CONGENITAL HEART DISEASES

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16.99 Incidence and relative frequency of congenital cardiac malformations

Lesion	% of all congenital heart defects
Ventricular septal defect	30
Atrial septal defect	10
Persistent ductus arteriosus	10
Pulmonary stenosis	7
Coarctation of aorta	7
Aortic stenosis	6
Tetralogy of Fallot	6
Complete transposition of great arteries	4
Others	20

16.100 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
- Eisenmenger's syndrome
- Hypertension (coarctation)
- Complications of previous cardiac surgery:
  - Arrhythmia related to scarring
  - Heart failure secondary to scarring

- **Eisenmenger’s syndrome**
- In patients with severe and prolonged pulmonary hypertension the left-to-right shunt may reverse, resulting in right-to-left shunt and marked cyanosis. This is termed Eisenmenger’s syndrome.
- The cyanosis in Eisenmenger’s syndrome may be more apparent in the feet and toes than in the upper part of the body, resulting in so-called differential cyanosis.
- Eisenmenger’s syndrome is more common with large ventricular septal defects or persistent ductus arteriosus than with atrial septal defects. Patients with Eisenmenger’s syndrome are at particular risk from abrupt changes in afterload that exacerbate right-to-left shunting, such as vasodilatation, anaesthesia and pregnancy.

Persistent ductus arteriosus

- Normally, the ductus arteriosus closes soon after birth but in this anomaly it fails to do so. Persistence of the ductus is often associated with other abnormalities and is more common in females.

Pathophysiology

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.

Persistence of the ductus causes a continuous AV shunt from the aorta to the pulmonary artery since pressure in the aorta is higher than that in the pulmonary circulation.

The volume of the shunt depends on the size of the ductus but as much as 50% of the left ventricular output may be recirculated through the lungs, with a consequent increase in the work of the heart.

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.

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, 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. If pulmonary vascular resistance increases, pulmonary artery pressure may rise until it equals or exceeds aortic pressure. The shunt through the defect may then reverse, causing Eisenmenger’s syndrome. The murmur becomes quieter, may be confined to systole or may disappear.

