Case number 11
The Complex Heart

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Case summary:

WB, a four-month-old Caucasian baby boy from Mosta was admitted to the paediatric ward in Mater Dei following an incidental finding of dextrocardia, failure to thrive and developing signs of respiratory distress, all indicating possible chronic heart failure.

Following examination and testing, baby WB was found to have a double outlet right ventricle, transposition of the great vessels, a VSD with blood mixing, and an element of heart failure. WB is termed to be having a complex heart. The patient is currently not fit for operation and is being managed on feeds to facilitate his growth.

Presenting complaint:

WB had incidental findings of dextrocardia, failure to thrive and signs of respiratory distress, raising concerns about chronic heart failure.

History of presenting complaint:

WB appeared well at home, with no obvious symptoms of chronic heart failure, no cyanosis, no chest infections, afebrile and seemingly feeling well. His referral was following an incidental finding during routine immunisation and was not prompted by any obvious signs noted by the parents.

Past medical and surgical history:

Past medical history:

Gestation & Birth:

- Normal gestation
- Twelve-hour labour
- Delivered by emergency caesarean section
- No perinatal complications
- No referrals to NPICU
- Birthweight: 3.89 kg
- Other: nil of note

Past surgical history:

Nil of note

Drug history:

Nil of note

Family history:

WB is the second child of a 38-year-old mother, and a 36-year-old father, who also have an 8-year-old son.

WB's mother is allergic to penicillin, but other than this, there is no history of ill family members.

Social history:

WB is the second child in the family, and lives in a supporting household.

Systemic inquiry:

- General Health: Generally looks well and alert but is pale and mildly cyanosed.
- Cardiovascular System: Tachycardic (heart rate of 120 bpm), well perfused, capillary refill under 2 seconds, cyanotic tinge around the mouth, apex beat felt on the right with heart sounds loudest in the right side of the chest, heaves felt in right 4-5 intercostal space veering to the right anterior axillary line and also felt along the right parasternum, and no murmurs, loud S2 with some splitting.
- Respiratory System: Tachypnoeic (respiratory rate 40/min), SpO_2 75% on air, significant subcostal recessions, indrawing of intercostal muscles, tracheal tug, clear chest sounds.
- Gastrointestinal System: Currently feeding, liver edge palpable on the left side extending 1.5 cm, abdomen is soft and non-tender.
- Genitourinary System: Nil of note.
- Central Nervous System: Nil of note.
- Musculoskeletal System: Reduced subcutaneous fat.
- Endocrine System: Nil of note.
- Others: Nil of note.

Current Therapy:

Cannula for venous access inserted and a CXR and echocardiogram ordered.

Discussion of results of general and specific examinations:

During the general examination it was evident that the baby was in distress and cyanosed. The apex beat was remarkably strong on palpation and the second heart sound was very loud because the aortic valve lies very superficial and close to the chest wall.

Diagnostic procedures:

Laboratory exams:

Test: Complete Blood Count.
Justification for test: Check haemoglobin level because of cyanosis.
Result: Patient had an increased red cell count, increased haemoglobin, a high haematocrit and increased red cell distribution width (RDW).
Conclusion: Shows attempted compensation for hypoxia by increasing the oxygen carrying capacity of the blood.

Test: Renal Profile (Serum).
Justification for test: Check that there is adequate renal function due to new cyanosis and heart failure.

Result: Patient had a low Creatinine and a high potassium.
Conclusion: Low Creatinine indicates that the kidneys are functioning normally while the high potassium is due to the potassium sparing diuretics.

Test: Calcium and Phosphate (Serum).
Justification for test: Check to see if electrolytes are normal because $CaPO_4$ tends to get reduced in children in heart failure and on diuretics.
Result: Patient had high calcium and a high phosphate.
Conclusion: Patient not having $CaPO_4$ lowering side effects from treatment.

Instrumental exams:

Test: Chest X-ray.
Justification for test: To confirm dextrocardia and check for situs inversus or radiological signs of heart failure.
Result: There is dextrocardia and total viscerus inversus. The liver is left-sided and the spleen is right-sided.
Conclusion: Diagnosis confirmed and mild signs of heart failure seen.

