Medicine – Dr. Omed – Lecture 6 - CONGENITAL HEART DISEASE

- usually manifests in childhood but may pass unrecognised and
- not present until adult life.
- Defects which are well tolerated, e.g. atrial septal defect,
- may cause no symptoms until adult life or may be detected
- incidentally on routine examination or chest X-ray.
- Congenital defects that were previously fatal in childhood can
- now be corrected, or at least partially corrected, so that survival
- to adult life is the norm. Such patients may remain well for many
- years and subsequently re-present in later life with related
- problems such as arrhythmia or ventricular dysfunction

The fetal circulation

- Understanding the fetal circulation helps to understand how some forms of congenital heart disease occur.
- The fetus has only a small flow of blood through the lungs, as it obviously does not breathe in utero.
- The fetal circulation therefore allows oxygenated blood from the placenta to pass directly to the left side of the heart through the foramen ovale without having to flow through the lungs. Congenital defects may arise if the changes from fetal circulation to the extraterine circulation are not properly completed.

PRESENTATION OF CONGENITAL HEART DISEASE THROUGHOUT LIFE

Birth and neonatal period
Cyanosis
Heart failure

Infancy and childhood
Cyanosis
Heart failure
Arrhythmia
Murmur
Failure to thrive

Adolescence and adulthood
Heart failure
Murmur
Arrhythmia
Cyanosis due to shunt reversal (Eisenmenger’s syndrome)
Hypertension (coarctation)
Late consequences of previous cardiac surgery, e.g. arrhythmia, heart failure
INCIDENCE AND RELATIVE FREQUENCY OF CONGENITAL CARDIAC MALFORMATIONS

Lesion% of all CHD

<table>
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<th>Lesion</th>
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<tbody>
<tr>
<td>Ventricular septal defect</td>
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<tr>
<td>Atrial septal defect</td>
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<tr>
<td>Patent ductus arteriosus</td>
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<td>Pulmonary stenosis</td>
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<td>Coarctation of aorta</td>
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<td>Aortic stenosis</td>
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Aetiology and incidence

- The incidence of haemodynamically significant congenital cardiac abnormalities is about 0.8% of live births.
- Maternal infection or exposure to drugs or toxins may cause congenital heart disease.
- Maternal rubella infection is associated with persistent ductus arteriosus, pulmonary valvular and/or artery stenosis, and atrial septal defect.
- Maternal alcohol misuse is associated with septal defects, and maternal lupus erythematosus with congenital complete heart block.
- Genetic or chromosomal abnormalities such as Down's syndrome may cause septal defects, and gene defects have also been identified as causing specific abnormalities, e.g. Marfan's and DiGeorge's (deletion in chromosome 22q) syndromes.

PERSISTENT DUCTUS ARTERIOSUS

Aetiology:

- During fetal life, before the lungs begin to function, most of the blood from the pulmonary artery passes through the ductus arteriosus into the aorta.
- Normally the ductus closes soon after birth but sometimes it fails to do so.
- Persistence of the ductus is more common in females.
- Since the pressure in the aorta is higher than that in the pulmonary artery (PA), there will be a continuous arteriovenous shunt.

Clinical features

- With small shunts there may be no symptoms for years, but when the ductus is large, *growth and development may be retarded.*
cardiac failure may eventually ensue, dyspnoea being the first symptom.

A continuous 'machinery' murmur is heard with late systolic accentuation, maximal in the second left intercostal space below the clavicle.

It is frequently accompanied by a thrill. Pulses are increased in volume.

Enlargement of the pulmonary artery may be detected radiologically.

The ECG is usually normal.

**Persistent ductus with reversed shunting:**

- If pulmonary vascular resistance increases, pulmonary artery pressure rises and may continue to do so until it equals or exceeds aortic pressure.
- The shunt through the defect may then reverse, causing central cyanosis (Eisenmenger's syndrome), which may be more apparent in the feet and toes than in the upper part of the body.
- The murmur becomes quieter, may be confined to systole or may disappear.
- The ECG shows evidence of right ventricular hypertrophy.

**Management:**

- It is now usual practice to close a patent ductus at cardiac catheterisation with an implantable occlusive device.
- Closure should be undertaken in infancy if the shunt is significant and pulmonary resistance not elevated,
- but this may be delayed until later childhood in those with smaller shunts, for whom closure remains advisable to reduce the risk of endocarditis.
- Pharmacological treatment in the neonatal period a prostaglandin synthetase inhibitor (indometacin or ibuprofen) may be used in the first week of life
- However, in the presence of a congenital defect with impaired lung perfusion (e.g. severe pulmonary stenosis and left-to-right shunt through the ductus), it may be advisable to improve oxygenation by keeping the ductus open with prostaglandin treatment.