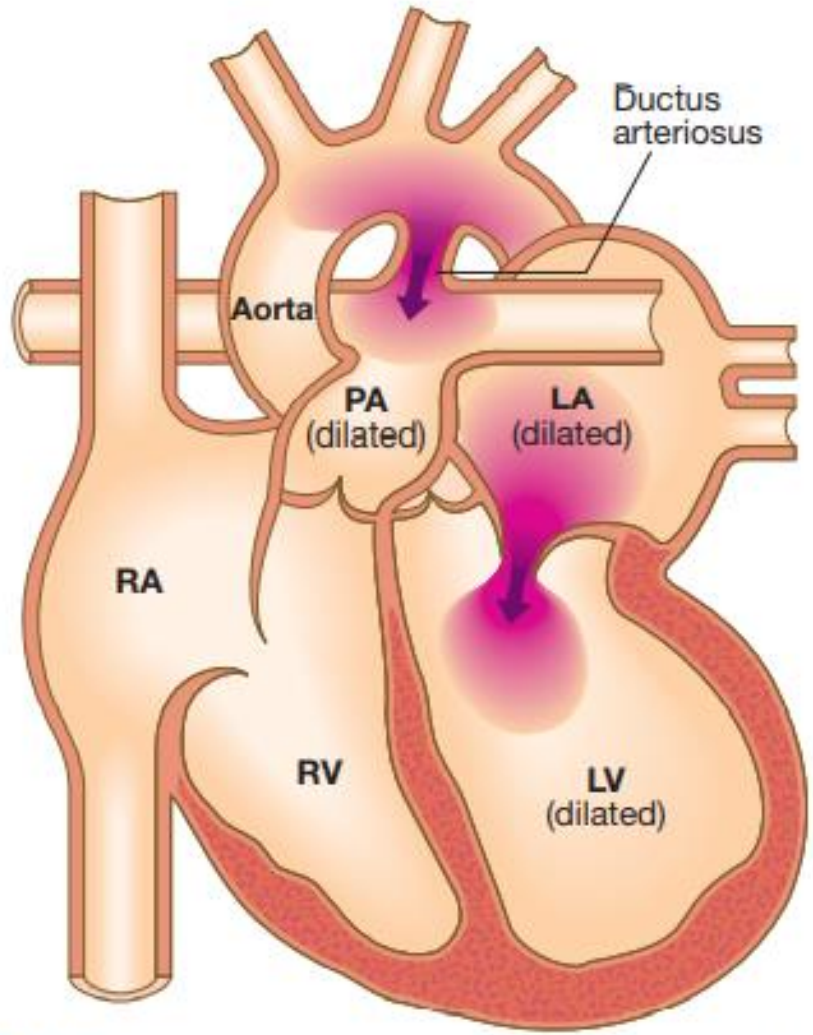


Fig. 16.92 Persistent ductus arteriosus. There is a connection between the aorta and the pulmonary artery with left-to-right shunting. (LA = left atrium; LV = left ventricle; PA = pulmonary artery; RA = right atrium; RV = right ventricle)

- **Investigations**

Echocardiography is the investigation of choice although the persistent ductus requires specific echocardiographic views, such as from the suprasternal notch, to reveal it.

The ECG shows evidence of right ventricular hypertrophy

- **Management**
- A persistent ductus can be closed 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. When the ductus is structurally intact, a prostaglandin synthetase inhibitor (indomethacin or ibuprofen) may be used in the first week of life to induce closure. However, in the presence of a congenital defect with impaired lung perfusion, such as occurs in 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. Unfortunately, these treatments do not work if the ductus is intrinsically abnormal.

- **Coarctation of the aorta**
- More in males
- It is associated with other abnormalities, most frequently **bicuspid aortic valve and 'berry' aneurysms of the cerebral circulation**
- Acquired coarctation of the aorta is rare but may follow trauma or occur as a complication of a progressive arteritis (Takayasu's disease)
- . **Pathogenesis**
- Narrowing of the aorta occurs in the region where the ductus arteriosus joins the aorta, at the isthmus just below the origin of the left subclavian artery
- This causes raised BP affecting vessels of the head and neck proximal to the coarctation, and reduced BP and impaired circulation distally.
- **Clinical features**
- Aortic coarctation is an important cause of cardiac failure in the newborn but symptoms are often absent 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 BP 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; they mainly involve the periscapular, internal mammary and intercostal arteries, and may result in localised bruits**.

- **Investigations**
- Imaging by MRI is the investigation of choice
- The chest X-ray in early childhood is often normal but later 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.**
- The ECG may show evidence of left ventricular hypertrophy, which can be confirmed by echocardiography.
- **Management**
- In untreated cases, death may occur from left ventricular failure, dissection of the aorta or cerebral haemorrhage.
- Surgical correction is advisable in all but the mildest cases. If this is carried out 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 on. 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.

