CONGENITAL HEART DISEASE

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.

Congenital defects may arise if the changes from fetal circulation to the extrauterine circulation are not properly completed. Atrial septal defects occur at the site of the foramen ovale. A patent ductus arteriosus may remain if it fails to close after birth. Failure of the aorta to develop at the point of the aortic isthmus and where the ductus arteriosus attaches can lead to narrowing or coarctation of the aorta.

In fetal development, the heart develops as a single tube which folds back on itself and then divides into two separate circulations. Failure of septation can lead to some forms of atrial and ventricular septal defect. Failure of alignment of the great vessels with the ventricles contributes to transposition of the great arteries, tetralogy of Fallot and truncus arteriosus.

Incidence:

The incidence of haemodynamically significant congenital cardiac abnormalities is about 0.8% of live births (Table 1).

Table 1: INCIDENCE AND RELATIVE FREQUENCY OF CONGENITAL CARDIAC MALFORMATIONS

<table>
<thead>
<tr>
<th>Lesion</th>
<th>% of all CHD defects</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ventricular septal defect</td>
<td>30</td>
</tr>
<tr>
<td>Atrial septal defect</td>
<td>10</td>
</tr>
<tr>
<td>Patent ductus arteriosus</td>
<td>10</td>
</tr>
<tr>
<td>Pulmonary stenosis</td>
<td>7</td>
</tr>
<tr>
<td>Coarctation of aorta</td>
<td>7</td>
</tr>
<tr>
<td>Aortic stenosis</td>
<td>6</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>6</td>
</tr>
<tr>
<td>Complete transposition of great arteries</td>
<td>4</td>
</tr>
<tr>
<td>Others</td>
<td>20</td>
</tr>
</tbody>
</table>
Aetiology:

1. *Maternal infection:* e.g. maternal rubella infection is associated with persistent ductus arteriosus, pulmonary valvular and/or artery stenosis, and atrial septal defect.
2. *Maternal exposure to drugs or toxins:* Maternal alcohol misuse is associated with septal defects.
3. *Maternal diseases:* Maternal lupus erythematosus may be associated with congenital complete heart block.
4. *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.

Clinical features of CHD in general:

1. Symptoms may be absent (asymptomatic).
2. Some defects are not compatible with extrauterine life, or compatible only for a short time.
3. Abnormal chest X-ray taken for other reasons.
4. Murmurs, thrills or signs of cardiomegaly may be present on routine physical examination or felt by the mother.
5. The child may be breathless & has other features of heart failure as oedema & gallop rhythm.
6. In coarctation of the aorta, hypertension & radio-femoral delay may be noted.
7. Skeletal abnormalities from associated congenital conditions. Tall stature with long limbs and lens dislocation may be obvious in Marfan's syndrome or features of other congenital conditions such as Down's syndrome, may also be apparent.
8. Central cyanosis and digital clubbing. Central cyanosis of cardiac origin occurs when desaturated blood enters the systemic circulation without passing through the lungs (i.e. a right-to-left shunt). In the neonate, the most common cause is transposition of the great arteries. In older children, cyanosis is usually the consequence of a ventricular septal defect combined with severe pulmonary stenosis (tetralogy of Fallot) or with pulmonary vascular disease (Eisenmenger's syndrome). Prolonged cyanosis is associated with finger and toe clubbing.
9. Cerebrovascular accidents and cerebral abscesses are complications of severe cyanotic congenital disease.
10. Growth retardation and learning difficulties. These may be a feature with large left-to-right shunts at ventricular or great arterial level, but can also occur with other defects, especially if they form part of a genetic syndrome. Major intellectual impairment is uncommon in children with isolated congenital heart disease; however, minor learning difficulties can occur and may also be the consequence of cardiac surgery.
11. Syncope. In the presence of increased pulmonary vascular resistance or severe left or right ventricular outflow obstruction, exercise may provoke syncope as systemic vascular resistance falls on exercise but pulmonary vascular resistance may rise, worsening right-to-left shunting and cerebral oxygenation.
12. Pulmonary hypertension and Eisenmenger's syndrome. Persistently raised pulmonary flow (e.g. with left-to-right shunt) leads to increased pulmonary resistance followed by pulmonary hypertension. Progressive changes, including obliteration of distal vessels, take place in the pulmonary vasculature and, once established, the increased pulmonary resistance is irreversible. Central cyanosis appears and digital clubbing develops. The chest X-ray shows enlarged central pulmonary arteries and peripheral 'pruning' of the pulmonary vessels. The ECG shows right ventricular hypertrophy. If severe pulmonary hypertension develops, a left-to-right shunt may reverse, resulting in right-to-left shunting and marked cyanosis (Eisenmenger's syndrome). Patients with Eisenmenger's syndrome are at particular
risk from abrupt changes in afterload that exacerbate right-to-left shunting, e.g. vasodilatation, anaesthesia, pregnancy.

