Figure/table/box	Title	Source	Page number 1st Ed	Permission status
Figure 24.1	The developmental stages of a human heart beginning with the fusion of two endocardial tubes up to specific compartmentalization. Illustration of development of heart by OpenStax	OpenStax https://openstax.org/books/anatomy-and-physiology/pages/19-5-development-of-the-heart#fig-ch20_05_01	New	CC Attribution 4.0 International License.
Table 24.1	Classification of congenital heart disease by cyanosis/acyanosis, blood flow patterns and clinical findings	Author	New	Not required
Box 24.1	Cardiac shunts	Author	New	Not required
Box 24.2	Eisenmenger syndrome	Author	New	Not required
Table 24.2	ACHD AP Classification (CHD Anatomy þ Physiological Stage ¼ ACHD AP Classification)	Stout, K. K., Daniels, C. J., Aboulhosn, J. A., Bozkurt, B., Broberg, C. S., Colman, J. M., ... & Khairy, P. (2019). 2018 AHA/ACC guideline for the management of adults with congenital heart disease: a report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. *Journal of the American College of Cardiology*, *73*(12), e81-e192.	New	Permission granted 2/7/20 Request ID 600017738

Chapter 24

Congenital, Valve Abnormalities and Cardiomyopathies in Adults

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Learning Objectives

After reading this chapter, you should be able to:

- Describe major congenital cardiac abnormalities
- Describe foetal blood flow and relationship to heart defect
- Discuss the anatomy and pattern of blood flood flow leading to cyanotic and acyanotic congenital heart disease
- Discuss specific health issues relating to adults with congenital heart disease
- Discuss the need for ongoing monitoring and follow-up for adults with congenital heart disease

Key Concepts

Congenital heart disease; genetic heart disease; cyanotic and acyanotic heart disease; cardiac shunt; Eisenmenger syndrome

Overview

This chapter will introduce the formation and management of congenital and genetic heart defects, valve abnormalities and genetic cardiomyopathies. Strictly speaking *congenital* refers to defects occurring at birth but in common use the term congenital describes defects that occur during fetal development, at birth, or immediately after. Congenital heart disease is a complex sub-specialty of cardiac care. This chapter is presented as an introductory view to congenital heart disease, focusing on the continued needs of the congenital heart disease population in adulthood.

Background

Congenital heart defects are a heterogeneous group of abnormalities, ranging in severity and associated outcome. Critical cases of CHD may be incompatible with survival without specific

intervention in the newborn period/early infancy whilst other forms may not require intervention until adulthood, while milder forms may not require any intervention at all (Patel et al. 2015). In some instances, an anomaly may be identified antenatally through screening, but others are not identified until after birth.

Epidemiology

Congenital heart disease (CHD) refers to a structural abnormality of the heart and great vessels present at birth that is, or could be, of functional significance. It is the most common birth defect and affects approximately1% of all liveborn infants. However, significant geographical variations exist, influenced by intrinsic maternal and environmental factors, as well as extrinsic factors such as access to antenatal screening, diagnostics, and wider policies and laws (for example, options for termination) (Knowles et al., 2017).

Before the era of cardiac surgery, a serious congenital defect would usually result in death. Improved diagnostic techniques, as well as medical and surgical interventions, have resulted in 10-year survival rates exceeding 80%, even in complex cases of CHD (Schwerzmann et al., 2017). Nonetheless, CHD remains the leading cause of mortality from birth defects in the developed world, and in addition is associated with significant cardiac and extracardiac comorbidities, in particular neurodevelopmental disabilities, that affect quality of life,(Zaidi & Brueckner 2017).

Antenatal detection of severe congenital heart defects is associated with a reduction in mortality and morbidity (Thakur et al., 2016, Van Velzen et al., 2015). In some instances, parents-to-be may make the decision to terminate the pregnancy (Smith et al., 2011), whilst for others, antenatal detection provides the opportunity to optimise follow up and make plans for appropriate birthing procedures.

Key Point	Improved detection and management of CHD means that there are now more adults than children living with CHD (Zaidi & Brueckner 2017).

Risk factors for the development of congenital heart disease

Various risk factors are associated with the development of a congenital heart defect. Many cases of CHD are multifactorial and result from a combination of genetic predisposition and environmental risk factors (Ossa Galvis et al. 2020). Maternal factors that have been associated with CHD include pregestational diabetes, phenylketonuria, obesity, exposure to smoking, al-

cohol and some drugs, infections such as rubella, whereas periconceptional folic acid supplementation or fortification has been found to have a protective effect ((Morris et al., 2018)Roos-Hesselink & Johnson 2017; Dolk et al. 2020).

In addition,gene defects, chromosomal disorders, environmental factors or infections, teratogens or micronutrient deficiencies have been identified as contributory factors. However, in around 80% of cases but the anomaly cannot be attributed to a specific cause (Blue et al., 2012, Feldkamp et al., 2017).

Key Point	The vast majority of fetal heart defects are seen in families without a known risk factor for CHD, which highlights the importance of having an effective screening program to detect fetal heart disease (Roos-Hesselink & Johnson 2017).

A number of CHD lesions occur alongside underlying genetic syndromes. These include Down syndrome (the most common chromosomal cause of CHD), Turner syndrome, Klinefelter syndrome, Noonan syndrome, Williams syndrome, DiGeorge syndrome (velocardiofacial syndrome), and Holt-Oram syndrome (Stout et al. 2019).

Suggested resource website	If you would like to know more about genetic congenital heart disease, read the following article: Pierpont, M. E., Brueckner, M., Chung, W. K., Garg, V., Lacro, R. V., McGuire, A. L., ... & Ware, S. M. (2018). Genetic basis for congenital heart disease: revisited: a scientific statement from the American Heart Association. *Circulation*, *138*(21), e653-e711. https://doi.org/10.1161/CIR.0000000000000606

Embryology

The heart begins as a flat sheet of cells near the head of the embryo See Figure 24.1. By day 19 after conception, these cells form into two *endocardial tubes* with endothelial, myocardial and pericardial layers. Around day 22, these endocardial tubes merge into a single tube that forms the *primitive heart tube*. This tube has five regions from top to bottom: *truncus arteriosus*, *bulbus cordis*, *primitive ventricle*, *primitive atrium*, and *sinus venosus.* The heart develops

by looping, ballooning and reforming these regions into atrial, ventricular and outflow tract areas, which then differentiate into the left and right heart structures seen in the mature heart (Moorman et al., 2003).

In the foetus, the heart is the first organ to begin functioning and beats by day 21 or 22 with the formation of the primitive heart. Cardiac cells (*myocytes*) spontaneously contract by electrical depolarisation. Specialised conducting tissue develops in the *primitive atrium* and *sinus venosus* into the *sinoatrial* and *atrioventricular* nodes respectively. The foetal heart rate starts at a similar rate to the mother's but then increases in the second trimester before decelerating to the 120-160 bpm typically seen at birth (von Steinburg et al., 2013).