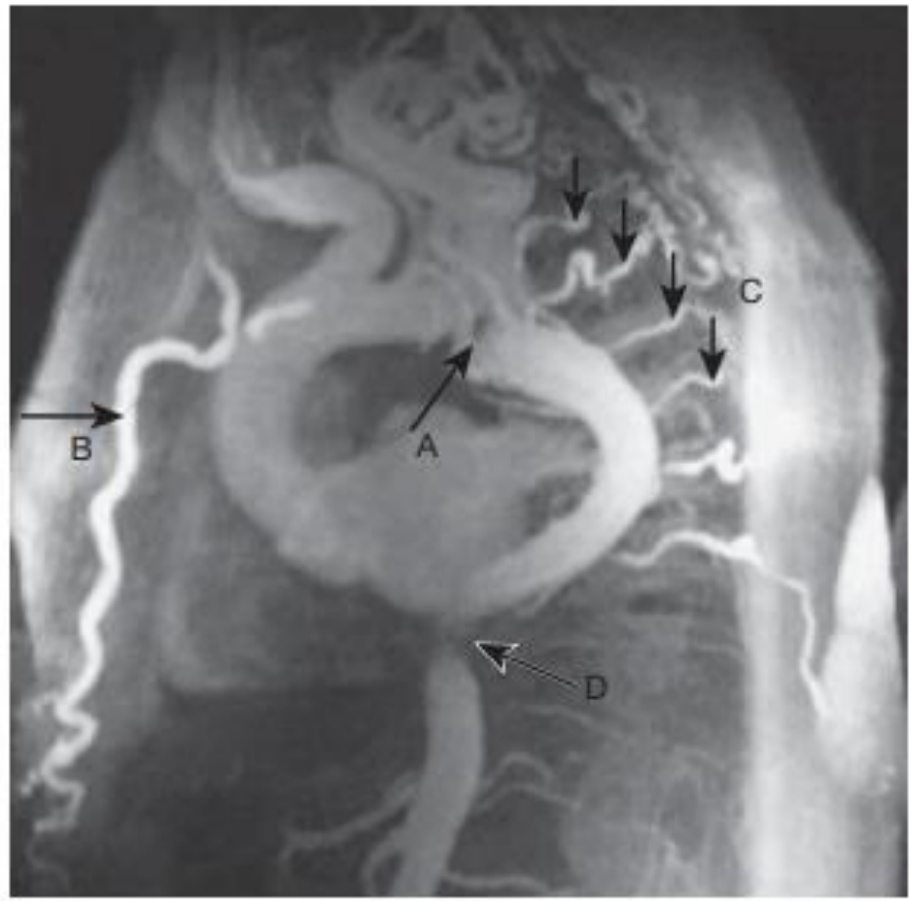


Fig. 16.93 MRI scan of coarctation of the aorta. The aorta is severely narrowed just beyond the arch at the start of the descending aorta (arrow A). Extensive collaterals have developed; a large internal mammary artery (arrow B) and several intercostal arteries (arrows C) are shown. Unusually, in this case, there is also a coarctation of the abdominal aorta (arrow D).

- **Atrial septal defect**
- Atrial septal defect is one of the most common congenital heart defects and occurs twice as frequently in females. Most are ‘ostium secundum’ defects, involving the fossa ovalis that, 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).
- **Pathogenesis** Since the normal RV is more compliant than the LV, a patent foramen ovale is associated with shunting of blood from the LA to the RA, and then to the RV 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 asymptomatic for many years and the condition is often detected at routine clinical examination or following a chest X-ray. Symptoms that can occur include dyspnoea, chest infections, cardiac failure and arrhythmias, especially AF. **The characteristic physical signs are the result of the volume overload of the RV:**

