Congenital heart disease in infancy and childhood

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Abstract

Congenital heart disease occurs in approximately 8/1000 live births. The most common lesion at birth is a ventricular septal defect but many are small and do not need surgery. Cyanotic heart disease includes Fallot's tetralogy and transposition of the great arteries, which are both amenable to correction in childhood. More complicated cyanotic lesions are treated by separation of the systemic venous flow by a cavopulmonary connection, often referred to as a Fontan circulation. Some genetic syndromes are associated with congenital heart disease (Marfan's, Noonan's, Williams'), as are chromosomal disorders such as Down's and Turner's. DiGeorge's syndrome (thymic aplasia, hypoparathyroidism and cono-truncal cardiac defect) and velocardiofacial syndrome (palatal abnormalities, heart defects and dysmorphic features) are associated with microdeletions within the q11 region of chromosome 22.

Keywords Atrial septal defect; coarctation of the aorta; congenital heart disease; Fontan; paediatric cardiology; tetralogy of Fallot; transposition of the great arteries; ventricular septal defect

Neonatal data collection gives an incidence of significant congenital heart disease of 8/1000 live births. This does not include minor defects, which often present later in childhood or adult life (e.g. bicuspid aortic valves occur in 1/100 of the population). Congenital heart disease is even more common in the antenatal period; the discrepancy is explained by the greater incidence of spontaneous abortions and stillbirths in fetuses with congenital heart disease and, increasingly, by prenatal diagnosis and termination of pregnancy.

Specific lesions

The most common congenital heart defects at birth are shown in Figure 1.

Ventricular septal defect (VSD): the physical signs and symptoms depend on the size of the defect. Assessment is by echocardiography (using 3D for further definition of complex defects).

- Small defects are associated with a loud pansystolic murmur (reflecting a high-pressure difference between the left and right ventricle). Up to 60% close spontaneously in the first 5 years of life. If they persist, surgical closure is not usually undertaken.
- Large defects usually present with heart failure in infancy (failure to thrive, breathlessness and a history of poor feeding). There may be only a soft murmur, because large defects result in equalization of the ventricular pressures. A diastolic flow murmur can be heard across the mitral valve when pulmonary blood flow is more than twice systemic flow. Surgical closure is indicated in those who fail to respond to medical therapy or who have pulmonary hypertension with a high pulmonary blood flow (i.e. without pulmonary vascular disease). Transcatheter closure is also sometimes possible but membranous defects are usually closed with surgery because of the risk of early and late heart block.

Atrial septal defects (ASDs): ASDs seldom cause symptoms in childhood, but findings on auscultation include fixed splitting of the second heart sound, an ejection systolic pulmonary flow murmur and, sometimes, a diastolic tricuspid flow murmur. The ECG typically shows right axis deviation and partial right bundle branch block, though primum defects (see below) may have a superior (left) axis. Echocardiography demonstrates the interatrial septum clearly and in children with large shunts there may be evidence of right ventricular volume overload. ASDs occur in different positions in the atrial septum:

- Ostium secundum defects (about 70% of ASDs in infancy) result from incomplete development of the septum secundum with a defect at the site of the oval fossa. Small secundum ASDs noted in infancy may close spontaneously, but there is consensus that larger defects should be closed in childhood. Transcatheter closure is usually feasible, but large defects with deficient rims may require surgical closure.
- Ostium primum or partial atrioventricular septal defects (about 25% of ASDs) result from failure of the septum primum to reach the endocardial cushions. There are normally two atrioventricular valve orifices, but in some cases there is a common atrioventricular valve. The left-sided atrioventricular valve has three leaflets and there may be regurgitation through the 'cleft' or zone of apposition. In complete atrioventricular septal defect a ventricular communication is also present (Figure 2a & b) and, typically, there is severe heart failure in infancy. Atrioventricular septal defects are associated with Down's syndrome and almost all require surgical closure. In patients with a large 'cleft' extensive valve repair or replacement may be required.
- Failure of absorption of the sinus venosus into the right atrium causes a defect at the superior or inferior portion of the atrial septum (about 5% of ASDs). In superior defects the right upper lobe pulmonary vein usually drains into the lower part of the superior vena cava. Sinus venosus lesions all require surgical closure.

Patent ductus arteriosus (PDA): the ductus arteriosus is a normal fetal structure allowing blood flow from the pulmonary artery into the aorta (because little cardiac output passes to the lungs in the prenatal period). Closure of the duct normally occurs within a few hours of birth; persistence beyond the neonatal period is abnormal. Physical findings depend on the size of the

