the vessels in the umbilical cord. Blood then goes to the liver and splits into 2 main branches. The blood then reaches the inferior vena cava, a major vein to the heart. Blood enters the right atrium. Most of the blood flows to the left side through the foramen ovale. Blood then passes into the left ventricle and then to the aorta. From the aorta, blood is sent to the head and upper extremities. After circulating there, the blood returns to the right atrium of the heart through the superior vena cava. About one third of the blood entering the right atrium does not flow through the foramen ovale, but, instead, stays in the right side of the heart, eventually flowing into the pulmonary artery.

Because the placenta does the work of exchanging oxygen and carbon dioxide through the mother's circulation, the foetal lungs are not used for breathing. Instead of blood flowing to the lungs to pick up oxygen and then flowing to the rest of the body, the foetal circulation shunts most of the blood away from the lungs. In the foetus, blood is shunted from the pulmonary artery to the aorta through a connecting blood vessel called the ductus arteriosus.

With the first breaths of air the baby takes at birth, the foetal circulation changes. A larger amount of blood is sent to the lungs to pick up oxygen

- Because the ductus arteriosus is no longer needed, it begins to wither and close off. This process is mediated by the hormone, prostaglandin.

- The circulation in the lungs increases and more blood returns to the left atrium of the heart. This increased pressure causes the foramen ovale to close and blood circulates normally

Cyanosis in Tetralogy of Fallot depends on the severity of the pulmonary outflow obstruction. Severe obstruction results in low pulmonary blood flow and important cyanosis while babies with mild obstruction may have minimal cyanosis. Usually clinical cyanosis correlates with at least moderate desaturation and babies with mild desaturation may not look obviously blue. It is important to measure saturations with an oximeter if in doubt.

A harsh ejection systolic murmur is audible at the left sternal edge and/or in the pulmonary area (infundibular stenosis) and radiates through to the back. The second heart sound is often quite loud but single because the pulmonary closure sound is inaudible. When the right ventricular outflow becomes severe, the murmur becomes increasingly high pitched, softer and shorter. During a so-called ‘tet spell’ (also called hypercyanotic or hypoxic spell), the murmur may be very soft for the same reason.

Cyanosis appears gradually during the first 6-12 months of life or rarely later, and is characteristically more obvious on crying or on exertion. A characteristic feature is the development of intermittent episodes of severe cyanosis (spells as above), which may appear spontaneously but are quite commonly precipitated by stress or exercise. Such spells are characterized by marked pallor or cyanosis with dyspnoea and distress. Loss of consciousness may occur. Hypoxic spells are associated with increased right-to-left shunting and a sharp reduction in pulmonary flow.

Cyanosis generally progresses gradually, with diminishing exercise tolerance, finger clubbing and in severe cases growth retardation. Development of cardiac failure is unusual.

Question 2
Briefly outline the three main subgroups of cyanotic heart disease.

Tetralogy of Fallot
Of the four components that comprise Fallot's tetralogy (VSD, pulmonary stenosis, right ventricular hypertrophy, overriding aorta) the important ones are pulmonary stenosis and the VSD.
In Tetralogy, the VSD is always large and severity refers to the degree of pulmonary outflow obstruction. Mild Tetralogy (i.e. with mild RVOTO) may actually have a pure left to right shunt and will function like a simple large VSD (so called “pink Tetralogy”). As the RVOTO gets more severe, the shunt across the VSD becomes less left to right and will then reverse to allow a right to left shunt which will result in systemic desaturation. Progressive RVOTO will result in progressive desaturation/cyanosis. The natural history is for the RVOTO to progress but the time frame may be quite variable. Some babies will be intensely blue in the neonatal period (once the duct starts closing) while reports of asymptomatic tetralogy in adulthood do exist. In general children with this condition are repaired within the first 6-12 months of life.

Transposition of the great arteries
In this condition the aorta and pulmonary arteries arise from the incorrect ventricles. Systemic venous blood is directed through the right side of the heart back into the aorta and pulmonary venous blood through the left side of the heart back into the pulmonary circulation. Survival is dependent on transfer of blood across from each circuit into the other via a foramen ovale, ductus arteriosus or a septal defect. Affected infants generally survive for several days or even weeks because of shunting through the foramen ovale and/or ductus arteriosus, but few live longer than a month without help, unless they have a coexisting septal defect, e.g. a VSD.

