CASE ONE

Short case number: 3_23_01
Category: Children and young people
Discipline: Paediatrics_Medicine
Setting: Urban_hospital
Topic: Congenital Heart disease_acyanotic

CASE

Finley Mae, is 6 weeks old, she is presenting with her mother to newborn follow-up clinic. You are the paediatric intern in the clinic today.
Finley's mother informs you that she has been feeding and sleeping well, but she is concerned that occasionally Finley appears short of breath when breast feeding.
You note that Finley is alert and pink and there are no features of respiratory distress.
On auscultation you detect a high pitched pansystolic cardiac murmur along the left sternal edge.

QUESTIONS

1. What further history would you explore with Finley's mother and why?
2. What clinical findings on history and examination would suggest the presence of cardiac failure?
3. Finley has no clinical features of cardiac failure, but you are concerned that Finley may have a ventricular septal defect [VSD]. With the assistance of diagrams explain to Finley's mother what a VSD is how they may develop.
4. You speak with the Paediatrician in the clinic who suggests a cardiology consult. Outline the details that you would include on the referral letter.
5. The cardiology registrar explains that a chest x-ray and ECG should be organised before Finley attends the paediatric cardiology clinic. What information can be obtained from these investigations?
6. You decide to attend Finley's appointment at the cardiology clinic. The chest x-ray and ECG have been reported as normal. The murmur is still present and Finley undergoes an echocardiogram which reveals a moderate VSD. The cardiologist explains the normal progress of VSD to Finley's parents. Outline what the cardiologist would have explained to her parents and details the complications that may occur with larger ventricular septal defects.
7. Following the clinic, you decide to revise acyanotic heart disease, briefly outline the pathophysiology of the following conditions, persistent ductus arteriosis, atrial septal defect, pulmonary & aortic stenosis and coarctation of the aorta.

Resources

- Cheung M, Heart disease, Chapter 15.2 in South M, Isaacs D. Practical Paediatrics 7th Ed. Churchill Livingston, Edinburgh 2012.
- Menahem S. Recognising Heart Disease in Childhood. Australian Doctor, How to Treat. 5th December 2008. http://search.ebscohost.com.ipacez.nd.edu.au/login.aspx?direct=true&db=anh&AN=35778757&site=ehost-live&scope=site

1. What further history would you explore with Finley's mother and why?

- When else does this occur
- Pregnancy Hx
- Developmental milestones
- Colour at birth (born blue (TOF?)
- Breathing difficulties
- Sleep
- **All the ususal paeds stuff*

2. Clinical findings that would suggest cardiac failure in child (5)

- Tachycardia, Sweating
- Dyspnoea/Tachypnoea -> Feeding Difficulties -> Failure to Thrive
- Increased WoB (Intercostal recession)
- Periorbital Edema (other edema hard to assess on child)
- Liver Enlargement (due to systemic venous congestion)

Finley has no clinical features of cardiac failure, but you are concerned that Finley may have a ventricular septal defect [VSD]. With the assistance of diagrams explain to Finley's mother what a VSD is how they may develop.

Hole in heart (develop due to incomplete formation in embryological development)
L to R Shunt

The cardiology registrar explains that a chest x-ray and ECG should be organised before Finley attends the paediatric cardiology clinic. What information can be obtained from these investigations?

Small VSD - CXR & ECG normal

For larger defects - CXR shows cardiomegaly, ECG shows biventricular hypertrophy

The chest x-ray and ECG have been reported as normal. The murmur is still present, and Finley undergoes an echocardiogram which reveals a moderate VSD. The cardiologist explains the normal progress of VSD to Finley's parents. Outline what the cardiologist would have explained to her parents and details the complications that may occur with larger ventricular septal defects.

Avg sized VSD - No worries, should be fine
Larger VSD - May need repair as too much mixing of blood
Risk of Paradoxical embolism with both

List and outline the pathophysiology of some acyanotic congenital heart conditions:

L to R shunts are acyanotic, but lead to issues later on when they increase pressures in pulmonary circulation and cause R-Ventricular Hypertrophy.

Persistent ductus arteriosus (Communication between aorta and Pulmonary Aorta remains open leading to a Left to Right Shunt - e.g. Oxygenated Blood shunted from to Pulmonary Circulation - good in ToF

Atrial septal defect (hole between atria, leads to fixed split S2 systolic murmur)

Pulmonary & Aortic stenosis (stiff valve, outflow obstruction)

Coarctation of the aorta - Narrowing of Aorta just proximal to ductus arteriosus - high pressures, can lead to rupture or aneurism.

ANSWERS

Question 1
What further history would you explore with Finley's mother and why?