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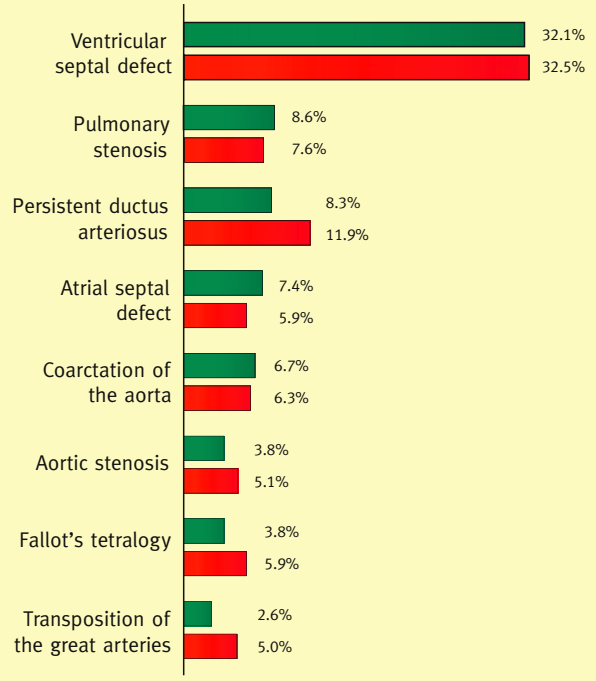
The most common congenital heart defects



These figures are taken from two classic studies. The green bars are from a USA study[1] of 56,109 births; the red bars are from a UK study[2] of 160,480 births. The lesions listed account for 70–80% of all congenital heart defects.

[1]Mitchell S C *et al. Circulation* 1971; **43:** 323–32.
[2]Dickinson D F *et al. Br Heart J* 1981; **46:** 55–62.

Figure 1

lesion: small ducts usually cause no symptoms but a continuous murmur through systole and diastole; large ducts cause cardiac failure but there may be no murmur. High pulmonary flow may cause left atrial and left ventricular enlargement. A patent duct is easily diagnosed in children using echocardiography and colour flow Doppler (Figure 3). The ECG shows an increase in left-sided voltages and there may be ST- and T-wave repolarization changes.

Intervention is usually catheter-based, unless the lesion is very large or the patient is very small, when surgical closure is preferred. Treatment of small ducts is often recommended because the risks of intervention are considered less than the risk of endocarditis. PDA is more common in premature babies when medical treatment with a prostaglandin synthesis inhibitor (indomethacin or ibuprofen) can induce duct closure. Paradoxically, patients with cyanotic congenital heart disease and some left-heart obstructive lesions can be dependent on a patent duct in the neonatal period; duct patency can be maintained with prostaglandin.

Pulmonary stenosis: this is usually valvar, but occasionally subvalvar, supravalvar or peripheral pulmonary artery stenoses are seen. On auscultation, the intensity of the murmur is related to the gradient between the right ventricle and pulmonary artery. Critical pulmonary stenosis presents in infants with cyanosis due

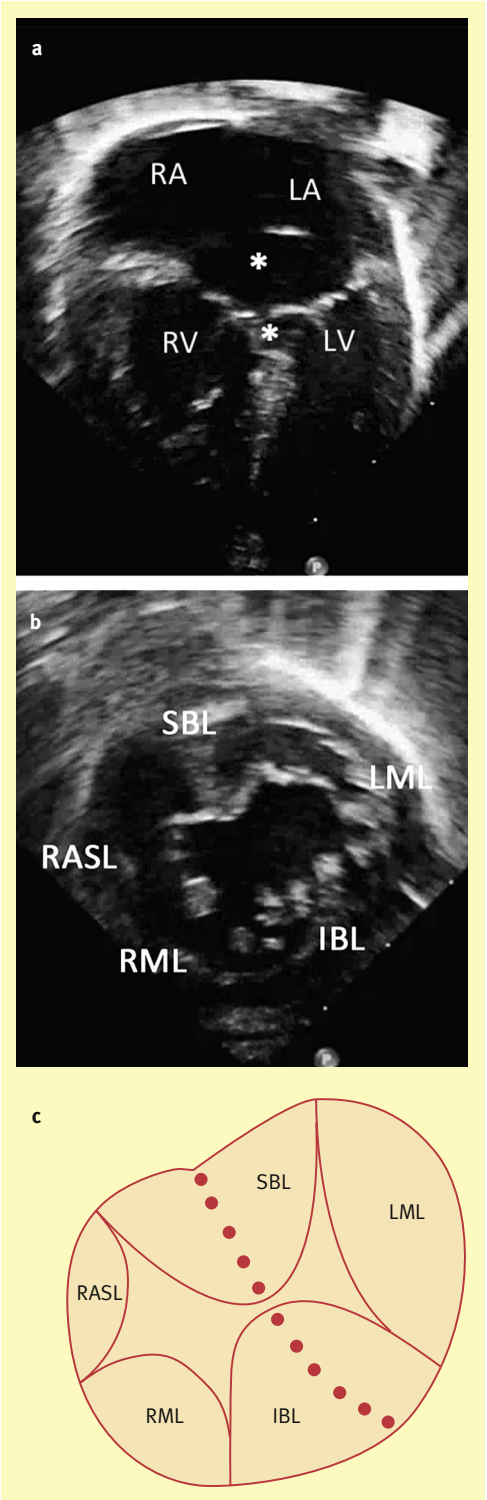


Figure 2 (a) Two-dimensional transthoracic echocardiography standard 'four chamber' view showing an atrioventricular septal defect; **(b)** Two-dimensional transthoracic echocardiography short axis view of the common atrioventricular valve **(c)** Diagram from the short axis view (SBL, superior bridging leaflet; IBL, inferior bridging leaflet, RASL, right anterior superior leaflet; LML, left mural leaflet; RML, right mural leaflet; dotted line represents the position of the interventricular septum).