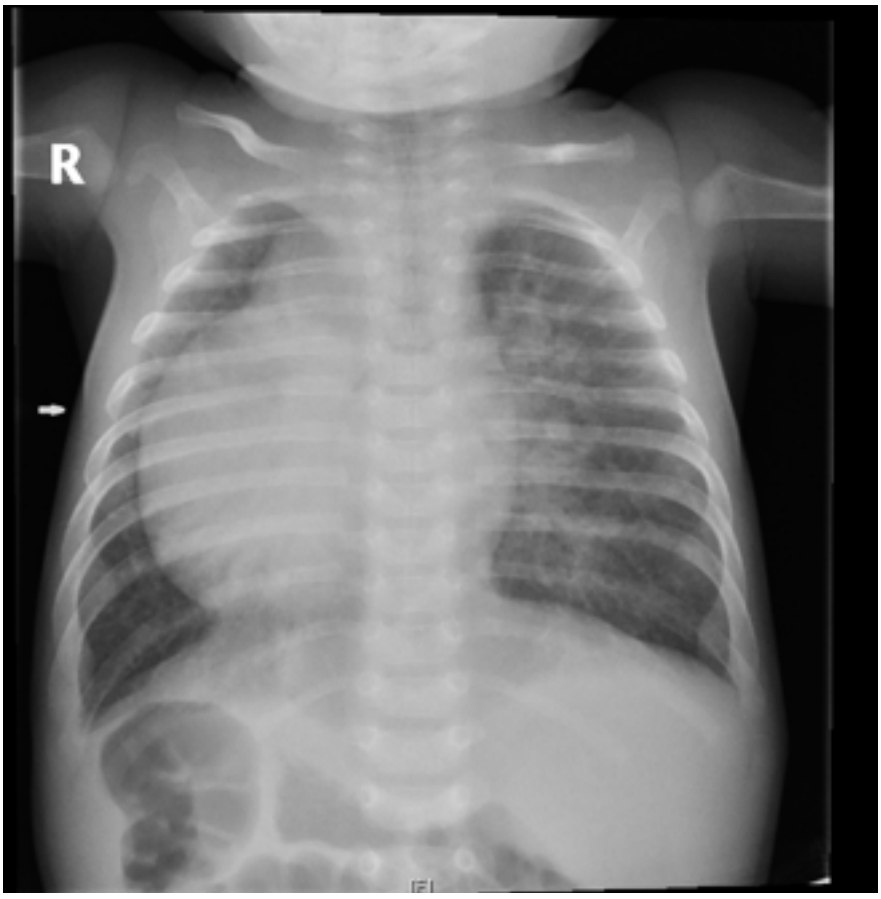

Figure 1 Chest X-ray showing dextrocardia.

Test: Abdominal Ultrasound.
Justification for test: To visualise all abdominal organs and their anatomy since situs inversus was found on chest X-ray.
Result: There is total viscerus inversus. The liver is left-sided. The spleen is right-sided. The liver, spleen, pancreas and kidneys are of normal size and structure. The gall bladder is contracted. The bladder is empty. No ascites or pleural effusions.

Conclusion: Situs inversus is present but all abdominal organs are still normal in size and structure although they are mirror images.