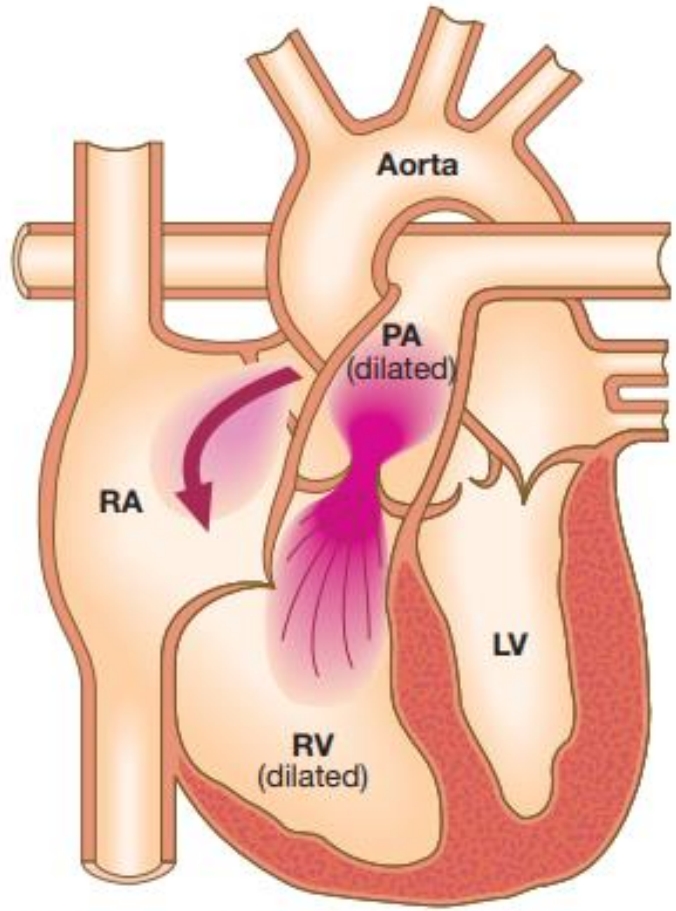


Fig. 16.94 Atrial septal defect. Blood flows across the atrial septum (arrow) from left to right. The murmur is produced by increased flow velocity across the pulmonary valve, as a result of left-to-right shunting and a large stroke volume. The density of shading is proportional to velocity of blood flow. (LV = left ventricle; PA = pulmonary artery; RA = right atrium; RV = right ventricle)

- wide, fixed splitting of the second heart sound: wide because of delay in right ventricular ejection (increased stroke volume and RBBB), 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.

- **Investigations**
- Echocardiography is diagnostic. It directly demonstrates the defect and typically shows right ventricular dilatation, right ventricular hypertrophy and pulmonary artery dilatation. The precise size and location of the defect are best defined by TOE .
- The chest X-ray typically shows enlargement of the heart and the pulmonary artery, as well as pulmonary plethora. The ECG usually demonstrates incomplete RBBB because right ventricular depolarisation is delayed as a result of ventricular dilatation (with a ‘primum’ defect, there is also left axis deviation).
- **Management**
- Atrial septal defects in which pulmonary flow is increased 50% above systemic flow (i.e. a flow ratio of 1.5 : 1) are often large enough to be clinically recognisable and should be closed surgically. (Smaller defects may be managed conservatively and patients are monitored with echocardiography.) 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

- **<u>Ventricular septal defect</u>**
- Ventricular septal defect is the most common congenital cardiac defect
- The defect may be isolated or part of complex congenital heart disease.
- **Pathogenesis**
- 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', occurring at the junction of the membranous and muscular portions of the septum.
- **Clinical features**
- Flow from the high-pressure LV to the low-pressure RV 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 produces a softer murmur, particularly if pressure in the RV 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 becomes 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 becomes apparent only in the first 4–6 weeks of life. In addition to the murmur, there is prominent parasternal pulsation, tachypnoea and indrawing of the lower ribs on inspiration.