General

- Pregnancy complications
- Birth complications
- Immunisations
- Family history of congenital heart disease or other
- Allergies
- Growth centiles
- Developmental 6 month check
- PMHx : wheezing episodes, bronchiolitis etc

Specific

- Feeding difficulties (breast or bottle)
- Sleep
- Breathing difficulties
- Presence of murmur
- Cyanosis (permanent or intermittent – eg with bathing, crying, feeding)

Question 2
What clinical findings on history and examination would suggest the presence of cardiac failure?

Cardiac failure in infancy tends to be dominated by pulmonary congestion, which leads to dyspnoea/tachypnoea.

Dyspnoea contributes to feeding difficulties, reduced intake and increased metabolic rate. Failure to thrive often results. Chronic dyspnoea may lead to the appearance of Harrison's sulci, which are deformations of the ribcage at the site of the diaphragmatic attachments. Crepitations at the lung bases are usually a manifestation of superimposed infection rather than heart failure in infants

Systemic venous congestion is manifest by liver enlargement and/or oedema. Liver engorgement results in an enlarged, abnormally firm liver with its edge palpable 2.5-5 cm below the costal margin. In infants, oedema is often diffuse and difficult to detect. It is often best seen around the face and eyes (periorbital oedema).

Elevated jugular venous pressure cannot be assessed easily in infancy

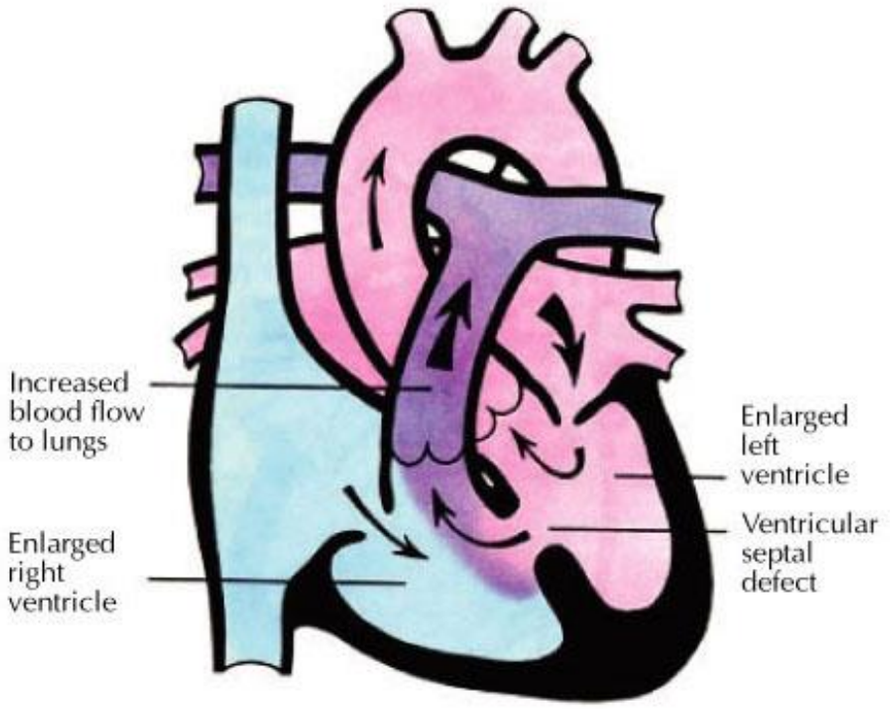
Other evidence of cardiac failure may include persistent tachycardia, a chronic dry cough and profuse sweating, especially of the forehead and scalp

Question 3

Finley has no clinical features of cardiac failure, but you are concerned that Finley may have a ventricular septal defect [VSD]. With the assistance of diagrams explain to Finley's mother what a VSD is how they may develop

Ventricular Septal Defect (VSD) comprises around 30% of all cardiac defects. They vary from tiny defects, of pinhole size, to huge defects. Small defects are more common than large ones and are usually asymptomatic.

Ventricular Septal Defect



Question 4

You speak with the Paediatrician in the clinic who suggests a cardiology consult. Outline the details that you would include on the referral letter.

- History of presenting symptom
- History – background information (see above)
- Examination findings
- Immunisation history
- Investigations (CXR)

Question 5

The cardiology registrar explains that a chest x-ray and ECG should be organised before Finley attends the paediatric cardiology clinic. What information can be obtained from these investigations?

With small defects, the chest X-ray and electrocardiogram (ECG) are frequently normal. With larger defects, the X-ray shows cardiomegaly and increased pulmonary vascular markings. The ECG often shows biventricular hypertrophy.

Question 6

You decide to attend Finley's appointment at the cardiology clinic. The chest x-ray and ECG have been reported as normal. The murmur is still present and Finley undergoes an echocardiogram which reveals a moderate VSD. The cardiologist explains the normal progress of VSD to Finley's parents. Outline what the cardiologist would have explained to her parents and details the complications that may occur with larger ventricular septal defects.