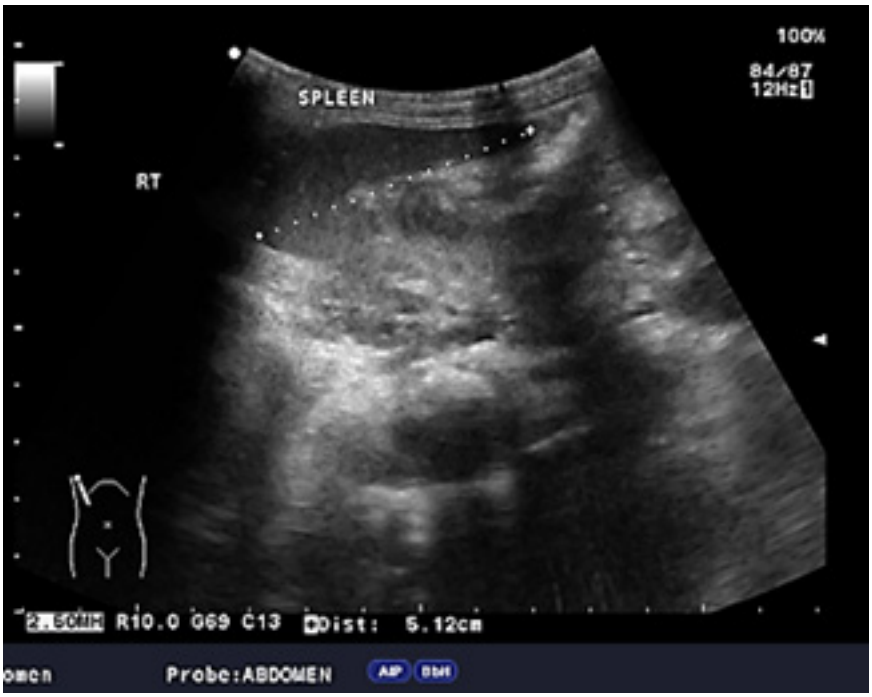

Figure 2: Ultrasound scan showing spleen on the right side.

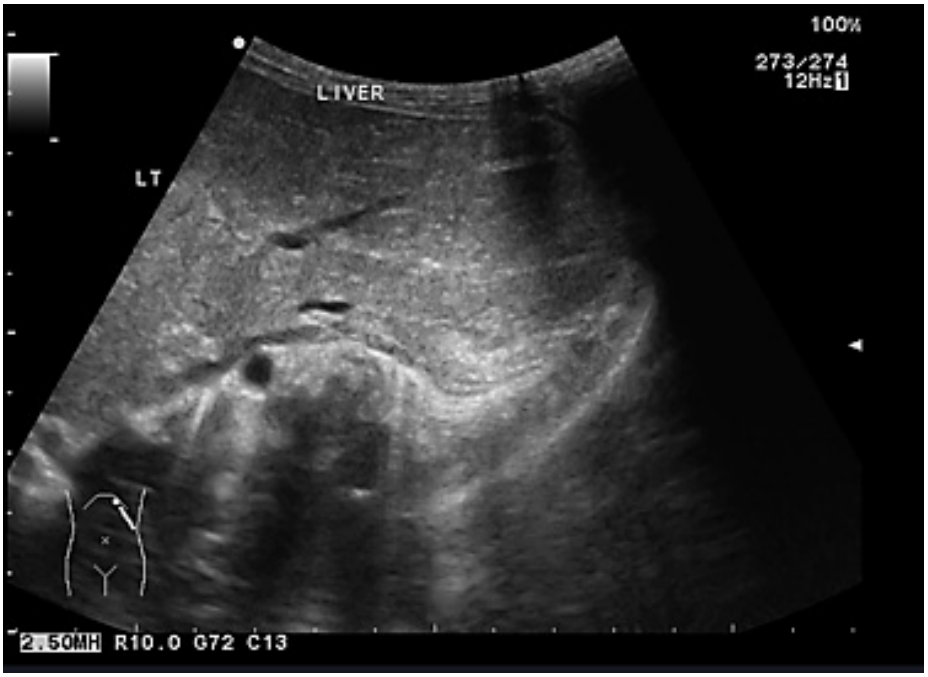

Figure 3: Ultrasound scan showing liver located on the left

Test: Echocardiogram.
Justification for test: To screen for congenital cardiac defects since patient has situs inversus and has a cyanotic tinge.
Result: There is dextrocardia and abdominal situs inversus. Inferior vena cava on the left and ductus arterious on the right. There is a very large right atrium and a smaller left atrium, inter-atrial septum is intact except for a small patent foramen ovale.
The right ventricle is enlarged and hypertrophied with multiple muscle bands and gives off both great arteries. The aorta is smaller and is centrally placed overriding a ventricular septal defect (VSD). The

pulmonary artery is much larger and is anterior and to the right of the aorta and can be seen to bifurcate. The aorta turns directly backwards and narrows down a bit where it joints the ductus arteriosus but there was no turbulence on Doppler.