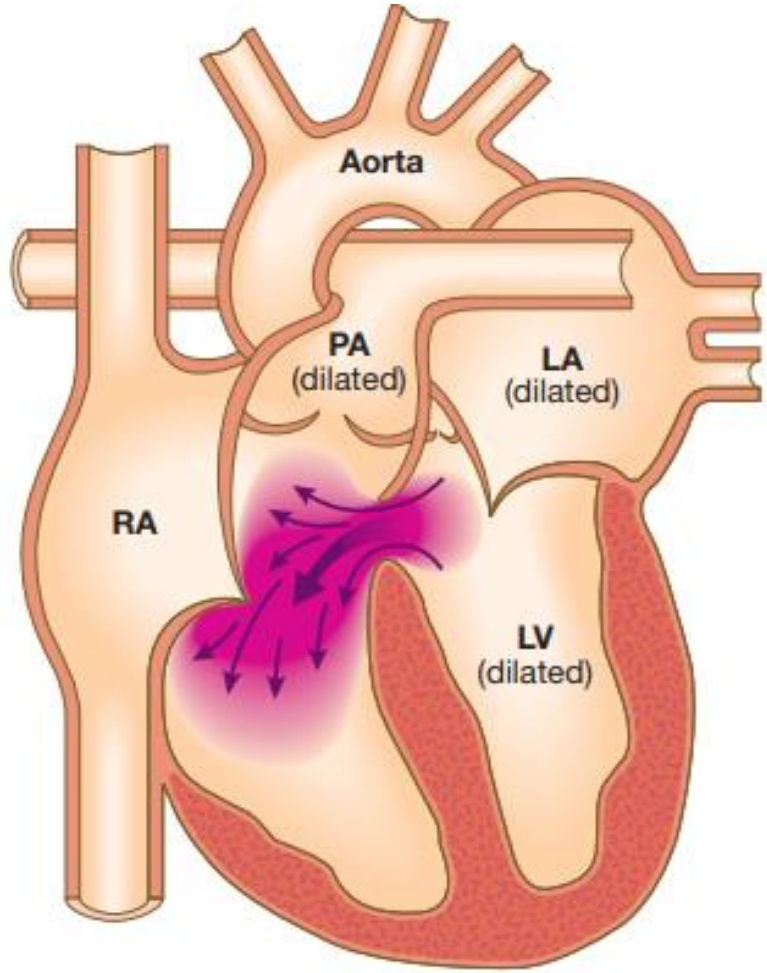


Fig. 16.97 Ventricular septal defect. In this example, a large left-to-right shunt (arrows) has resulted in chamber enlargement. (LA = left atrium; LV = left ventricle; PA = pulmonary artery; RA = right atrium)

- **Investigations**
- Doppler echocardiography should be performed since it helps to identify the small septal defects that are not haemodynamically significant and are likely to close spontaneously.
- Patients with larger defects should be monitored by serial ECG and echocardiography to screen for signs of pulmonary hypertension. With larger defects, the chest X-ray shows pulmonary congestion and the ECG shows bilateral ventricular hypertrophy.
- **Management:**
- Small ventricular septal defects require no specific treatment. If there is cardiac failure in infancy, this should initially be treated medically with digoxin and diuretics.
- Persisting failure is an indication for surgical repair of the defect. Percutaneous closure devices are under development. If serial ECG and echocardiography suggest that pulmonary hypertension is developing, surgical repair should be performed. Surgical closure is contraindicated in fully developed Eisenmenger's syndrome, in which case heart–lung transplantation is the only effective treatment.
- The long-term prognosis is generally very good. An exception is in Eisenmenger's syndrome, when death normally occurs in the second or third decade of life, but a few individuals survive to the fifth decade without transplantation.

- **Tetralogy of Fallot**
- This is complex defect consisting of right ventricular outflow tract obstruction and right ventricular hypertrophy, a large ventricular septal defect and an over-riding aorta that, when combined with the septal defect, allows blood to be pumped directly from the RV into the aorta.
- **It is the most common cause of cyanosis in infancy after the first year of life.**
  Pathogenesis
- Tetralogy of Fallot occurs as the result of abnormal development of the bulbar septum that separates the ascending aorta from the pulmonary artery, and which normally aligns and fuses with the outflow part of the interventricular septum. The right ventricular outflow obstruction is most often subvalvular (infundibular) but may be valvular, supravalvular or a combination of these.
- The subvalvular component of the right ventricular outflow obstruction is dynamic and may increase suddenly under adrenergic stimulation. The ventricular septal defect is usually large and similar in aperture to the aortic orifice. The combination results in elevated right ventricular pressure and right-to-left shunting of cyanotic blood across the VSD into aorta.