**COARCTATION OF THE AORTA**

**Aetiology:**

Narrowing of the aorta most commonly occurs in the region where the ductus arteriosus joins the aorta, i.e. at the isthmus just below the origin of the left subclavian artery

- The condition is twice as common in males as in females and occurs in 1 in 4000 children.
- It is associated with other abnormalities, of which the most frequent are bicuspid aortic valve and 'berry' aneurysms of the cerebral circulation.
- Acquired coarctation of the aorta is rare but may follow trauma or occur complication of a progressive arteritis Takayasu’s disease.

**Clinical features and investigations**

- Aortic coarctation is an important cause of cardiac failure in the newborn, but symptoms are often absent when it is detected in older children or adults.
Headaches may occur from hypertension proximal to the coarctation.

- weakness or cramps in the legs may result from decreased circulation in the lower part of the body.

- The blood pressure is raised in the upper body but normal or low in the legs.

- The femoral pulses are weak, and delayed in comparison with the radial pulse.

- A systolic murmur is usually heard posteriorly, over the coarctation.

- There may also be an ejection click and systolic murmur in the aortic area due to a bicuspid aortic valve.

- As a result of the aortic narrowing, collaterals form, mainly involving the periscapular, internal mammary and intercostal arteries.

- These may result in localized bruits.

- Chest X-ray in early childhood is often normal but at a later age may show changes in the contour of the aorta (indentation of the descending aorta, '3 sign') and notching of the under-surfaces of the ribs from collaterals.

- MRI is ideal for demonstrating the lesion.

- The ECG may show left ventricular hypertrophy

**Management**

- In untreated cases, death may occur from left ventricular failure, dissection of the aorta or cerebral hemorrhage.

- Surgical correction is advisable in all but the mildest cases.

- If this is done sufficiently early in childhood, persistent hypertension can be avoided.

- Patients repaired in late childhood or adult life often remain hypertensive or develop recurrent hypertension later in life.

- Recurrence of stenosis may occur as the child grows, and this may be managed by balloon dilatation, which can also be used as the primary treatment in some cases.

**ATRIAL SEPTAL DEFECT**

**Aetiology:** is one of the most common congenital heart defects.

- occurs twice as frequently in females.

- Most are 'ostium secundum' defects, involving the fossa ovale which in utero was the foramen ovale

- 'Ostium primum' defects result from a defect in the atrioventricular septum and are associated with a 'cleft mitral valve' (split anterior leaflet).
Since the normal right ventricle is more compliant than the left, a large volume of blood shunts through the defect from the left to the right atrium and then to the right ventricle and pulmonary arteries.

As a result there is gradual enlargement of the right side of the heart and of the pulmonary arteries.

Pulmonary hypertension and shunt reversal sometimes complicate atrial septal defect, but are less common and tend to occur later in life than with other types of left-to-right shunt.

Clinical features:

- Most children are free of symptoms for many years and the condition is often detected at routine clinical examination or following a chest X-ray.
- Dyspnoea, chest infections, cardiac failure and arrhythmias, especially atrial fibrillation, are other possible modes of presentation.
- Wide fixed splitting of the second heart sound: wide because of delay in right ventricular ejection (increased stroke volume and right bundle branch block) and fixed because the septal defect equalises left and right atrial pressures throughout the respiratory cycle.
- A systolic flow murmur over the pulmonary valve.
- In children with a large shunt, there may be a diastolic flow murmur over the tricuspid valve. Unlike a mitral flow murmur, this is usually high-pitched.

The chest X-ray

- Typically shows enlargement of the heart and the pulmonary artery as well as pulmonary plethora.
- The ECG usually shows incomplete right bundle branch block because right ventricular depolarisation is delayed as a result of ventricular dilatation. (With a 'primum' defect there is also left axis deviation).
- Echocardiography can directly demonstrate the defect and typically shows RV dilatation, RV hypertrophy and pulmonary artery dilatation. The precise size and location of the defect can be shown by transoesophageal echocardiography.

Management

- Atrial septal defects in which pulmonary flow is increased 50% above systemic flow (i.e. flow ratio of 1.5:1) are often large enough to be clinically recognisable and should be closed surgically. Closure can also be accomplished at cardiac catheterisation using implantable closure devices.
- The long-term prognosis thereafter is excellent unless pulmonary hypertension has developed.
- Severe pulmonary hypertension and shunt reversal are both contraindications to surgery.
VENTRICULAR SEPTAL DEFECT

- Aetiology: occurs as a result of incomplete septation of the ventricles.
- Embryologically, the interventricular septum has a membranous and a muscular portion, and the latter is further divided into inflow, trabecular and outflow portions.
- Most congenital defects are ‘perimembranous’, i.e. at the junction of the membranous and muscular portions.
- V.S.D are the most common congenital cardiac defect, occurring once in 500 live births.
- The defect may be isolated or part of complex congenital heart disease.
- Acquired ventricular septal defect may result from rupture as a complication of acute myocardial infarction, or rarely from trauma.