13. Pregnancy. CHD may present as exacerbation of hemodynamic changes of pregnancy. Abnormalities causing severe outflow tract obstruction, such as aortic stenosis, are not well tolerated and are associated with significant maternal morbidity and mortality. Pregnancy is particularly hazardous in the presence of conditions associated with cyanosis or severe pulmonary hypertension.

14. Infective endocarditis. All CHD except ostium secondum ASD can be complicated by infective endocarditis.

**Classification:** Two main categories.

<table>
<thead>
<tr>
<th>A- Acyanotic CHD</th>
<th>B- Cyanotic CHD</th>
</tr>
</thead>
<tbody>
<tr>
<td>Atrial septal defect</td>
<td>Tetralogy of Fallot</td>
</tr>
<tr>
<td>Ventricular septal defect</td>
<td>Transposition of great arteries</td>
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</tr>
<tr>
<td>Pulmonary stenosis</td>
<td>Epstein anomaly</td>
</tr>
<tr>
<td>Coarctation of aorta</td>
<td></td>
</tr>
</tbody>
</table>

These can be further classified according to the presence or absence of shunt (left to right or right to left).

**PERSISTENT DUCTUS ARTERIOSUS**

**Pathophysiology:**

Normally the ductus closes soon after birth but sometimes it fails to do so. Persistence of the ductus may be associated with other abnormalities and 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 arterio-venous shunt, the volume of which depends on the size of the ductus. As much as 50% of the left ventricular output may be re-circulated through the lungs, increasing pulmonary blood flow (pulmonary plethora on CXR), with a consequent volume overload of left-sided cardiac chambers & increase in the work load of these chambers. Untreated, this will lead to increased pulmonary vascular resistance → pulmonary hypertension → Eisenmenger’s syndrome.

**Clinical features:**

With small shunts there may be no symptoms for years, but when the ductus is large, growth and development may be retarded. Usually there is no disability in infancy but cardiac failure may eventually ensue, dyspnea being the first symptom, and there may be recurrent chest infections. 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.

A large left-to-right shunt in infancy may cause a considerable rise in pulmonary artery pressure, and sometimes this leads to progressive pulmonary vascular damage. Enlargement of the pulmonary artery may be detected radiologically.

**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.

**Investigations:**

*ECG:* can be normal or shows LA & LV hypertrophy & dilatation. In Eisenmenger’s syndrome, it shows right ventricular hypertrophy & right axis deviation.

*CXR:* can be normal or shows pulmonary plethora & cardiomegaly (mainly LA & LV)& enlargement of aortic root & pulmonary artery. In Eisenmenger’s syndrome, it shows enlargement of right-sided chambers & pulmonary artery & pulmonary oligemia (↓ pulmonary vascularity).

*Echocardiography:* confirms the Dx & measures the size of cardiac chambers & estimates the pulmonary artery pressure.

*Cardiac catheterization:* may be required to confirm the Dx & prior to closure.

**Management:**

*Pharmacological treatment in the neonatal period*

When the ductus is structurally intact, a prostaglandin synthetase inhibitor (indometacin or ibuprofen) may be used in the first week of life to induce closure. Unfortunately, these treatments do not work if the ductus is intrinsically abnormal.

*Percutaneous closure with an implantable occlusive device*

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.

*Surgical closure:* if percutaneous closure is not feasible.

In Eisenmenger’s syndrome, closure is contraindicated.
ATRIAL SEPTAL DEFECT

Pathophysiology:

Atrial septal defect is one of the most common congenital heart defects, and occurs twice as frequently in females. It is classified according to location:

1. Ostium secundum defect: involving the fossa ovalis (mid portion). Most common type.
2. Ostium primum defect: result from a defect in the lower portion of septum and are associated with abnormalities of atrio-ventricular valves.

ASD → a large volume of blood shunts through the defect from the left to the right atrium → right ventricle → 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 (Eisenmenger’s syndrome) 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. Dyspnea, recurrent chest infections, cardiac failure and arrhythmias, especially atrial fibrillation, are other possible modes of presentation. The characteristic physical signs are the result of the volume overload of the right ventricle:

- Wide fixed splitting of the second heart sound
- A systolic flow murmur over the pulmonary valve (Lt second intercostal space).

In children with a large shunt, there may be a diastolic flow murmur over the tricuspid valve. In ostium primum ASD, pansystolic murmer of mitral &/or tricuspid regurgitation may be heard also.

Investigations:

The chest X-ray: typically shows enlargement of the heart and the pulmonary artery as well as pulmonary plethora.

The ECG: usually shows incomplete or complete right bundle branch block.

In secondum defect → right axis deviation.

In primum defect → 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 ≥1.5:1) 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

Pathophysiology:

Congenital ventricular septal defect 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.

Ventricular septal defects 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.

VSD → Lt to Rt shunt at ventricular level → ↑pulmonary blood flow → volume overload of LV.

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). Conversely, a large defect may produce a softer murmur.

Congenital ventricular septal defect may present as dyspnea on exertion, cardiac failure, growth retardation and recurrent chest infections 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.

In addition to the murmur, there is prominent parasternal pulsation, tachypnoea and indrawing of the lower ribs on inspiration.