Tricuspid atresia
In this malformation the tricuspid valve is blocked completely and there is no communication between the right atrium and ventricle. Systemic venous blood passes via the foramen ovale or an ASD into the left side of the heart, and at ventricular or arterial level a left-to-right shunt exists (via a VSD or PDA). This allows blood to perfuse the pulmonary circulation, usually in reduced amounts.

Question 3
Subsequent investigations reveal that Harry has transposition of the great arteries, outline the key features and pathophysiology of this condition.

Transposition of the great arteries is a congenital heart defect. Due to abnormal development of the foetal heart during the first 8 weeks of pregnancy, the large vessels that take blood away from the heart to the lungs, or to the body, are improperly connected.

In transposition of the great arteries, the aorta is connected to the right ventricle, and the pulmonary artery is connected to the left ventricle. Oxygen-poor blood returns to the right atrium from the body, passes through the right atrium and ventricle, and then goes into the misconnected aorta back to the body. Oxygen-rich blood returns to the left atrium from the lungs, passes through the left atrium and ventricle, and then goes into the pulmonary artery and back to the lungs.

Two separate circuits are formed - one that circulates oxygen-poor blood from the lungs back to the lungs, and another that recirculates oxygen-rich blood from the body back to the body.

Other heart defects are often associated with TGA, and they actually may be necessary in order for an infant with transposition of the great arteries to live. An opening in the atrial or ventricular septum will allow blood from one side to mix with blood from another.

Question 4

Following Harry's delivery you recall a condition called Tetralogy of Fallot that you covered during one of your PBL cases, you decide to revise these conditions as your next rotation is Paediatrics. Briefly outline the key features and underlying pathophysiology of the following conditions tetralogy of fallot, tricuspid atresia and pulmonary atresia.

Tetralogy of Fallot

Tetralogy of Fallot is a complex condition of several congenital defects that occur due to abnormal development of the foetal heart during the first 8 weeks of pregnancy. These problems include the following

- Ventricular septal defect (VSD)
- Pulmonary (or right ventricular outflow tract) obstruction
- Overriding aorta - the aorta is shifted towards the right side of the heart so that it sits over the ventricular septal defect.
- Right ventricular hypertrophy secondary to pulmonary artery obstruction.

Normally, oxygen-poor blood returns to the right atrium from the body, travels to the right ventricle, and then is pumped through the pulmonary artery into the lungs where it receives oxygen. Oxygen-rich blood returns to the left atrium from the lungs, passes into the left ventricle, and then is pumped through the aorta out to the body.

In tetralogy of Fallot, blood flow within the heart varies, and is dependent on the severity of the RVOTO, the VSD always being large.

- With mild right ventricle obstruction, the pressure in the right ventricle can be slightly higher than the left. Some of the oxygen-poor blood in the right ventricle will pass through the VSD to the left ventricle, mix with the oxygen-rich blood there, and then flow into the

aorta. The rest of the oxygen-poor blood will go its normal route to the lungs. These children may have slightly lower oxygen levels than usual, but may not appear blue.

- With more serious obstruction in the right ventricle, it is harder for oxygen-poor blood to flow into the pulmonary artery, so more of it passes through the VSD into the left ventricle, mixing with oxygen-rich blood, and then moving on out to the body. These children will have lower than normal oxygen levels in the bloodstream, and may appear blue. (Because the VSD is large, the RV pressure is actually similar to that on the left side. The resistance to pulmonary artery flow produced by the severe RVOTO is the important factor.)

Tetralogy of Fallot occurs in about two out of every 10,000 live births. It makes up about 8 percent of all cases of congenital heart disease. Tetralogy of Fallot occurs equally in boys and in girls.

Pulmonary Stenosis (PS) PS is either isolated or associated with other cardiac anomalies. Isolated lesions may be range from trivial to severe. Severe lesions will produce RVH and there may be some infundibular narrowing associated. Most children with PS are asymptomatic but important obstruction will be treated with balloon dilatation or failing that surgery.

PS can be due to isolated valvular (90%), subvalvular, or peripheral (supravalvular) obstruction, or it may be found in association with more complicated congenital heart disorders.

With severe valvular PS, right ventricular hypertrophy can cause infundibular narrowing and contribute to the right ventricular outflow obstruction. With severe PS and decreased right ventricular chamber compliance, cyanosis can occur from right-to-left shunting if a concomitant patent foramen ovale, atrial septal defect, or ventricular septal defect is present.