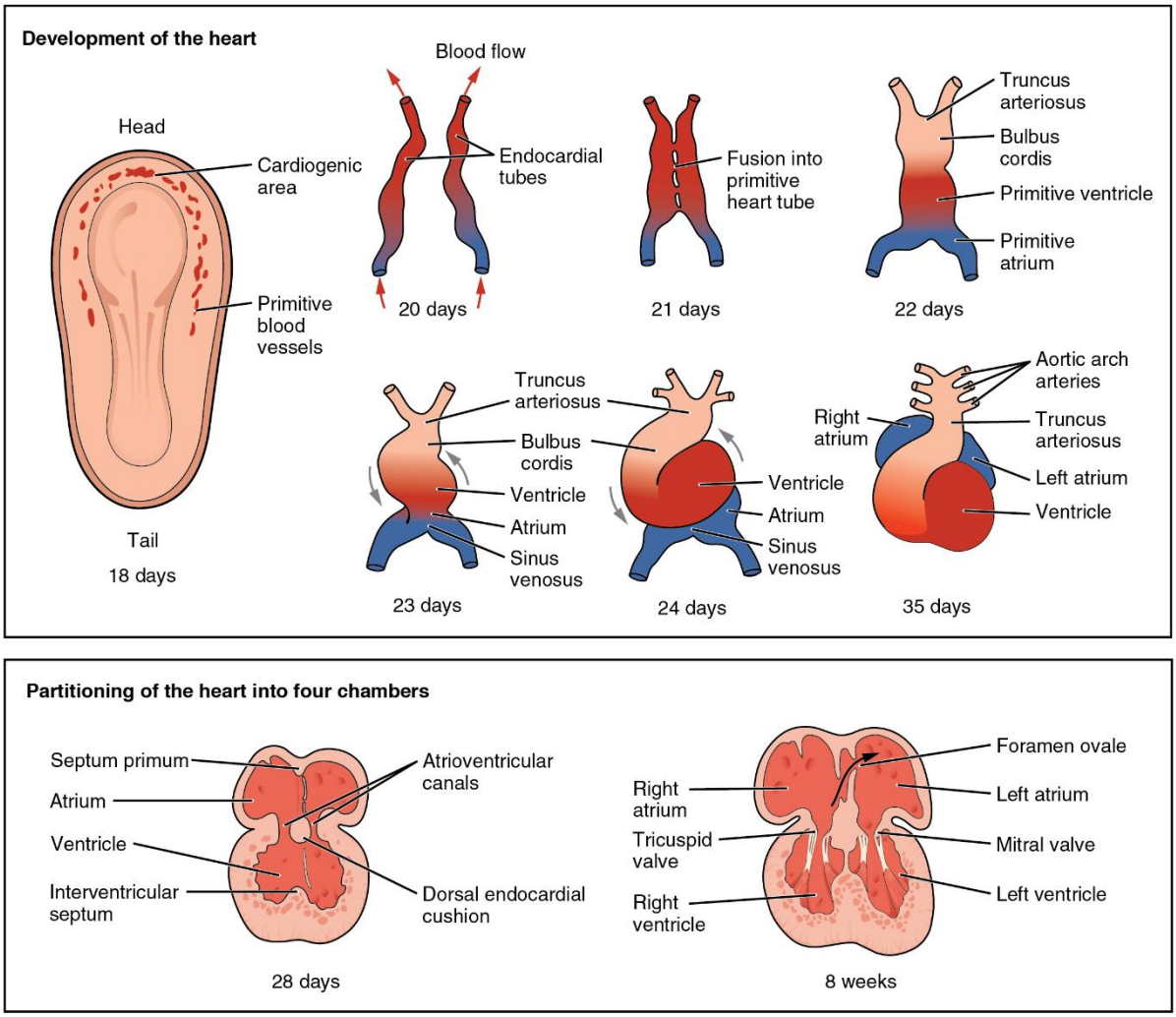

Figure 24.1 The developmental stages of a human heart beginning with the fusion of two endocardial tubes up to specific compartmentalization. Illustration of development of heart by OpenStax is licensed under a CC Attribution 4.0 International License. Accessed from https://openstax.org/books/anatomy-and-physiology/pages/19-5-development-of-the-heart#fig-ch20_05_01

The walls of the heart form between day 27 and 37 as tissue grows and merges. In the atrioventricular area the growth of the endocardial cushions forms the atrioventricular valves and the tracts for the aortic and pulmonary channels. In the atria, tissue growth leads to the closing of the original hole between the atria (ostium primum) and formation of a second hole (ostium secundum) with a slit (foramen ovale) through which blood can continue to flow from right to left during foetal circulation. Foetal gas exchange occurs in the placenta. Oxygenated blood travels through the umbilical vein into the inferior vena cava through the ductus venosus and bypasses the liver circulation. Oxygenated blood is shunted from the right atrium to the left atrium through the foramen ovale and then blood pumped to the left ventricle and into the aorta to reach systemic circulation. Only a small portion of blood is pumped from the right atrium to right ventricle and pulmonary artery (PA) where it is shunted to the aorta through the ductus arteriosus thereby bypassing the lungs. Deoxygenated blood returns to the placenta via the umbilical arteries (Ossa Galvis et al. 2020). At birth, blood flow through the placenta stops and respiration begins, in normal circumstances, with a rise in right atrial pressure closing the foramen ovale and the ductus arteriosus beginning to close.

Patent Foramen Ovale (PFO)

In around 25% of the population the foramen ovale fails to close (Homma et al., 2016). This anomaly is referred to as a patent foramen ovale. Of these, around 30% will be transient where the PFO is fused, opening only during high pressure activities such as coughing. In these patients, no left to right shunt is visible on imaging. Patients with PFOs are usually asymptomatic, and therefore may not be diagnosed until later in life, when they present with neurological problems such as strokes, transient ischaemic attacks and migraines. Medical therapy subsequently may include antiplatelet or anticoagulant medication (Zhang et al., 2018). Where patients are symptomatic, PFOs can be closed using percutaneous devices or through open-heart surgery.

Suggested resource website	For a more comprehensive information about fetal development of the cardiovascular system, see the suggested resource below: Development of the cardiovascular system Teach Me Anatomy URL: https://teachmeanatomy.info/the-basics/embryology/cardiovascular-system/

Congenital heart disease classification

Classifying CHD is complex, with crossover between categories of classification, particularly in cases where patients present with different combinations of anomalies.0020At least 18 distinct types of CHDs have been recognised, with many additional anatomic variations (American Heart Association [AHA] 2020). CHD is traditionally classified according to the presence or absenceof cyanosis,. Cyanotic heart disease can be subdivided into conditions with decreased pulmonary blood flow and conditions with mixed blood flow. The most common lesions include tetralogy of Fallot and transposition of the great arteries. (see Box 24.1) Management of hypoxia is a clinical priority in this group of patients.

Acyanotic CHD includes congenital heart defects with dominant left-to-right shunt and obstructive lesions. The most common lesions include Atrial or Ventricular Septal defects (ASD or VSD). Congestive cardiac failure is a significant risk in this group of patients.

Key Point CHD lesions can be categorized as cyanotic or acyanotic. Cyanosis occurs when deoxygenated blood bypasses the lungs and enters the systemic circulation resulting in low saturations and commonly a 'blue' colour. Approximately 25% of cases of CHD are cyanotic lesions (Ossa Galvis et al. 2020).

Cyanotic lesions can be further characterised based on theblood flow patterns through the heart. This makes it easier to predict clinical manifestations. Congenital heart anomalies according to classification, blood flow patterns and clinical findings are shown in Table 24.1. Using this classification system, the clinical presentation and management of the most commonly encountered congenital heart defects are outlined in this chapter.

Table 24.1 Classification of congenital heart disease by cyanosis/acyanosis, blood flow patterns and clinical findings

	Acyanotic		**Cyanotic**	
Blood flow pattern	Increased pulmonary blood flow (left to right shunt)	Obstruction to blood flow from ventricles	Decreased pulmonary blood flow	Mixed blood flow
Anomaly	• ASD • VSD • PDA	• Valvular AS • CoA • IAA • PVS	• TOF • Tricuspid atresia • Ebstein anomaly	• TGA • TAPVR • Truncus arteriosus

	• Complete AV canal defect			• Hypoplastic left heart syndrome
Clinical findings	• ↑ PVR • ↑ right heart pressures • PH • HF • Reversal of shunt • ES (see Box 24.1)	• Left side: HF • Right side: cyanosis	• Cyanosis	• Variable based on the degree of mixing and amount of PBF. • Hypoxemia (with or without cyanosis) and HF usually occur together.

PVR = pulmonary vascular resistance; PH = pulmonary hypertension; HF = heart failure; ES = Eisenmenger syndrome; ASD = atrial septal defect; VSD = ventricular septal defect; PDA = patent ductus arteriosus; AV = atrioventricular; AS = aortic stenosis; CoA = coarctation of the aorta; IAA = interrupted aortic arch; PVS = pulmonic valve stenosis; TOF = Tetralogy of Fallot; TA = tricuspid atresia; TGA = transposition of the great arteries; TAPVR = total anomalous pulmonary venous return

Box 24.1 Cardiac shunts

Fetal circulation

Shunts are crucial during fetal life as the placenta provides the exchange of gases and nutrients. Fetal circulation has four sites of shunting: the placenta, ductus venosus through which umbilical vein drains into inferior vena cava, foramen ovale within the interatrial septum, and the arterial duct through which blood in the PA flows into descending aorta. Shortly after birth, the placental circulation disappears, and pulmonary circulation is established. Interruption of the umbilical cord results in an increase in systemic vascular resistance and closure of the ductus venosus (Micheletti 2019).

Circulation following birth

Normal function of the heart involves the flow of blood from the atria to the respective ventricle and subsequently around the body or to the lungs. Pressures in the left side of a normal heart are persistently higher than those in the right. Patterns of blood flow that deviate from this normal circulation are called cardiac shunts. These may occur from left to right, right to left, or bidirectional. The direction of the shunt is determined by pressure in the respective chamber, with blood shunted from areas of high pressure to low pressure. In a normal heart,

the left heart pumps blood into circulation and therefore is a higher-pressure system. Conversely, the right heart has a smaller muscle mass and lacks the central constricting muscle fibres that are responsible for generating the force of contraction and is a lower-pressure system (Sheehan and Redington, 2008). Therefore, shunt direction affects the status of pulmonary blood flow which can vary from normal, increased, or decreased (Micheletti 2019).