The VSD extends from the inlet septum where it is relatively small to the outlet septum where it is 1.3cms across. There is another small VSD closer to the apex. There is moderate atrioventricular valve regurgitation. The pulmonary blood flow is unrestricted.
Conclusion: There is Dextrocardia, DORV (double-outlet right ventricle), a large VSD and unrestricted pulmonary blood flow present.

Therapy:

Drugs:

Drug	Dosage	Frequency	Type	Reason
Spirinolactone	4.5mg	BD	Aldosterone antagonist Diuretic	Reduce fluid load and prevent congestive heart failure
Furosemide	4.5mg	BD	Loop Diuretic	Reduce fluid load and prevent congestive heart failure
Duocal			Feeds	Increase calorie intake for weight gain as the patient is currently failing to thrive due to the excess energy consumption from his abnormal circulation

Diagnosis:

Dextrocardia, Double-outlet right ventricle, Ventricular septal defects and Transposition of the great vessels.

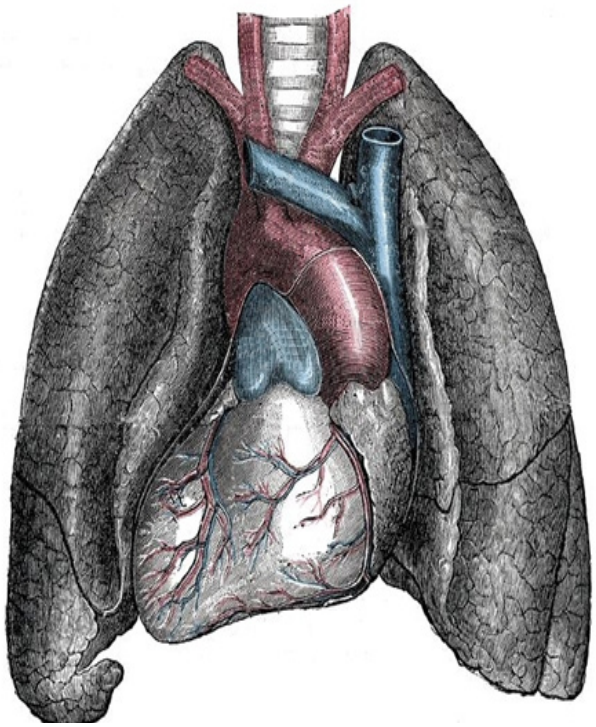

Figure 4: Diagram showing dextrocardia and transposition of the great vessels.

Final treatment and follow-ups:

The aim of treatment is to correct his cardiac defect by closing the VSD and exchanging the arteries with each other so as to make the heart function efficiently. However the patient cannot undergo this surgery as of yet because his weight is still too low at 3.8kgs. He needs to be at least 5kgs.

When the necessary weight is achieved he will be sent abroad to a specialised hospital where he will undergo a corrective procedure. However, prior to this the patient may need pulmonary artery banding to reduce excessive pulmonary blood flow and improve his heart failure whilst decreasing his failure to thrive.

Fact Box 11- Part 1

Title: Situs Inversus

Definition: Situs inversus (also called situs transversus or oppositus) is a congenital condition in which the major visceral organs are mirrored from their normal positions. The normal arrangement being known as situs solitus.

Effect on Anatomy: The condition affects all major structures within the thorax and abdomen. Generally, the organs are simply transposed through the sagittal plane. The heart is located on the right side of the thorax, the stomach and spleen on the right side of the abdomen and the liver and gall bladder on the left side. The left lung is trilobed and the right lung bilobed and blood vessels, nerves, lymphatics and the intestines are also transposed.