The natural history of a VSD varSs. Small defects frequently undergo spontaneous closure, which may occur in 50% or more. Some moderate defects may also diminish in size and the shunt becomes minor.

Important complications include progressive aortic incompetence, when one leaflet of the aortic valve is sucked into (prolapses) into an adjacent VSD, or the development of infundibular pulmonary stenosis.

Small isolated defects may be left alone if the evidence shows no significant haemodynamic disturbance and the patient remains symptom-free.

Large VSDs are associated with a variable degree of pulmonary hypertension. Pulmonary 'hypertension' is present from birth and may lead to the development of pulmonary vascular obliterative disease. Progression of pulmonary vascular damage, with increasing vascular resistance in the pulmonary circulation, will eventually result in reversal of the shunt, with the appearance of cyanosis (Eisenmenger syndrome), developing in adolescence or early adult life.

Question 7

Following the clinic, you decide to revise acyanotic heart disease, briefly outline the pathophysiology of the following conditions, persistent ductus arteriosis, atrial septal defect, pulmonary & aortic stenosis and coarctation of the aorta.

Left-to-right shunts (such as ASD, VSD, and patent ductus arteriosus (PDA)) increase pulmonary blood flow and are not initially associated with cyanosis. However, left- to-right shunts raise both flow volumes and pressures in the normally low-pressure, low-resistance pulmonary circulation, which can lead to right ventricular hypertrophy and pathological changes in the pulmonary vasculature. The muscular pulmonary arteries (<1 mm diameter) first respond to increased pressure and flow by undergoing hypertrophy and vasoconstriction, which reduces distal pulmonary capillary and venous pressures, and prevents pulmonary oedema. Prolonged pulmonary arterial vasoconstriction, however, stimulates the proliferation of the vascular wall cells and the consequent development of irreversible obstructive intimal lesions. Eventually,

pulmonary vascular resistance approaches systemic levels, thereby producing a new right-to-left shunt that introduces unoxygenated blood into the systemic circulation (Eisenmenger syndrome).

Persistent ductus arteriosis

The ductus arteriosus is a normal vascular channel during intrauterine life. It is a large vessel with a muscular wall which courses between the pulmonary artery and the aorta.

The ductus arteriosus normally closes within the first 48 hours of life. If it remains patent longer than this it is unlikely to close spontaneously. The exception is in premature babies where closure should occur by roughly the expected due date.

Atrial septal defect

An atrial septal defect (ASD) is an abnormal, fixed opening in the atrial septum caused by incomplete tissue formation that allows communication of blood between the left and right atria (not to be confused with patent foramen ovale, see below). ASDs are often asymptomatic in childhood. A small number will be importantly symptomatic in the first year of life and they are generally repaired before the age of 5.

Aortic stenosis

Aortic valve stenosis may be congenital or acquired. The congenital lesions are associated with thickening of the leaflets and variable fusion between the cusps producing most commonly a functionally bicuspid valve. The degree of stenosis varies from very mild (which may be asymptomatic well into adulthood) to severe, producing heart failure in the newborn period. If the obstruction is very severe the systemic circulation may be duct dependent in the newborn period and neonatal surgery/balloon dilatation will be required. In extreme cases the aortic valve obstruction may be associated with underdevelopment of the left heart structures and even opening the valve will not allow satisfactory forward systemic flow. These babies remain duct dependent and will generally not survive without complex surgery.

Subaortic stenosis is not commonly congenital but develops most commonly as a membrane below the valve. This is often associated with aortic valve regurgitation and usually requires surgical resection as it tends to be a progressive obstruction.

Supra-aortic stenosis is usually associated with Williams Syndrome and often these children also have small peripheral pulmonary arteries and a small proximal descending aorta. The supraaortic stenosis is commonly but not necessarily progressive.

Pulmonary Stenosis

This relatively frequent malformation constitutes an obstruction at the pulmonary valve, which may be mild to severe; the lesion can be isolated or part of a more complex anomaly-either tetralogy of Fallot or transposition of the great arteries. Right ventricular hypertrophy often develops. Mild stenosis may be asymptomatic and compatible with long life, whereas more severe cases warrant intervention, which is usually balloon dilatation.

Coarctation of the aorta

Coarctation {narrowing, constriction) is an obstruction of the aorta usually just proximal to the ductus arteriosis, beyond the left subclavian artery. It ranks high in frequency among the common structural anomalies. Males are affected twice as often as females, although females with Turner syndrome frequently have a coarctation. Encroachment on the aortic lumen is of variable severity, sometimes leaving only a small channel and at other times producing only minimal narrowing. Although coarctation of the aorta may occur as a solitary defect, it is accompanied by a bicuspid aortic valve in 50% of cases and may also be associated with congenital aortic stenosis, ASD, VSD, mitral regurgitation, or berry aneurysms of the circle of Willis in the brain.