CHD with shunt between systemic and pulmonary circulation

In conditions with left-to-right shunt, blood from the systemic arterial circulation mixes with systemic venous blood. The extent of flow through the shunt and its physiologic effects are impacted by multiple factors. An abnormal connection between the systemic and pulmonary circulations gives rise to potential excess volume of blood to flow from the systemic (left side) circulation to the pulmonary circulation (right side) resulting in the recirculation of already oxygenated pulmonary venous blood through the pulmonary vasculature. Left-to-right shunting can occur due to communications at the atrial, ventricular, and arterial level, with blood preferentially shunting from a higher to lower resistance circulation (Kung & Triedman 2019). The clinical presentation of a patient with left-to-right shunt depends on the size of the shunt, the physiological effects of the shunt and presence of other cardiac anomalies. Any large left-to-right (L-R) shunt results in increased blood flow into the lungs associated with shortness of breath and prominent vascular markings on chest x-ray, volume overload of left ventricle (LV) associated with chamber dilatation, and subsequently heart failure (Micheletti 2019). The most common CHD causes of left to right shunt include atrial septal defect (ASD), ventricular septal defect (VSD), patent ductus arteriosus (PDA), and atrioventricular septal defect (also known as atrioventricular canal defect). When pressure in the right side of the heart becomes higher than that in the left, Eisenmenger syndrome may develop (see Box 24.2).

Box 24.2. Eisenmenger Syndrome

Eisenmenger syndrome (ES) is a rare progressive disorder. Whilst an increasing number of CHDs are diagnosed antenatally or soon after birth, complications can arise where an anomaly is not identified early. ES refers to any untreated congenital cardiac defect with intracardiac communication that over time leads to pulmonary hypertension (PH), reversal of flow, and cyanosis. In the normal heart, the left side of the heart generates high pressure to supply the extensive high resistance systemic circulation. The right side of the heart generates much lower pressures to allow blood to pass through the low resistance and high

compliance pulmonary circulation. In the case of a defect that allows communication between the two sides of the heart, a shunt will develop causing blood to flow from the area of high pressure to lower pressure (left to right). The amount of blood shunted is proportional to the size of the defect. The abnormally high and pressure of blood directed through the shunt from the left side of the heart to the right side damages the pulmonary vasculature causing scar tissue and ultimately PH. The right side of the heart has to work harder to supply blood to the lungs, leading to hypertrophy of the right ventricle (RV) and increased right heart pressures. Once right heart pressures exceed those of the left side of the heart, the left to right shunt will reverse and deoxygenated blood returning to the right side of the heart will be shunted to the left, bypassing the lungs, and pumped to the systemic circulation leading to cyanosis and organ damage. The kidneys, sensing the decrease in oxygen saturation, try to compensate by increasing production of erythropoietin and red blood cells, leading to an increase in reticulocyte count and the risk of hyperviscosity syndrome. As reticulocytes are immature blood cells, they are not as efficient at carrying oxygen and changing shape compared with mature cells, and are unable to transit easily through capillaries, leading to death of capillary beds. Patients with ES are at risk of both blood clots due to hyperviscosity and bleeding from their damaged lung capillaries. Clinically, ES manifests in cyanosis, desaturation, dyspnoea, syncope, and clubbing (Dakkak & Oliver 2019).

The natural history of ES is variable and depends upon the complexity of lesions but it is typical for complications to start in the patient's third decade with high mortality in the third and fourth decades. Ventricular failure, haemoptysis, pregnancy complications, and strokes are common causes of death (Basit et al 2020). Surgical repair is usually possible before PH becomes too high. Once severe PH is established, pulmonary vasodilators may help symptoms (D'Alto and Diller, 2014). Surgical transplant of the lungs or heart-lungs is an option but usually only a viable treatment for a small number of people (Le Pavec et al., 2018).

Acyanotic CHD

Acyanotic CHD with increased blood flow

Atrial Septal Defect (ASD)

An atrial septal defect (ASD) is the most common form of CHD found in adults. Most ASDs occur by chance, though familial transmission has also been reported. In the fetal circulation

there is normally an opening between the left and right atria to allow blood to bypass the lungs. This opening usually closes about the time the baby is born, but if the opening persists it is called an ASD. Around 90% of ASDs occur in the central portion of the atrial septum and are referred to as secundum ASD (ASD II). An ASD may be managed conservatively or require closure using surgical or percutaneous device implantation.

ASD in adults

Patients with an isolated ASD often remain asymptomatic during childhood and adolescence, but supraventricular arrhythmias, right ventricular dysfunction, and PH increase with age, while exercise tolerance and life expectancy are reduced (Kuijpers et al. 2015). Conservative management includes reducing thromboembolic risks with anticoagulation or antiplatelet drugs. Risk of complications with ASD closure increases with age.

Key Point	Where the atrial shunt has reversed the sudden closure of the ASD can lead to the right ventricle having to overcome a high PA pressure and this may result in HF.

Suggested resource Reading	Oster, M., Bhatt, A. B., Zaragoza-Macias, E., Dendukuri, N., & Marelli, A. (2019). Interventional therapy versus medical therapy for secundum atrial septal defect: a systematic review (part 2) for the 2018 AHA/ACC guideline for the management of adults with congenital heart disease: a report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. *Journal of the American College of Cardiology*, *73*(12), 1579-1595. URL: https://www.onlinejacc.org/content/accj/73/12/1579.full.pdf

Ventricular Septal Defect (VSD)

Ventricular septal defect (VSD) refers to a hole in the septum dividing the right and left ventricle that occurs in the embryonic stage of heart development. It is the most common congenital cardiac anomaly in children, and the second most common congenital abnormality in adults (Dakka & Oliver 2020; Mavroudis, Dearani & Anderson 2020). Several genetic factors including Down syndrome have been identified to cause VSD, but non-inheritable factors have also been implicated. VSD may occur in isolation but can occur in association with other congenital heart defects and are a frequent component of complex CHD.

VSDs are classified according to location and size. Most VSDs are restrictive (<5 mm) and undergo spontaneous closure during the first year of life (Mavroudis et al. 2020). Small congenital VSDs may not need treatment but if a large VSD is not repaired children are likely to develop pulmonary vascular obstructive disease as early as 18 months to 2 years of age (Rao & Harris 2018). Management must be considered in the context of VSD size, location, coexisting congenital heart defects and clinical findings.

VSD in adults

Adult patients with restrictive VSDs have about a 10% spontaneous closure rate during any decade of life, but there is a 25% chance of developing complications relating to the uncorrected VSD, including infective endocarditis (see Box 24.3), aortic regurgitation, and symptomatic arrhythmias. Surgery is necessary if the VSD is large or associated with significant complications (Mavroudis et al. 2020). Where the position of the VSD allows, percutaneous device VSD closure will be undertaken. In addition those in whom surgery is very risky due to severe PAH or multiple comorbidities, percutaneous closure may be offered. . Complications of surgical closure include residual or recurrent VSD, valvular incompetence such as tricuspid regurgitation and aortic insufficiency, arrhythmias (atrial fibrillation, complete heart block and uncommonly, ventricular tachycardia), LV dysfunction and progression of pulmonary hypertension (PH).

Key Point	Lifelong follow-up is recommended for patients with VSD whether they have had corrective surgery or not, as there is a measurable incidence of major complications during late follow-up of adult patients with restrictive VSDs regardless of operative status (Mavroudis et al. 2020).

Patent Ductus Arteriosus (PDA)

The *ductus arteriosus* is a foetal vessel that allows oxygenated blood from the placenta to by-pass the lungs in utero (Gillam-Krakauer & Mahajan 2020). At birth, as lungs fill with air and normal circulation begins, the ductus arteriosus is redundant and functionally closes within 12-24 hours of birth, with permanent anatomic closure becoming complete within 2-3 weeks. 98% of infants will have a closed ductus by the time they are discharged from hospital. Preterm infants weighing less than 1000 grams at birth are most at risk of PDA. Conservative management of a PDA with fluid restriction, supportive peak end-expiratory pressure to treat pulmonary oedema and indomethacin, ibuprofen, or acetaminophen/paracetamol increasing peak expiratory may be sufficient to support the infant while awaiting spontaneous closure. In more

severe cases, a PDA can result in blood flowing from the descending aorta across the PDA into the pulmonary circulation (left to right shunt) resulting in pulmonary oedema and pulmonary hemorrhage, intraventricular hemorrhage, congestive heart failure, bronchopulmonary dysplasia, with decreased blood flow to the lower body leading to renal failure and necrotising enterocolitis (Gillam-Krakauer & Mahajan 2020). PDAs are closed either using an open-heart surgery technique or, more commonly, percutaneously using a coil or plug (Baruteau et al, 2014). After closure of the PDA, most children have a normal life expectancy (Gillam-Krakauer & Mahajan 2020).