If the heart is swapped to the right side of the thorax, it is known as situs inversus with dextrocardia or situs inversus totalis. If the heart remains on the normal left side of the thorax, a much rarer condition (1 in 22,000 of the general population), it is known as situs inversus with levocardia or situs inversus incompletus. This case is thus a situs inversus with dextrocardia.

Prevalence: Situs inversus is thought to be present in 0.01% of the population.

Genetics: It is an autosomal recessive condition but is also X- linked in some cases.

Significance:

- In the absence of congenital heart defects, individuals with situs inversus are phenotypically normal, and can lead normal healthy lives, without any complications related to their medical condition.
- But there is a 5 –10% prevalence of congenital heart disease in individuals with situs inversus totalis, most commonly transposition of the great vessels, as in this case.
- The incidence of congenital heart disease is 95% in situs inversus with levocardia.

Diagnosis:

- Many people with situs inversus totalis are unaware of their unusual anatomy until they seek medical attention for an unrelated condition. The reversal of the organs may then lead to some confusion, as many signs and symptoms will be on the atypical side.
- For example, if an individual with situs inversus develops appendicitis, they will present to the physician with lower left abdominal pain, since that is where their appendix is located. Thus, in the event of a medical problem, the knowledge that the individual has situs inversus can expedite diagnosis especially if the person is unable to communicate.

Advice: Patients with this rare condition may inform their physicians before an examination, so the physician can redirect their search for heart sounds and other signs. Wearing a medical identification tag can help to inform health care providers in the event of the person being unable to communicate.

It is advisable to screen for Kartagener's Syndrome since about 25% of individuals with situs inversus have an underlying condition known as primary ciliary dyskinesia (PCD). PCD is a dysfunction of the cilia that manifests itself during the embryologic phase of development. Normally functioning cilia determine the position of the internal organs during early embryological development, and so individuals with PCD have a 50% chance of developing situs inversus. If they do, they are said to have Kartagener syndrome, characterised by the triad of situs inversus, chronic sinusitis and bronchiectasis. Cilia are also

responsible for clearing mucus from the lung and thus the dysfunction causes increased susceptibility to lung infections.

Other complications: Situs inversus also complicates organ transplantation operations as donor organs will more likely come from situs solitus (normal) donors. As hearts and livers are chiral, geometric problems arise when placing an organ into a cavity shaped in the mirror image. For example, a person with situs inversus who requires a heart transplant needs all the vessels from the transplant donor heart reattached to their existing ones. However, the orientation of these vessels in a person with situs inversus is reversed, thus alterations have to be made.

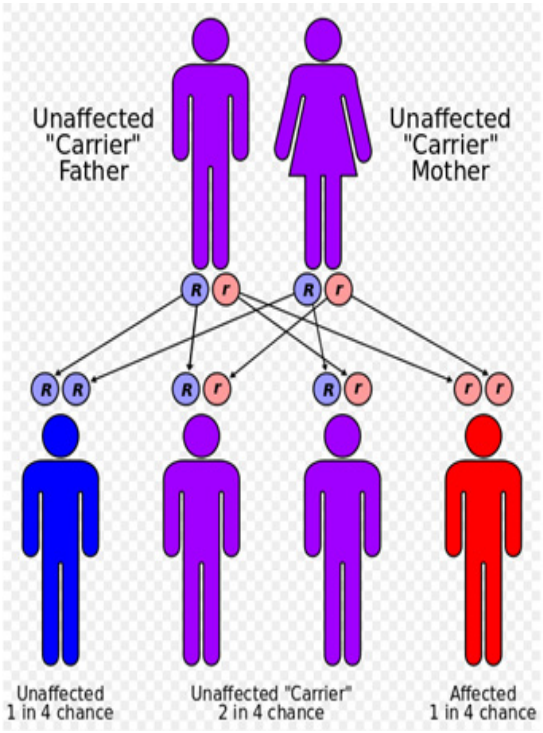

Figure 5: Diagram showing the autosomal pattern of transmission of dextrocardia.