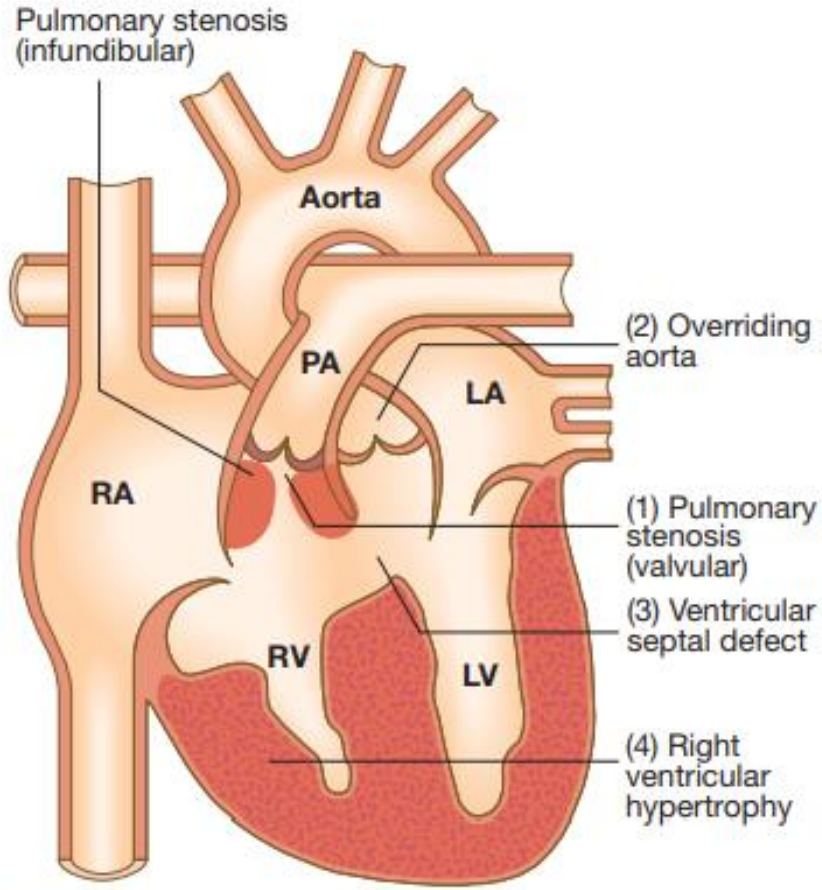
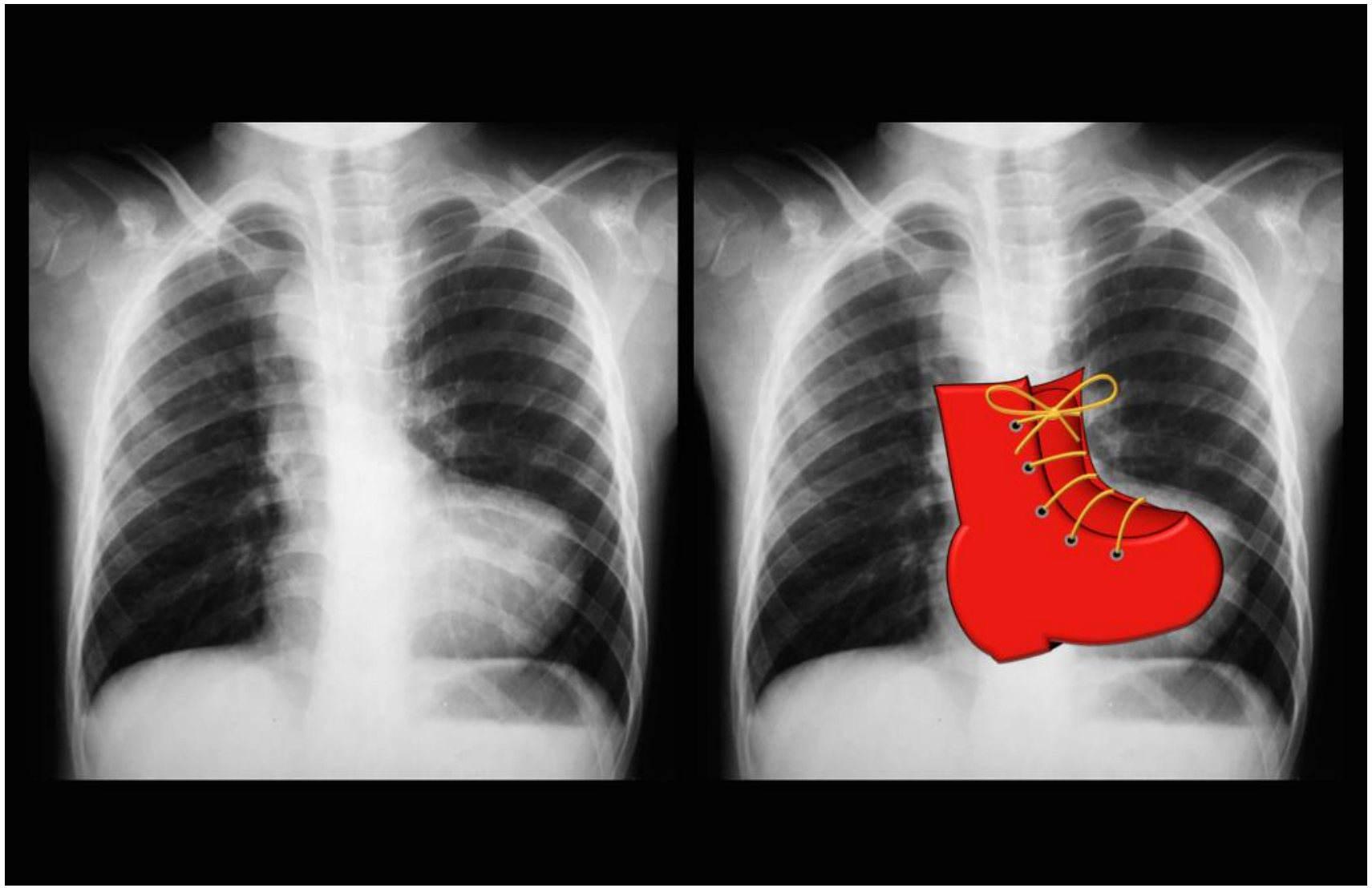