PDA in adults

PDA is rare in adults as symptoms and signs usually occur in infancy and most cases are treated shortly after diagnosis. Untreated PDA can lead to PH, Eisenmenger syndrome, HF and endarteritis. In adults, closure of a PDA is required if the patient has not developed fixed PH. In general, PDA's are closed by transcatheter intervention. Before the ductus is closed in an adult, the status of the pulmonary vasculature must be determined. Those with only mild PH have a normal life expectancy (Gillam-Krakauer & Mahajan 2020).

Atrioventricular septal defect (atrioventricular canal defect; endocardial cushion defect)

Atrioventricular septal defect (AVSD) accounts for around 5% of congenital heart anomalies. An AVSD may be complete, with a large non-restrictive inlet ventricular septal defect (VSD); transitional with a small or moderate-sized restrictive VSD, or partial with no VSD. Patients with no VSD component or a small VSD and good AV valve function may be asymptomatic. In the case of a large VSD or significant AV valve regurgitation, patients often have signs of HF (Baffa 2018a). A complete AVSD (also called a complete common AV canal defect) may result in left-to-right shunt at the atrial and ventricular levels. AV valve regurgitation sometimes causes a direct left ventricle-to-right atrial shunt. Over time, Eisenmenger syndrome may develop if not corrected (see Box 24.1). Complete AVSD heart failure is evident in an infant between 4-6 weeks old and requires surgical repair by age 2-4 months old. Symptoms in partial AVSD vary with the degree of mitral regurgitation: if mild or absent, symptoms may develop during adolescence or early adulthood (Baffa 2018a).

AVSD in adults

Patients with milder disease may present in late childhood or early adulthood with congestive heart failure, PH with Eisenmenger syndrome, infective endocarditis, or arrhythmia. Surgical repair is indicated unless there is proven irreversible PH in which case medical management for PH with pulmonary vasodilators should be attempted (Kochav 2018).

Key Point	Most patients with the complete form of AVSD have Down Syndrome (Baffa 2018a).

Acyanotic CHD with outflow obstruction

This group of congenital heart defects all present with an obstruction to the outflow tract. A ventricular outflow tract is the section of left ventricle or right ventricle through which blood passes in order to reach the great arteries. Conditions associated with acyanotic obstructive CHD include aortic valve stenosis (AVS), coarctation of the aorta, interrupted aortic arch and pulmonic stenosis (PS).

Aortic Valve Stenosis

Aortic stenosis (AS) may be congenital or acquired. In paediatric patients AS is almost always congenital. Congenital valvular AS (CVAS) accounts for approximately 3–6% of congenital heart defects and may be associated with other forms of CHD including patent ductus arteriosus, coarctation of the aorta and ventricular septal defects (Singh 2019). CVAS displays several morphologic types of abnormal valves: unicuspid, bicuspid, tricuspid and quadricuspid, and is a spectrum in which the degree of obstruction ranges from mild to severe. Clinical findings depend upon the age of the patient at the presentation, severity of the CVAS and the presence of associated cardiac lesions. The majority of infants with severe CVAS present with progressive congestive heart failure (HF) by 2 months of age. Older children and adolescents are usually asymptomatic but symptoms of dyspnoea, angina or syncope (particularly during exercise) should be investigated as there is a risk of sudden death in in children aged 5-15 years with moderate to severe CVAS (Singh 2019). CVAS is a progressive disorder and management is determined by the age of the patient at presentation, the severity of the obstruction and adequacy of left heart structures. Current therapeutic intervention options to relieve LVOT obstruction are percutaneous balloon aortic valvuloplasty, surgical aortic valvotomy and valve replacement (Singh 2019).

Congenital AVS in the adult

Congenital aortic stenosis is a lifelong disease that often requires multiple interventions. Presentation in adults is most commonly associated with a bicuspid valve, or, less commonly, following previous intervention as a neonate or infant. Balloon aortic valvuloplasty is the intervention of choice in many centres for neonates or infants, as it is seen to be a safer and a less invasive alternative to surgery. , (Donald et al. 2019). However, this is rarely performed in

adults. Those who have had prior intervention, or surgical repair of replacement of the AV, may present symptoms resulting from patient-prosthesis mismatch due to the normally functioning valve prosthesis being too small in relation to the patient's body size, leading to high transvalvular pressure gradients (Dahou et al. 2016).

Key Point	The bicuspid aortic valve is the most common cause of aortic stenosis in patients less than the age of 70 years in developed countries (Pujari & Agasthi 2020). Aortic aneurysm formation and aortic dissection are the two major complications of bicuspid aortopathy, mostly encountered beyond pediatric age (Singh 2019).

Coarctation of the aorta

Coarctation of the aorta (CoA) accounts for 6 to 8% of congenital heart anomalies. No single cause has been identified, although evidence suggests a combination of genetic, environmental and haemodynamic factors. CoA involves a localised narrowing of the aortic lumen that results in upper-extremity hypertension (HT), left ventricular hypertrophy (LVH), and impaired perfusion of the abdominal organs and lower extremities (Baffa 2018b). It can occur in different sites in the aorta but usually occurs just beyond the branch of the left subclavian artery. The aorta can bulge either side of the narrowing and the subclavian artery can be dilated (Suradi and Hijazi, 2015). CoA may occur alone or with various other congenital anomalies including bicuspid aortic valve, VSD, AS, patent ductus arteriosus and mitral valve disorders (Baffa 2018b). Neonates may present in shock and require prostaglandin E_1 to maintain ductal patency until the time of surgical repair. Older children usually present with upper extremity hypertension or murmur. Surgical repair in neonates and infants depends on the presence or absence of concomitant defects and the degree of associated arch hypoplasia (Nelson, Stone & Gamgeni 2019). Treatment is balloon angioplasty with stent placement, or surgical correction (Baffi 2018b).

Adults with CoA

CoA is commonly treated after birth or during childhood and is rarely seen in adults. For those identified in adulthood, treatment in the form of a covered stent to the aortic coarctation. Untreated CoA has a poor long-term prognosis, with a 75% mortality by the time the patient reaches their mid-forties (Suradi and Hijazi, 2015). Adults may present with a history of a previous coarctation procedure, rupture of an old repair, heart failure, aortic aneurysm, aortic

dissection, undersized grafts of previous repairs, intracranial hemorrhage, hypertension with exercise, and infections (Shah & Shah, 2018).

Key Point	CoA is more common in females than males (2:1 ratio) and occurs in 10-20% of patients with Turner syndrome (Baffa 2018b).

Interrupted Aortic Arch

Interrupted aortic arch (IAA) accounts for approximately 1.5% of all CHD. IAA differs from a coarctation because the aorta is not narrowed but discontinuous at a location between the ascending and descending aortic arch. IAA is associated with other cardiac anomalies, commonly VSD, PDA, and bicuspid aortic valve. In IAA lower body perfusion is entirely PDA-dependent and in the absence of prenatal diagnosis, patients with IAA present in shock within the first few days to weeks of life when the PDA closes. PGE_1 infusion is usually given to keep the arterial duct open, and inotropic agents, diuretics and mechanical ventilation may be required to stabilise the patient in preparation for surgery. Mortality rate for patients with IAA is estimated to be 3.6% at 2 years and about 39% at 21 years (Micheletti 2019).

Adults with interrupted aortic arch

IAA is associated with a significant burden in terms of the need for reintervention and deficits in exercise performance, health status and health-related quality of life (Micheletti 2019). Lifelong follow-up is needed for patients with repaired IAA, primarily to monitor for left ventricular outflow tract obstruction and recurrent aortic coarctation (Friedman 2018).

Pulmonary valve stenosis

Pulmonary valve stenosis (PVS) with normal cardiac connections accounts for 8-12% of all CHDs (Micheletti 2019; McCarthy et al., 2019) and is the most common cause of RV outflow tract obstruction (RVOTO). Concomitant presence of right ventricular hypertrophy (RVH) is dependent on the significance of the stenosis. PVS may occur as an isolated lesion but can coexist with other congenital heart lesions – notably it is one of the four cardinal features of tetralogy of Fallot (TOF). Isolated mild PVS is usually asymptomatic, and the probability of long-term survival is similar to the general population. Patients with severe PVS most often present during childhood with RV failure and cyanosis, most often in the presence of interatrial shunting (Fathallah & Krasuski 2017). Neonates with critical stenosis typically present with central cyanosis at birth. PVS may be recognised in infants and children by the presence of ejection murmurs auscultated in the pulmonic area. Symptoms of PVS progress with time. Newborns with critical PVS

need emergency treatment including PGE_1 infusion and cardiac catheterisation for percutaneous pulmonary when stabilised. If percutaneous balloon dilation is unsuccessful, surgical pulmonary valvotomy is urgently indicated in critically ill patients (Micheletti 2019). If PVS is successfully treated (balloon pulmonary valvuloplasty or pulmonic valve replacement, prognosis is generally excellent (Fathallah & Krasuski 2017). Treatment decisions are highly dependent on concomitant disease.

