Case Number 13
Transposition of the Great Arteries, Atrial Septal Defect & Ventricular Septal Defect

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Case Summary:

Demographic Details:
Ms. BB, 3-day-old female, Żebbug.

Three-day-old baby girl, transferred to NPICU because of low saturations and a development of a murmur. Following a number of rigourous test and the appropriate investigations, she was diagnosed with Transposition of the Great Arteries (TGA), a large Atrial Septal Defect, and a malaligned Ventricular Septal Defect. She was then transferred to Great Ormond Street Hospital (GOSH) for corrective surgey.

Presenting complaint:

39 week gestation baby girl was transferred to NPICU on third day of life because she became increasingly cyanotic and developed a murmur.

History of presenting complaint:

Baby BB was born to a 34-year-old, healthy, primigravida mother, blood group A positive. There were no significant problems during the pregnancy, antenatal scans were normal and birth was uneventful. The time from the rupture of membranes until delivery was 18 hours.

Baby BB was born at 39 weeks of gestation by normal vaginal delivery and weighed 2.97kg. Apgar scores her both 9 at 1 and 5 mins respectively. Examination at NPICU revealed that the baby was tachypnoeic, centrally cyanosed (with dusky lips) and jaundiced.

Past medical and surgical history:

Baby B.B was previously healthy. Her medical and surgical history are unremarkable.

Drug History:

During Pregnancy:

Drug	Dosage	Frequency	Reason
Folic Acid	400mcg	daily	To prevent Neural Tube Defects in the growing embryo
Iron Supplements		daily	To prevent iron deficiency anaemia

During Labour:

Drug	Dosage	Frequency	Reason
Entonox		prn	Pain relief
Pethidine	100mg	once	Pain relief
Maxalon	10mg	once	Anti-emetic

Family history:

Baby BB was born to healthy non-consanguineous parents, who have no relevant family history of medical problems. Maternal grandmother suffers from Diabetes Mellitus and Hypertension. Paternal grandmother died from myocardial infarction at age 64.

Current therapy:

On admission to NPICU, baby BB was on no medication.

Examination:

Baby BB was well perfused, with a capillary-refill time of 2 seconds and warm peripheries. She was slightly jaundiced and had no dysmorphic features.

Weight: 2.73kg
Saturations: 89-90% in air.
Temperature: 36.7^{o}C
Pulse: 100bpm

Respiratory rate: 32 breaths per minute.

Cardiovascular system: S1 + S2 + 2/6 ejection systolic murmur radiating to the back.

Chest: clear, with good air entry bilaterally.

Abdomen: soft, non-tender, no organomegaly and no palpable masses.

Neurological examination: baby was active, had a soft fontanelle and both femorals were palpable.

Blood pressure:
- Right upper limb: 88/41mmHg
- Right lower limb: 85/39mmHg
- Left upper limb: 117/89mmHg
- Left lower limb: 93/78mmHg

Discussion of results and general and systemic examinations:

From the above findings, it can be concluded that the baby had relatively low saturations in air, which explains why she was tachypnoeic, had dusky lips and was therefore centrally cyanosed. The low saturations, which did not respond to oxygenation, were most probably related a cardiac condition, as was the murmur which was heard on auscultation. She does not have signs of organomegaly and therefore she was not in heart failure. Since there are no differences between the blood pressure of her right upper limb and the other blood pressure readings, it is unlikely that she was suffering from coarctation of the aorta.

Differential Diagnosis:

- Congenital Heart Disease (involving mainly right to left shunts)
  -Transposition of the Great Arteries
  -Right Obstruction: Pulmonary Stenosis, Tetralogy of Fallot
  -Left Obstruction: Aortic Stenosis, Coarctation of the aorta, Hypoplastic left heart syndrome

- Sepsis
- Respiratory Distress
- Methaemoglobinaemia

Diagnostic Procedures:

Laboratory exams:

Test: Arterial Blood Gases
Justification for test: Due to cyanosis
Result: pH:7.40, pCO_2: 30mmHg, pO_2: 40mmHg, HCO_3:20.1; Base Excess: -4.6; Saturations: 76%.
Conclusion: Hypoxaemia.

Test: Complete Blood Count
Justification for test: As a baseline and moreover, to assess if patient has polycythaemia (which would also present with cyanosis) or has an infection (to exclude sepsis).
Result: Normal
Conclusion: The patient does not have polycythaemia and does not have increased white cells, which would indicate a possible infection.