Investigations:

The chest X-ray: shows pulmonary plethora & cardiomegaly in large defects.

ECG: may be normal in small defects or shows bilateral ventricular hypertrophy in large defects.

Echo: shows the defect & measures its size.

Management:

- Small ventricular septal defects require no specific treatment apart from endocarditis prophylaxis.

- Large defects with pulmonary: systemic flow $\geq$ 1.5:1 with LV volume overload → surgical closure.
Cardiac failure caused by a ventricular septal defect 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.

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.

**COARCTATION OF THE AORTA**

**Pathophysiology:**

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.

**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, and occasionally 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 localised bruits. In untreated cases, death may occur from left ventricular failure, dissection of the aorta or cerebral haemorrhage.

**Investigations:**

*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.

*ECG:* may show left ventricular hypertrophy.
Management:

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. Coexistent bicuspid aortic valve, which occurs in over 50% of cases, may lead to progressive aortic stenosis or regurgitation and also requires long-term follow-up.

PULMONARY STENOSIS

Clinical features, investigations and management

The principal finding on examination is an ejection systolic murmur, loudest to the left of the upper sternum and radiating towards the left shoulder. There may be a thrill, best felt when the patient leans forward and breathes out. The murmur is often preceded by an ejection sound (click). Delay in right ventricular ejection may cause wide splitting of the second heart sound. Severe pulmonary stenosis is characterised clinically by a loud harsh murmur, an inaudible pulmonary closure sound (P₂), an increased right ventricular heave, prominent a waves in the jugular pulse, ECG evidence of right ventricular hypertrophy, and post-stenotic dilatation in the pulmonary artery on the chest X-ray. Doppler echocardiography is the definitive investigation.

Mild to moderate isolated pulmonary stenosis is relatively common, does not usually progress and does not require treatment; it is a low-risk lesion for infective endocarditis. Severe pulmonary stenosis (resting gradient > 50 mmHg with a normal cardiac output) is treated by percutaneous pulmonary balloon valvuloplasty or, if not available, by surgical valvotomy. Long-term results are very good.

TETRALOGY OF FALLOT

The four components of the tetralogy are:

1. Right ventricular outflow obstruction: is most often subvalvular (infundibular), but may be valvular, supravalvular or a combination of these.
2. Ventricular septal defect: usually large and similar in aperture to the aortic orifice.
3. RV hypertrophy.
4. Overriding aorta.

TOF → Elevated right ventricular pressure and right-to-left shunting of cyanotic blood across the ventricular septal defect.

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 polycythaemia. 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:

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 between the pulmonary artery and subclavian artery). This improves pulmonary blood flow and pulmonary artery development, and may facilitate definitive correction at a later stage.

The prognosis after total correction is good, especially if the operation is performed in childhood. Follow-up is needed to identify residual shunting, recurrent pulmonary stenosis and rhythm disorders.
Table 2: OTHER CAUSES OF CYANOTIC CONGENITAL HEART DISEASE

<table>
<thead>
<tr>
<th>Defect</th>
<th>Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tricuspid atresia</td>
<td>Absent tricuspid orifice, hypoplastic RV, RA to LA shunt, VSD shunt, other anomalies Surgical correction may be possible</td>
</tr>
<tr>
<td>Transposition of the great vessels</td>
<td>Aorta arises from the morphological RV, pulmonary artery from LV Shunt via atria, ductus and possibly VSD Palliation by balloon atrial septostomy/enlargement Surgical correction possible</td>
</tr>
<tr>
<td>Pulmonary atresia</td>
<td>Pulmonary valve atretic and pulmonary artery hypoplastic RA to LA shunt, pulmonary flow via ductus Palliation by balloon atrial septostomy Surgical correction may be possible</td>
</tr>
<tr>
<td>Ebstein's anomaly</td>
<td>Tricuspid valve is dysplastic and displaced into RV, right ventricle 'atrialised' Tricuspid regurgitation and RA to LA shunt Wide spectrum of severity Arrhythmias Surgical repair possible, but significant risks</td>
</tr>
</tbody>
</table>

OTHER CAUSES OF CYANOTIC CONGENITAL HEART DISEASE

Other causes of cyanotic congenital heart disease are summarised in table 2. Echocardiography is usually the definitive diagnostic procedure, supplemented if necessary by cardiac catheterization.

ADULT CONGENITAL HEART DISEASE

There are increasing numbers of children who have had surgical correction of congenital defects and who may have further cardiological problems as adults. For example, those who have undergone correction of coarctation of the aorta may develop hypertension in adult life. Those with transposition of the great arteries who have had a 'Mustard' repair, where blood is re-directed at atrial level leaving the right ventricle connected to the aorta, may develop right ventricular failure in adult life. The right ventricle is unsuited for function at systemic pressures and may begin to dilate and fail when patients are in their 20s or 30s.

Those who have had surgery involving the atra may develop atrial arrhythmias, and those who have ventricular scars may develop ventricular arrhythmias. Such patients require careful follow-up from the teenage years through adult life so that problems can be identified early and appropriate medical or surgical treatment instituted.