Adults with PVS

PVS is most often associated with the failure of the valvular leaflets to fuse and is clinically detected at different stages of life. The more severe the obstruction, the earlier the valvular abnormality is typically detected. Adults present with symptoms of congestive HF and RVOT that is progressive in nature (Loewenthal 2019).

Key Point	Patients with Noonan's syndrome are often resistant to balloon dilatation due to the associated pulmonary trunk narrowing (Bellsham-Revell and Burch, 2018). Surgery is usually the first line treatment for this group.

Cyanotic CHD

Cyanotic CHD with decreased pulmonary blood flow

Tetralogy of Fallot

Tetralogy of Fallot (TOF) occurs in 10% of all CHD (Micheletti 2019) and is the most common cyanotic heart defect (McCarthy et al., 2019) occurring in 3 to 5 of every 10,000 live births (Diaz-Frias & Guillaume 2020). It is a complex condition defined by the presence of four conditions: (1) large malaligned VSD; (2) pulmonary stenosis causing RVOT obstruction; (3) RV hypertrophy resulting from RVOT obstruction; and (4) an over-riding aorta above the VSD which allows blood from both ventricles to enter the aorta (Micheletti 2019). The degree of patient cyanosis depends upon the degree of RVOT obstruction which is variable. "Pink TOF" is associated with mild pulmonary stenosis and has a similar presentation to that of VSD with high pulmonary blood flow, normal oxygen saturation, and potential CHF symptoms early in life. All other forms of TOF may experience characteristic hypoxic or hypercyanotic spells that typically reach peak incidence in infants between two-four months of age. Immediate recognition and treatment is required to avoid severe complications including death.

TOF is typically treated by open heart surgery in the first year of life. The procedure involves reconstruction of the RVOT and repairing the VSD. Most patients undergoing surgery

have a good long-term outcome, but residual post-surgical defects may affect life expectancy and increase the need for reoperation. Left untreated, TOF carries a 35% mortality rate in the first year of life, and a 50% mortality rate in the first three years of life (Diaz-Frias & Guillaume 2020).

Adults withToF

Adults with repaired ToF develop late complications, such as progressive exercise intolerance, arrhythmias, and heart failure (Dennis et al., 2017).These complications are mainly due to consequences of the operation, in particular pulmonary regurgitation, leading to right ventricle dysfunction (Babu-Narayan and Gatzoulis, 2018) Due to the increased survival rates of ToF patients, PV replacement is now becoming one of the most common ACHD surgeries performed, although the timing of the procedure remains controversial (Dorobantu et al., 2020)

Although rare, there are a small number of patients who present with untreated ToF in adulthood, These patients appear to have a unique haemodynamic balance that provides protection (Chaudhry et al., 2017). In a study of 30 patients who had total correction of ToF between the ages of 40 to 60 years, long term survival rate at 5 years and 10 years postoperatively of 92% and 74% respectively (Hu et al., 1985)

Tricuspid Atresia

Failure of the tricuspid valve to grow during foetal development results in the right side of the heart developing without a connection between the atrium and ventricle. Whilst the left side of the heart functions normally, there is an underdeveloped right ventricle. Tricuspid atresia is a relatively rare anomaly, occurring in about two out of every 10000 live births, and accounting for around 1-2% of all cases of congenital heart disease (Murthy et al., 2019). Like many congenital cardiac conditions, tricuspid atresia is often associated with a series of additional defects in various combinations and with varying degrees of severity. ASD or VSD commonly occur alongside tricuspid atresia. This enables blood to shunt between the left and right side of the heart, allowing blood to mix, and therefore provide oxygen to the systemic circulation. Tricuspid atresia may also be classified as a univentricular (single ventricle) heart (discussed later in this chapter).

Ebstein Anomaly

Ebstein anomaly (EA) accounts for <1% of all CHD (Micheletti 2019) and is rare, with a prevalence of around 1 in 20 000 live births (Boyle et al., 2018). Patients with EA have an abnormal tricuspid valve (TV) with malformed leaflets leading to tricuspid regurgitation. The valve is

also malpositioned, sitting lower in the RV than in normal anatomy. In addition, the right atrium (RA) is large, and attached to a small RV. As with all CHD, this anomaly presents on a spectrum of severity depending on the precise anatomy (Geerdink et al., 2017). In some instances, the severity of the tricuspid regurgitation leads to raised pressures in the right atrium. This results in a right to left shunt across the PFO after birth, preventing the PFO closing, with deoxygenated blood then entering the systemic system. As a result, the baby may become cyanotic. Anomalies associated with EA include an ASD and less commonly a VSD or PDA. Complex forms of EA may be associated with pulmonary valve stenosis or atresia, tetralogy of Fallot, or left-sided abnormalities such as mitral valve stenosis or regurgitation (Micheletti 2019). It is common for these patients to have an accessory (extra) electrical conduction pathway in the heart, potentially leading to episodes of supraventricular tachycardia known as Wolff-Parkinson-White syndrome. As well as medical and surgical treatment, they may also require electrophysiology assessment.

EA in the adult

Mortality and morbidity for children with EA have reduced with improved medical management and surgical techniques; most children live well into adulthood. Around 50% of patients will have required reoperation by 20 years after the initial procedure

Cyanotic CHD with mixed blood flow

Transposition of the Great Vessels (TGV)

Transposition of the great vessels (TGV) refers to a group of congenital heart in which there is abnormal arrangement of any of the great vessels (superior and/or inferior venae cavae, PA, pulmonary veins, and aorta).

Transposition of the great vessels/transposition of the great arteries

Transposition of great arteries (TGA) accounts for 2–5% of all CHD with a prevalence of about 0.2–0.3 of 1000 births and is more common in males than in females (Micheletti 2019). There are two types of TGA, simple (TGA with intact ventricular septum) and complex (with a VSD, an hypoplastic aortic arch or Interrupted aortic arch, or both). In a TGA the aorta arises from the RV, and the PA arises from the LV causing deoxygenated blood from the right heart to bypass the lungs and be pumped into the aorta and circulated throughout the body and the heart itself. The left heart continuously pumps oxygenated blood back into the lungs via the PA. This hemodynamic condition is incompatible with life unless a communication exists between the two circulations which allows a mixing between oxygenated and deoxygenated blood (Micheletti 2019). Prostaglandin E1 can be given to newborns to keep the ductus arteriosus open to allow mixing of the pulmonary and systemic circuits until surgery, though emergent balloon atrial

septostomy may be required in some cases (Sarris et al. 2017). Prognosis for TGA is good following surgical intervention (through the Arterial Switch operation) with survival rates greater than 90% (Szymanski et al. 2020).

Less common, is the congentially corrected TGA (ccTGA) (also known as Levo or l-TGA), accounting for less the 0.5% of CHD (Wallis et al., 2011) This is a rare acyanotic form of TGA that is associated with transposed primary arteries, and ventricles. This results in a functioning cardiac circuit, although the left side of the heart supplies the pulmonary system, while the right maintains the systemic circulation. , TGA can occur in isolation or as part of other complex CHD anomalies. Whilst surgery may be required when other complex anomalies are present, ccTGA alone will not require correction.

TGA in the adult

In adolescents and adults, longitudinal monitoring is required for patients who have undergone the Arterial Switch operation. Long term sequelae include right ventricular outflow tract obstruction at PV level or PA branch level, as well as AV regurgitation and ascending aorta dilatation (Safi and Bhatt, 2017). Coronary problems are rare.

Total and Partial Anomalous Pulmonary Venous Connection (TAPVC/PAPVC)

The reported birth prevalence of TAPVC ranges from around 0.5% to 2.0% of live births although this may be an underestimate due to historical difficulties in antenatal detection (Tongsong et al., 2016).

The four pulmonary veins take oxygenated blood from the lungs back to the left atrium. In TAPVC the veins are abnormally connected to the venous system – sometimes connected to the veins outside the heart and sometimes connecting with the right atrium. Oxygenated blood returns to the right atrium. The mixed blood moves across a PFO or ASD in order to reach the left atrium and then circulation. Surgery can reconnect the pulmonary veins to the left atrium and close shunts. In PAPVC some but not all the pulmonary veins connect to the right atrium. In this case, the effects on the patient will depend upon the amount of blood flow through the abnormal circuit.