Fig. 16.98 Tetralogy of Fallot. The tetralogy comprises (1) pulmonary stenosis, (2) overriding of the ventricular septal defect by the aorta, (3) a ventricular septal defect and (4) right ventricular hypertrophy. (LA = left atrium; LV = left ventricle; PA = pulmonary artery; RA = right atrium; RV = right ventricle)

- **Clinical features**
- Children are usually cyanosed but this may not be the case 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 affected child may suddenly become 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. This is called Fallot’s sign. 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). Cyanosis may be absent in the newborn or in patients with only mild right ventricular outflow obstruction, however. This is called acyanotic tetralogy of Fallot.

- **Investigations**
- Echocardiography is diagnostic and demonstrates that the aorta is not continuous with the anterior ventricular septum.
- The ECG shows right ventricular hypertrophy and the chest X-ray shows an abnormally small pulmonary artery and a **'boot-shaped'** heart.
- **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 the age of 5 years. If the pulmonary arteries are too hypoplastic, then palliation in the form of a Blalock–Taussig shunt may be performed, with an anastomosis created between the pulmonary artery and subclavian artery. This improves pulmonary blood flow and pulmonary artery development, and may facilitate later definitive correction. 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 arrhythmias. An implantable defibrillator is sometimes recommended in adulthood.



16.102 Other causes of cyanotic congenital heart disease

Defect	Features
Tricuspid atresia	Absent tricuspid orifice, hypoplastic RV, RA-to-LA shunt, ventricular septal defect shunt, other anomalies Surgical correction may be possible
Transposition of the great vessels	Aorta arises from the morphological RV, pulmonary artery from LV Shunt via atria, ductus and possibly ventricular septal defect Palliation by balloon atrial septostomy/ enlargement Surgical correction possible
Pulmonary atresia	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
Ebstein's anomaly	Tricuspid valve is dysplastic and displaced into RV, RV 'atrialised' Tricuspid regurgitation and RA-to-LA shunt Wide spectrum of severity Arrhythmias Surgical repair possible but significant risk

(LA = left atrium; LV = left ventricle; RA = right atrium; RV = right ventricle)


THANK YOU