Test: CRP, Blood Cultures
Justification for test: To assess if there is an inflammatory or infectious process occuring and therefore exclude sepsis.
Result: Normal
Conclusion: The patient does not have a inflammatory or infectious process and therefore sepsis is unlikely.

Test: Liver Function Tests especially Bilirubin (both total bilirubin and conjugated bilirubin)
Justification for test: Jaundiced patient
Result: Bilirubin: 271 (Normal: <230)
Conclusion: Patient was started on phototherapy.

Other Investigations:

Test: Chest X- Ray
Justification for test: To assess the lung fields for potential pneumonia or other inflammatory process, due to cyanosis.
Result: Clear lung fields, no consolidation or white shadowing.
Conclusion: Pneumonia or other infectious process unlikely.

Test: Echocardiogram
Justification for test: Possible congenital heart disease.
Result:
- Situs Solitus
- Transposition of the Great Arteries (TGA)
- Large Atrial Septal Defect (ASD)
- Malaligned over riding Ventricular septal defect (VSD)
- Turbulence in main pulmonary artery but the valve appears normal with velocities less than 2m/s

Conclusion: The patient was diagnosed with TGA and VSD; there was sufficient mixing of systemic and pulmonary circulations, through the VSD, and therefore did not require a Rashkind procedure (septostomy).

Therapy:

Drug Therapy at NPICU:

Drug	Dosage	Frequency	Type	Reason
Co-amoxiclav	90mg	Bd	Broad spectrum antibiotic	Against Listeria monocytogenes
Cefotaxime	150mg	Bd	Third Generation Cephalosporin	Against Group B streptococcus

Surgical Therapy:

The patient was transferred to Great Ormond Street Hospital – United Kingdom, were she had her TGA repaired and her VSD and ASD patched at 7 weeks of age.

Pre- operative: The patient was transferred to Great Ormond Street Hospital, London.

Operation:

- Median sternotomy, thymectomy, pericardial patch taken, atrio-aortic CPB 28C, left atrial vent.
- Duct ligated, divided and oversewn.
- Pulmonary arteries mobilised and controlled with sialastic loops, AXC, cold blood cardioplegia via isolated aortic root and then via coronary ostia.
- Cavae snared, right atriotomy.
- Ventricular septal defect closed with bovine pericardial patch and continuous 7-0 prolene, with one additional reinforcing pledgetted interrupted suture.
- Aorta transpected and coronary buttons mobilised.
- Autologous pericardial patch reconstruction of neo-pulmonary arterry with continuous 7-0 prolene.
- Coronary buttons relocated to medially hinged trapdoor flaps in neo-aorta with continuous 7-0 prolene.
- Lecompte maneouvre and neo-aortic anastomosis completed with continuous 7-0 prolene.
- Atrial communication closed with 6-0 prolene.
- AXC off following de-airing, heart regaining sinus rhythm.
- Neo-pulmonary artery completed with continuous 7-0 prolene, atriotomy closed in two layers with continuous 7-0 prolene.
- Off cardiopulmonary bypass easily in sinus rhythm on 0.05adr and 0.5 milrinone once warm.
- Modified ultrafiltration, prolonged haemostasis, x2 Right Atrium and x1 right ventricle pacing wires, mediastinal drain, x1 peritoneal dialysis catheter, vicryl to sternum, layered closure.

Post operative: Chest drains were removed on day 2 and the patient was extubated on day 3. Feeds re-established post operation and she was discharged on demand feeds. She was noted to be arching her back after feeds and occasional vomiting and so was started on anti-reflux medications.

Post operative medications were as follows:

Drug	Dosage	Frequency	Reason
Amiloride	0.6mg	BD	Diuretic
Furosemide	3mg	BD	Diuretic
Domperidone	0.9mg	QDS	Stomach/Reflux
Ranitidine	6mg	TDS	Stomach/Reflux

Diagnosis:

Ms. BB, was diagnosed with a congenital heart disease – namely a Transposition of the Great Arteries, a malaligned VSD and a large ASD.

Transposition of the Great artery is typically characterised by atrioventricular concordance and ventriculoarterial discordance (VA). In this type of malformation, the right atrium and the right ventricle are morphologically connected, giving rise to the aorta, whilst the left atrium and left ventricle are also morphologically connected, and these give rise to the pulmonary trunk. As a result, the systemic circulation is separate from the pulmonary circulation and therefore deoxygenated and oxygenated bloods do not mix.