Total/Partial Anomalous Pulmonary Venous Connection (TAPVC/PAPVC)

The four pulmonary veins take oxygenated blood from the lungs back to the left atrium.

TAPVC

In TAPVC the veins are abnormally connected – sometimes to the veins outside the heart and sometimes to the right atrium. Oxygenated blood from the lungs returns to the right atrium. Communication between the left and right atrium is crucial for survival in TAPVC. Mixing of

the oxygenated and deoxygenated blood occurs in the right atrium, which is then shunted right to the left at the level of atria. In most patients with TAPVC, an ASD is present, enabling the blood to shunt across until surgery is performed (Kim et al., 2014). The left atrium and aorta get mixed blood, which leads to cyanosis. In untreated patients, TAPVC is almost always fatal within the first few weeks of life. Surgery is offered soon after birth, in order to redirect the pulmonary veins to the left atrium and close the shunts (Sanjeev & Agarwal (2020).

PAPVC

In PAPVC, some but not all pulmonary veins connect into the right atrium (Sears et al., 2012). The main physiologic effects of PAPVC are similar to those of ASD, which is left to right shunt at the atrial level leading to recirculation of oxygenated blood through the pulmonary vasculature.

Patent Truncus Arteriosus

Truncus arteriosus (TA) occurs in 1–4% of all CHDs. During development, the truncus arteriosus should divide into a pulmonary trunk and an aorta. Rarely, it does not, leaving a common arterial trunk and VSD and a mixture of oxygenated and deoxygenated blood entering the circulation. Clinical presentation depends on the amount of pulmonary blood flow.

TA in the adult

Surgical repair is usually carried out within the first 6 months of life. This involves closure of the VSD and reconnection of the RV to PA branches with a conduit. Later presentation is often associated with PH and may lead to Eisenmenger syndrome (Micheletti 2019). Reoperation in order to change or upsize the RV-PA conduit may be required later in life.

Single Ventricle / Univentricular Hearts

Hearts with a functional single ventricle (functional univentricular heart) are rare, comprising 1–2% of all CHDs (Micheletti 2019) with a birth prevalence around 1 in 2000 births (Liu et al., 2019). The term ‘single ventricle’ is used to describe a heterogeneous group of anomalies with single atrioventricular connection (tricuspid atresia, mitral valve atresia, double-inlet LV) and/or severe hypoplasia of one ventricle and its own atrioventricular valve (hypoplastic left heart syndrome [HLHS], unbalanced complete AVSD). Although the anomalies may differ significantly from each other, they share the common feature that the patient develops with just one ventricle or one normal ventricle and a second undeveloped ventricle. The valves into the single functioning ventricle could be from both atria (*double inlet*), or from one atria, with the other valve being atresic, or have a single shared valve (*common inlet*). The developed ventricle chamber could be of either a right or left ventricular type. Over the past four decades, the

survival of these neonates has increased from 0 to nearly 90% (Ohye et al., 2016). Whilst predominantly a reflection of the development of surgical techniques, prenatal diagnosis is also associated with a higher survival in neonates with a single ventricle physiology (Weber et al., 2019). Mortality varies across the different anatomical presentations, with a lower mortality rate observed in patients with a dominant left ventricle and associated hyperplastic right ventricle (Beroukhim et al., 2015). Clinical findings will depend upon the amount of pulmonary blood flow and affected neonates may require PGE_1 infusion. Surgery is undertaken using a staged approach with several procedures, ultimately resulting in Fontan circulation.

Suggested resource	Hypoplastic left heart syndrome (HLHS) Cincinnati Children's
YouTube	URL: https://youtu.be/3CP3xZVgpdg

Adults with univentricular CHD

Patients with single ventricle and are at risk of multiple complications including cardiac, pulmonary, hepatic, gastrointestinal, and neurological problems that may start from 5 to 10 years after the operative procedures. Heart transplantation may the only effective option in some patients (Micheletti).

Whilst there have been significant developments in the treatment available to patients with CHD, not all lesions are correctable. Nonetheless, the offer of palliative procedures has seen a growing population of patients growing up with complex conditions. These lesions generally require multiple surgeries and patients will require lifelong follow up.

Most patients with single ventricle congenital heart disease are now expected to survive to adulthood. However, comorbidities are common (Collins II et al., 2016).

Suggested resource	The videos below from Cincinnati Children's outline the three operations undertaken for HLHS that result in Fontan circulation:
YouTube	Norwood operation URL: https://youtu.be/u5VbSDp2qMU Glenn repair URL: https://youtu.be/kBh8YDqcqxc Fontan operation URL: https://youtu.be/hIpvCUwml3c LHM Little Heart Matters: URL. https://www.lhm.org.uk/

Dextrocardia

Dextrocardia refers to alignment of the ? towards the right rather than the left. In most cases, it is diagnosed incidentally, typically on routine radiological examination, which reveals an abnormal location of the heart (Nair & Muthikuru 2020). Dextrocardia is often associated with other development anomalies with several possible presentations. If the heart is on the right side of the chest but all other organs are in their normal position it is known *situs solitus*; if the heart is in the opposite alignment facing the right with the abdominal organs also on the opposite side it is known as *situs inversus,* If the heart is turned to the right but the chamber positions are not changed and the abdominal organs stay in their usual position it is known as *dextroversion*. If the heart stays in the left position but the heart chambers invert and the abdominal organs also switch position it is known as *levoversion.* Both dextroversion and levoversion are almost always associated with additional heart defects such as transposition of the great vessels, pulmonary stenosis, VSDs, and ASDs.

The adult with dextrocardia

Most patients with dextrocardia are asymptomatic and lead a normal life. The prognosis of patients with dextrocardia depends on the presence or absence of other accompanying congenital defects and the type of congenital anomalies (Nair & Muthikuru 2020).

Key Point	People with dextrocardia may have heterotaxy syndrome. Organs in the chest and abdomen normally have a particular location on the right or left side of the body, termed "situs solitus." Rarely, the orientation of the internal organs is reversed, and is termed "situs inversus." Situs invertus does not normally cause problems unless it occurs as part of a syndrome affecting other parts of the body. "Heterotaxy syndrome" refers to an arrangement of the organs somewhere between situs solitus and situs inversus, but unlike situs invertus, it is associated with serious health issues including alterations to the structure of the heart and other organs.

Adult Congenital Heart Disease

With the growing success of heart surgery in infancy, many children born with congenital heart defects now survive to adulthood. In addition, improvements in imaging techniques mean that less complex heart defects are being in adults. The ratio of adults to children living with CHD is now estimated to be 2 to 1 (DeMaso et al., 2017) Understanding the needs of this emergent population is therefore essential. Anatomy alone is not the best measure of severity of adult congenital heart disease (ACHD) severity as anatomy and physiology in ACHD are not always

correlated. The severity of ADCH is determined by native anatomy, surgical repair, and current physiology. The recently developed ACHD Anatomical and Physiological (AP) classification system captures CHD anatomic variables as well as physiological variables, many of which have prognostic value in patients with ACHD. As with the New York Heart Association (NYHA) classification of functional status, patients may move from one ACHD AP classification to another over time (Stout et al 2019). Physiological variables used in ACHD AP Classification include aortopathy (diameter of the aorta); presence and type of arrhythmia; severity of concomitant valvular heart disease; end-organ dysfunction (including renal, hepatic, pulmonary); exercise capacity, hypoxaemia/hypoxia/cyanosis; NYHA functional classification; presence of PH; shunt (haemodynamically significant); and venous and arterial stenosis.

Table 24.2 gives an overview of the range of various ACHD types and the variables that affect functional status (Stout et al. 2019).