Clinical features

- Flow from the high-pressure left ventricle to the low-pressure right ventricle during systole produces a pansystolic murmur usually heard best at the left sternal edge but radiating all over the precordium.
- A small defect often produces a loud murmur (maladie de Roger) in the absence of other haemodynamic disturbance.
- Conversely, a large defect may produce a softer murmur, particularly if pressure in the right ventricle is elevated.
- This may be found immediately after birth, while pulmonary vascular resistance remains high, or when the shunt is reversed in Eisenmenger’s syndrome.
- Congenital ventricular septal defect may present as:
  - cardiac failure in infants,
  - as a murmur with only minor haemodynamic disturbance in older children or adults,
  - or rarely as Eisenmenger’s syndrome.
- In a proportion of infants, the murmur gets quieter or disappears due to spontaneous closure of the defect.
- If cardiac failure complicates a large defect, it is usually absent in the immediate postnatal period and only becomes apparent in the first 4-6 weeks of life.
- The chest X-ray shows pulmonary plethora
- ECG shows bilateral ventricular hypertrophy.

Management:

- Small VSD require no specific treatment apart from endocarditis prophylaxis.
- Cardiac failure in infancy is initially treated medically with digoxin and diuretics.
- Persisting failure is an indication for surgical repair of the defect.
- Percutaneous closure devices are under development.
- Doppler echocardiography helps to predict the small septal defects that are likely to close spontaneously.
Surgical closure is contraindicated in fully developed Eisenmenger's syndrome when heart-lung transplantation may be the only effective method of treatment.

**Prognosis**

- Except in the case of Eisenmenger's syndrome, long-term prognosis is very good in congenital ventricular septal defect.
- Many patients with Eisenmenger's syndrome die in the second or third decade of life, but a few survive to the fifth decade without transplantation.

**TETRALOGY OF FALLOT**

- The tetralogy comprises (1) pulmonary stenosis, (2) overriding of the ventricular septal defect by the aorta, (3) a ventricular septal defect and (4) right ventricular hypertrophy.

**Aetiology**

- The embryological cause is abnormal development of the bulbar septum which separates the ascending aorta from the pulmonary artery, and which normally aligns and fuses with the outflow part of the interventricular septum.
- The defect occurs in about 1 in 2000 births and is the most common cause of cyanosis in infancy after the first year of life.

**Clinical features**

- Children are usually cyanosed but this may not be present in the neonate because it is only when right ventricular pressure rises to equal or exceed left ventricular pressure that a large right-to-left shunt develops.
- The subvalvular component of the right ventricle outflow obstruction is dynamic, and may increase suddenly under adrenergic stimulation.
- The affected child suddenly becomes increasingly cyanosed, often after feeding or a crying attack, and may become apnoeic and unconscious.
- These attacks are called 'Fallot's spells'.
- In older children, Fallot's spells are uncommon but cyanosis becomes increasingly apparent, with stunting of growth, digital clubbing and polycythemia.
- Some children characteristically obtain relief by squatting after exertion, which increases the afterload of the left heart and reduces the right-to-left shunting.
- The natural history before the development of surgical correction was variable, but most patients died in infancy or childhood.

**On examination**

- the most characteristic feature is the combination of cyanosis with a loud ejection systolic murmur in the pulmonary area (as for pulmonary stenosis).
- However, cyanosis may be absent in the newborn or in patients with only mild right ventricular outflow obstruction ('acyanotic tetralogy of Fallot').

**Investigations**

- The ECG shows right ventricular hypertrophy,
chest X-ray shows an abnormally small pulmonary artery and a 'boot-shaped' heart.

Echocardiography is diagnostic and demonstrates that the aorta is not continuous with the anterior ventricular septum.

Management

- The definitive management is total correction of the defect by surgical relief of the pulmonary stenosis and closure of the ventricular septal defect.
- Primary surgical correction may be undertaken prior to age 5, unless the pulmonary arteries are too hypoplastic, when a palliative shunt may be performed (e.g. the Blalock-Taussig shunt, an anastomosis between the pulmonary artery and subclavian artery).
- The prognosis after total correction is good, especially if the operation is performed in childhood.

OTHER CAUSES OF CYANOTIC CONGENITAL HEART DISEASE

- Tricuspid atresia
- Transposition of great vessels
- Pulmonary atresia
- Ebstein's anomaly