The incidence of transposition of the great arteries is typically 1 in every 3500-5000 births, and it is typically commoner in males, with a male:female ratio of 1.5-3.2:1. Ventriculoarterial discordance is usually an isolated finding. This malformation can also be associated with other cardiac malformations, such as a VSD and left ventricular outflow obstruction. In these patients, the onset and presentation is typically delayed. If TGA is present with a VSD, but not left ventricular obstruction, cyanosis may be present only during exertion of the child; such as during crying or feeding. In such patients, heart failure is the prevalent feature.

Even though the aetiology of this congenital condition has been associated with some risk factors, such as exposure to rodenticides and antiepiletic medication, as well as a number of mutations such as growth differentiation factor-I, the exact aetiology remains unknown.

Diagnosis is made by echocardiography and further treatment and management is based on the findings at echocardiography. Typically, in isolated TGA, palliative therapy with prostaglandins and septostomy is required in order to ensure adequate mixing of blood, before corrective surgery. However, in this particular case, palliative therapy was not necessary, because there was sufficient mixing of blood through the VSD.

The type of corrective surgery performed was an arterial switch operation. In this procedure, the aorta and pulmonary trunks are sectioned, transposed and anastomosed. The coronary arteries, are then translocated to the neo-aorta.

Alternatively, a REV procedure (repair without extracardiac conduit) or its modification, and the Rastelli procedure can be used. Both these procedures involve creating an interventricular tunnel which connects the left ventricle to the aorta. In the Rastelli procedure, a patch is placed to create an interventricular tunnel and an extra cardiac conduit is placed between the right ventricle and the pulmonary arteries, whereas in the REV procedure the muscular outlet septum resected and the Lecompte manoeurve which avoids the use of the extracardiac conduit.

Final Treatment and Follow up:

Once the patient returned back to Malta, she underwent a follow-up echocardiogram. This demonstrated a patched VSD and no ASD. Furthermore, it also showed good function and suitable outflows with no significant obsturction. There were no effusions. Another follow-up echocardiogram was scheduled for a month later.

Fact Box 13:

Title: Transposition of the Great Arteries, Ventricular Septal Defect, Atrial Septal Defect

Description: Transposition of the Great artery is typically characterised by atrioventricular concordance and ventriculoarterial discordance (VA). The right atrium and the right ventricle are morphologically connected, giving rise to the aorta. The left atrium and left ventricle are also morphologically connected, and these give rise to the pulmonary trunk. As a result, the systemic circulation is separate from the pulmonary circulation and therefore deoxygenated and oxygenated bloods do not mix. This malformation can be assosicated with other lesions such as a Ventricular Septal Defect and Atrial Septal Defects (as in this case).

Risk Factors:

- Idiopathic
- Inutero exposure to rodenticides and anti-eplieptic treatment

Signs and Symptoms:

- The typical presentation is cyanosis in a newborn, with low saturations and hypoxaemia. Presentation, depends on the malformation present.
- Typically with an isolated TGA, cyanosis is present soon after birth. If the malformation is a TGA and VSD, the cyanosis is typically delayed until a few days after birth. With a TGA and VSD but no left ventricular obstruction, cyanosis is typically present only after exertion. These patients will develop heart failure earlier on in their life.

Diagnosis: Established by echocardiogram.

Management: According to findings at echocardiogram.

- In isolated TGA: Palliative treatment by septostomy and prostaglandins is required to ensure adequate mixing of blood, until corrective surgery is performed.
- In TGA and VSD: Palliative treatment is not usually required as there is adequate mixing of blood through the Ventricular Septal Defect.

Corrective Surgery: This is typically achieved by an arterial switch operation. Briefly, the aorta and pulmonary trunks are sectioned, transposed and anastomosed. The coronary arteries, are then translocated to the neo-aorta.

Prognosis: Prognosis is very good following corrective surgery, with more than 90% survival after 5 years.

References:

1. http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0002535/ as accessed on 9/02/2013
2. http://circ.ahajournals.org/content/114/24/2699.short as accessed as accessed on 9/02/2013
3. http://www.biomedcentral.com/content/pdf/1750-1172-3-27.pdf as accessed on 09/02/2013
4. http://www.nejm.org/doi/pdf/10.1056/NEJM196010132631506 as accessed on 09/02/2013
5. http://www.ncbi.nlm.nih.gov/pubmed/3747568 as accessed on 09/02/2013
6. http://circ.ahajournals.org/content/104/suppl_1/I-121.full as accessed on 09/02/2013