Fact Box 11 - Part 2

Title: Transposition of the great vessels (TGV)

Definition: TGV is a group of congenital heart defects involving an abnormal spatial arrangement of any of the great vessels: superior and/or inferior venae cavae, pulmonary artery, pulmonary veins and aorta. Transposed vessels can present a large variety of atriovenous, ventriculoarterial and/or arteriovenous discordance. The effects may range from a change in blood pressure to an interruption in circulation, depending on the nature and degree of the misplacement and which vessels are involved. Although "transposed" literally means "swapped", many types of TGV involve vessels that are in abnormal positions, while not actually being swapped with each other.

Variants:

- Transposition of the great arteries, a condition where the congenital heart defect involves only the primary arteries, that is the pulmonary artery and aorta.
- Dextro-Transposition of the great arteries.
- In dextro-transposition of the great arteries (dextro-TGA) deoxygenated blood from the right heart is pumped immediately through the aorta and circulated to the body and the heart itself, bypassing the lungs altogether, while the left heart pumps oxygenated blood continuously back into the lungs

through the pulmonary artery. In effect, two separate "circular" (parallel) circulatory systems are created. It is called a cyanotic congenital heart defect (CHD) because the newborn infant turns blue from lack of oxygen.

- Levo-Transposition of the great arteries.
- Levo-Transposition of the great arteries is an acyanotic heart defect in which the primary arteries are transposed, with the aorta anterior and to the left of the pulmonary artery, and the morphological left and right ventricles are also transposed.

Difference between simple and complex TGV: In many cases, TGV is accompanied by other heart defects, the most common type being intracardiac shunts such as atrial septal defects including patent foramen ovale, ventricular septal defect, and patent ductus arteriosus. Stenosis or other defects of valves and/or vessels may also be present.
When no other heart defects are present it is called 'simple' TGV; when other defects are present it is called 'complex' TGV.

Treatment: For newborns with transposition, prostaglandins can be given to keep the ductus arteriosus open which allows mixing of the otherwise isolated pulmonary and systemic circuits. Thus oxygenated blood that recirculates back to the lungs can mix with blood that circulates throughout the body. This was not needed in this case because blood was mixing due to the VSDs present. Surgical correction is the definitive treatment for a transposition.

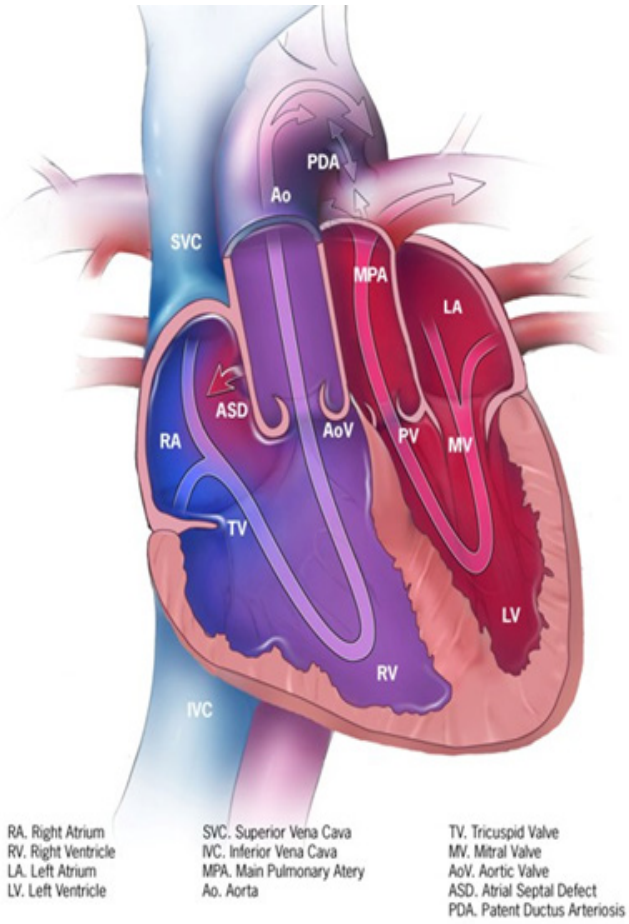

Figure 6: Diagram showing transposition of the great vessels.

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