Table 24.2 ACHD AP Classification (CHD Anatomy + Physiological Stage = ACHD AP Classification)

CHD Anatomy*
I: Simple
Native disease ▪ Isolated small ASD ▪ Isolated small VSD ▪ Mild isolated pulmonic stenosis **Repaired conditions** ▪ Previously ligated or occluded ductus arteriosus ▪ Repaired secundum ASD or sinus venosus defect without significant residual shunt or chamber enlargement ▪ Repaired VSD without significant residual shunt or chamber enlargement
II: Moderate Complexity
Repaired or unrepaired conditions ▪ Aorto-left ventricular fistula ▪ Anomalous pulmonary venous connection, partial or total ▪ Anomalous coronary artery arising from the Pa ▪ Anomalous aortic origin of a coronary artery from the opposite sinus ▪ AVSD (partial or complete, including primum ASD) ▪ Congenital aortic valve disease ▪ Congenital mitral valve disease ▪ Coarctation of the aorta ▪ Ebstein anomaly (disease spectrum includes mild, moderate, and severe variations) ▪ Infundibular right ventricular outflow obstruction ▪ Ostium primum ASD ▪ Moderate and large unrepaired secundum ASD ▪ Moderate and large persistently patent ductus arteriosus ▪ Pulmonary valve regurgitation (moderate or greater) ▪ Pulmonary valve stenosis (moderate or greater)

CHD Anatomy*

- Peripheral pulmonary stenosis
- Sinus of Valsalva fistula/aneurysm
- Sinus venosus defect
- Subvalvar aortic stenosis (excluding HCM; HCM not addressed in these guidelines)
- Supravalvar aortic stenosis
- Straddling atrioventricular valve
- Repaired tetralogy of Fallot
- VSD with associated abnormality and/or moderate or greater shunt

III: Great Complexity (or Complex)

- Cyanotic congenital heart defect (unrepaired or palliated, all forms)
- Double-outlet ventricle
- Fontan procedure
- Interrupted aortic arch
- Mitral atresia
- Single ventricle (including double inlet left ventricle, tricuspid atresia, hypoplastic left heart, any other anatomic abnormality with a functionally single ventricle)
- Pulmonary atresia (all forms)
- TGA (classic or d-TGA; CCTGA or l-TGA)
- Truncus arteriosus
- Other abnormalities of atrioventricular and ventriculoarterial connection (i.e., crisscross heart, isomerism, heterotaxy syndromes, ventricular inversion)

Physiological Stage

A

- NYHA FC I symptoms
- No hemodynamic or anatomic sequelae
- No arrhythmias
- Normal exercise capacity
- Normal renal/hepatic/pulmonary function

B

- NYHA FC II symptoms
- Mild hemodynamic sequelae (mild aortic enlargement, mild ventricular enlargement, mild ventricular dysfunction)
- Mild valvular disease
- Trivial or small shunt (not hemodynamically significant)
- Arrhythmia not requiring treatment
- Abnormal objective cardiac limitation to exercise

C

- NYHA FC III symptoms
- Significant (moderate or greater) valvular disease; moderate or greater ventricular dysfunction (systemic, pulmonic, or both)
- Moderate aortic enlargement
- Venous or arterial stenosis
- Mild or moderate hypoxemia/cyanosis
- Hemodynamically significant shunt
- Arrhythmias controlled with treatment
- PH (less than severe)
- End-organ dysfunction responsive to therapy

CHD Anatomy∗
D
▪ NYHA FC IV symptoms ▪ Severe aortic enlargement ▪ Arrhythmias refractory to treatment ▪ Severe hypoxemia (almost always associated with cyanosis) ▪ Severe PH ▪ Eisenmenger syndrome ▪ Refractory end-organ dysfunction

ACHD indicates adult congenital heart disease; AP, anatomic and physiological; ASD, atrial septal defect; AVSD, atrioventricular septal defect; CCTGA, congenitally corrected transposition of the great arteries; CHD, congenital heart disease; d-TGA, dextro-transposition of the great arteries; FC, functional class; HCM, hypertrophic cardiomyopathy; l-TGA, levo-transposition of the great arteries; NYHA, New York Heart Association; TGA, transposition of the great arteries; and VSD, ventricular septal defect. ∗ This list is not meant to be comprehensive; other conditions may be important in individual patients.
Reproduced with permission from Stout, K. K., Daniels, C. J., Aboulhosn, J. A., Bozkurt, B., Broberg, C. S., Colman, J. M., ... & Khairy, P. (2019). 2018 AHA/ACC guideline for the management of adults with congenital heart disease: a report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. *Journal of the American College of Cardiology*, *73*(12), e81-e192.

Adult congenital heart disease (ACHD) patients require ongoing monitoring and sometimes further treatment. There is also a need for consideration of genetics and pregnancy issues for ACHD patients choosing to have children of their own. Other issues, such as the risk of infective endocarditis (IE), are particularly pertinent in some subgroups of ACHD patients where risk of infection is significantly higher than the standard population. The lifelong nature of CHD means continuity of care is a priority. Specialist ACHD services are required for patients moving from the paediatric to adult phase of their lives (Hays, 2015), with the term *transition* applied to this particular timeframe. This is the time at which young adults are likely to be leaving home and perhaps taking some risks in their lives, including with their health, and during this phase they are often lost to follow up.

Key Point	CHD is a lifelong condition, in some cases requiring multiple surgeries and lifelong monitoring.

Common issues associated with ACHD are briefly discussed below.

Pharmacotherapy

Patients with ACHD are commonly excluded from clinical trials, and there are few data to guide pharmacological therapies. Treatments used for HF patients may not have the same benefit in the heterogeneous population of patients with ACHD, and in some cases may cause

harm. Pharmacological therapies in patients with ACHD are often directed to specific conditions, such as beta blockers for arrhythmia treatment. Some pharmacological therapies affecting the pulmonary vasculature have a beneficial effect on long-term outcomes in patients with Eisenmenger syndrome. If new symptoms occur in a patient with ADHD, it is important to review the patient's symptoms in the context of their individual anatomy, prior surgical repair and physiology to assess whether there are any residual anatomical causes that may be amenable to intervention.

Infective Endocarditis

IE is an infection of the endocardium, or inner lining of the heart. It results from a complex interaction between a bloodstream pathogen (most commonly staphylococcus or streptococcus) and platelets at sites of endocardial cell damage. Turbulent blood flow that arises as a result of certain types of congenital or acquired heart disease, such as shunts or blood flow across a narrowed orifice (such as valvular disease), traumatise the endothelium that fosters deposition of platelets and fibrin on the surface of the endothelium (Wilson et al., 2007). Activities that risk introducing bacteria to the blood stream similarly increase the chance of developing IE. These include interventions such as hemodialysis, intravenous drug taking or administration, or dental procedures. Mucosal surfaces, such as those in the mouth, are populated by a dense endogenous microflora. Disturbance of this surface, particularly the gingival crevice around teeth, gastrointestinal tract, urethra, and vagina, transiently releases different microbial species into the bloodstream (Wilson et al., 2007). IE may present as an acute, subacute or chronic condition and the clinical presentation is highly variable reflecting the variable causative microorganisms, underlying cardiac conditions and pre-existing comorbidities (Rajani & Klein 2020).

IE in the ACHD patient

IE is 15 to 30-fold higher in patients with CHD than in the general population (Ly et al. 2019). Cardiac anatomy and previous interventions place some ACHD patients at higher risk of developing IE than others and mortality is particularly high in patients with complex CHD (Ly et al., 2019). Whilst the difficulty in defining specific 'high risk' lesions are acknowledged, there is general agreement that the following conditions put CHD patients at 'high risk' for developing IE (Stout et al. 2019):

- previous IE;
- prosthetic valves (biological and mechanical, surgical and transcatheter);
- placement of prosthetic material within the previous six months;

- residual intracardiac shunts at the site of or adjacent to previous repair with prosthetic material or devices; or
- uncorrected cyanotic heart disease.

Recommendations for treatment

Currently here are no randomised controlled trials (RCTs) to support antibiotic prophylaxis to prevent IE following dental extraction (Cahill et al., 2017).Consequently, the National Institute for Health and Care Excellence (NICE) issued guidelines in 2008 recommending that antibiotic prophylaxis during invasive dental procedures should no longer be offered to people at risk of infective endocarditis in England. Subsequent epidemiological studies undertaken have pointed to an increase in cases of IE in England (Dayer et al., 2015). The 2016 NICE guideline update reaffirms this position but inserts the word *routinely* to the recommendation that antibiotic prophylaxis should no longer be offered, stating that doctors and dentists should apply clinical judgement on a case by case basis (Chambers et al. 2019). In contrast, the American Heart Association and European Society of Cardiology guidelines continue to recommend antibiotic prophylaxis in certain high-risk cases such as those listed above (Quan et al. 2020).

Exercise and Sports

Historically, the focus has been on restriction of activity for adults with CHD due to fears of adverse events such as sudden cardiac death (SCD) or aortic dissection. More recently, it has been suggested that most patients with ACHD can safely engage in regular, moderate physical activity but it is important to note that some conditions, such as systemic ventricular systolic dysfunction, systemic ventricular outflow tract obstruction, haemodynamically significant arrhythmias, or aortic dilation, warrant more cautious recommendations (Stout et al. 2019). The level of sports participation recommended must be individualised to the particular patient and take into account the training and the competitive aspects of the activity, the patient's functional status, and history of surgery. Noninvasive testing including exercise tolerance testing, holter monitoring, echocardiography, and cardiac magnetic resonance imaging may be useful in decision-making (Van Hare et al. 2015).

Suggested resource Reading	The reading below provides recommendations for competitive athletes with particular types of heart disease including CHD: Pelliccia, A, Sharma, S, Gati, S, Bäck, M, Börjesson, M, … Wilhelm, M, ESC Scientific Document Group, 2020 ESC Guidelines on sports cardiology and

exercise in patients with cardiovascular disease: The Task Force on sports cardiology and exercise in patients with cardiovascular disease of the European Society of Cardiology (ESC), *European Heart Journal*, ehaa605, https://doi.org/10.1093/eurheartj/ehaa605

Latest ESC guidance on ACHD https://www.escardio.org/The-ESC/Press-Office/Press-releases/what-happens-when-babies-with-heart-defects-become-adults?esctwitter

Key Point Physical activity is widely recognized as being beneficial for physical and mental health. Activity recommendations for people with ACHD should be individualised according to clinical status and the patient's interests (Stout et al. 2019).

Sexual health

Guidance on sexual functioning and reproductive health issues has largely been neglected for patients with ACHD. Sexuality is an important element of quality of life (QoL) and though sexual function is an issue that affects QoL for both women and men with ACHD, there appears to be minimal evidence to guide interventions (Stout et al. 2019) or advice on the safety of sexual activity. Most patients with ACHD have a reduced exercise capacity. It is uncertain to what extent this is caused by the heart defect per se or a sedentary life style because of restrictions and/or overprotection (Sandberg et al. 2016). There is evidence to suggest that adults with CHD have fears regarding the safety of performing sexual activities triggered by anecdotal reports of sudden deaths occurred during sexual arousal and 15% of men (and 9% of women reported cardiac symptoms (dyspnea, perceived arrhythmia, increased fatigue, syncope, chest pain) during sexual activity, with symptoms being more common in those with more severe disease (Vigl et al. 2009; Vigl et al. 2010). Concerns with sexual health are present in 20% to 40% of men with CHD and erectile dysfunction is reported by up to 42% of men with CHD (Stout et al 2019). The prevalence of sexual dysfunction in ACHD patients is high and the aetiology appears to be multifactorial. Psychological health, cardiac medications, clinical symptoms, and delay in psychosexual development may all play a part in sexual dysfunction for people with ACHD (Huang and Cook 2018).

Key Point	Patients often feel do not feel confident in voicing concerns about sexual health. Nurses should create an environment in which the patient with ACHD feels comfortable addressing concerns about their sexuality.

Early discussion and counselling about reproductive risk is essential. This includes information on the individual risk of inherited cardiac disease, and for women, the risks associated with pregnancy itself. These risks vary significantly depending on the defect and whether there has been surgical repair. Ideally, discussions should take place before the woman becomes pregnant.

Pregnancy

Pregnancy is frequently more complicated in women with CHD and requires specialty management (Stout et al. 2019). Women with CHD have greater rates of comorbidities that may complicate delivery, including cardiomyopathy, valvular heart disease, PH, systemic HT, cardiac conduction disorder, anaemia, and non-gestational diabetes (Schlichting et al. 2019). The risk of adverse pregnancy outcomes is much higher in women with CHDs due to the strain pregnancy places on the cardiovascular system, including increased blood volume, heart rate, and cardiac output which may lead to HF, stroke, arrhythmias, and myocardial infarction. Women with CHD are also have a higher frequency of adverse obstetric events including preterm labor, maternal death, operative vaginal delivery, and cesarean delivery. Babies born to women with CHDs are also at higher risk of including premature birth, being small for gestational age and CHD or other congenital anomalies (Schlichting et al. 2019).

Suggested resource Reading	Canobbio, M. M., Warnes, C. A., Aboulhosn, J., Connolly, H. M., Khanna, A., Koos, B. J., ... & Stout, K. (2017). Management of pregnancy in patients with complex congenital heart disease: a scientific statement for healthcare professionals from the American Heart Association. *Circulation*, *135*(8), e50-e87. URL: https://www.ahajournals.org/doi/pdf/10.1161/cir.0000000000000458

Pre-pregnancy counseling

Women with ACHD who are considering pregnancy should have an individualised risk assessment individual patient's anatomy and physiology, followed by discussion about maternal risks during pregnancy, delivery, and the postpartum period, as well as foetal risk in regard to CHD

transmission and overall risk to the health of the foetus. Ideally this should be undertaken with a cardiologist who specialises in ACHD. Women at extremely high risk of maternal mortality or severe morbidity include (but are not limited to) those with PH, LV EF <30% and/or NYHA III–IV symptoms, severe left heart obstruction, or severe native coarctation. In pregnant women with these conditions, the option of pregnancy termination should be discussed (Stout et al. 2019).

Key Point	If the patient with CHD or their partner is pregnant, there is an increased risk of CHD in the offspring. Foetal echocardiography can be useful in determining whether CHD is present, and if so, helps to inform the course of action at the time of delivery (Stout et al. 2019).

Birthing in high risk women should take place in a specialist centre with multidisciplinary input and close monitoring (Brennan and Hatch, 2018).

CHD in older adults

Many people with CHD are now living to middle-age and some into the geriatric age range leading to increased use of the medical system for both routine and episodic care. CHD and the sequelae of previous interventions must be treated in the setting of late complications, acquired cardiac disease, and the general effects of aging on other body systems.

Suggested resource Reading	Bhatt, A. B., Foster, E., Kuehl, K., Alpert, J., Brabeck, S., Crumb, S., ... & Mital, S. (2015). Congenital heart disease in the older adult: a scientific statement from the American Heart Association. *Circulation*, *131*(21), 1884-1931. https://doi.org/10.1161/CIR.0000000000000204

Genetic heart disease

Advances in family screening and human gene profiling has led to increased recognition of genetic abnormalities that can lead to congenital heart defects. Certain established patterns of gene disorders, often called by eponymous syndromes, are closely associated with heart defects. Where there are congenital birth defects, a family history should look to identify possible

genetic causes. Family screening should be considered and where particular genes are identified as disordered family members can have genotype screening. Certain physical characteristics and developmental delays or learning disabilities can suggest certain syndromes.

Psychosocial considerations

Whilst there have been marked improvements in understanding and subsequent support and care provision, patients with CHD remain at higher risk of psychological problems compared to a healthy population (Kronwitter et al., 2019). The risk and severity of psychological impairment, encompassing cognitive deficits, mood and anxiety disorders, and posttraumatic stress disorder increases with greater complexity of the cardiac lesion (Wilson et al., 2015). In particular, depression in adults with CHD is highly prevalent, and is strongly associated with poor prognosis (Huntley et al., 2019, Ladak et al., 2019). Similar patterns are observed within the paediatric and adolescent CHD population, in particular in those with single ventricle anatomy (DeMaso et al., 2017).

Learning activity 24.1	When patients (or their children) are diagnosed with CHD, most will seek information online about the condition. Identify some reliable resources to which you could direct patients. Consider also organisations that may relate to CHD-related complications. Some examples are provided below but there are many more.
Suggested resource YouTube	Adult Congenital Heart Disease Association http://www.achaheart.org/ Arrhythmia Alliance http://www.heartrhythmcharity.org.uk British Heart Foundation https://www.bhf.org.uk/ Canadian Congenital Heart Alliance http://www.cchaforlife.org/ Cardiomyopathy Association http://www.cardiomyopathy.org/ Heart Kids Australia http://www.heartkids.org.au/ Heart Kids New Zealand http://heartnz.org.nz/ Pulmonary Hypertension Association http://www.phassociation.uk.com/ The Somerville Foundation http://www.thesf.org.uk/

Conclusion

Congenital heart defects are the most common form of congenital anomaly. They are a heterogeneous group of defects affecting the structure of the heart or great vessels. Due to developments in technology and surgery, the majority of patients now survive into adulthood. The needs of this group of patients differ to those with acquired heart disease and therefore understanding the underlying pathophysiology of this group of conditions is essential in order to optimize care.

Learning activity 24.2	There are several video resources available on YouTube on the topic of CHD. As you read about the various types of CHD in this chapter, locate a video on YouTube that relates to the anomaly and describes surgical or percutaneous repair procedures. See the example below:
Suggested resource YouTube	The Cincinnati Children's Heart Encyclopedia provides detailed information on the congenital heart defects described in this chapter, including signs and symptoms, diagnoses and treatment options. A list can be found at: Cincinnati Children's Heart Encyclopedia URL: https://www.cincinnatichildrens.org/patients/child/encyclopedia/defects

Suggested resource YouTube	Congenital Heart Disease Joseph Alpert – Lecturio Medical (2018) URL: https://youtu.be/pNn7pICPAvU
Suggested resource YouTube	An Approach to Congenital Heart Disease R. Kannan Mutharasan (2018) URL: https://youtu.be/t0rJOIb10PQ
Suggested resource website	American Heart Association Congenital heart defects (2020) URL: https://www.heart.org/en/health-topics/congenital-heart-defects

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