Cardiac system 2: congenital heart disease and pathophysiology

Key points
- Any component of the cardiac system can become compromised, impairing normal heart function
- Increasing numbers of children with congenital heart defects survive into adulthood
- Damage to the cardiac valves may lead to them to become regurgitant or stenotic
- The most common cause of coronary artery disease is atherosclerosis
- An arrhythmia is defined as a heart rhythm that is not normal sinus rhythm

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Abstract
The heart can develop a plethora of diseases that impair its normal function, some of which are life-threatening and require urgent intervention. This article will raise nurses’ awareness and improve their knowledge of commonly encountered cardiac diseases. It is the second in a two-part series, with part 1 covering cardiac anatomy and physiology.

Citation

There is a plethora of diseases that directly affect the heart, as well as many inflammatory, infective or systemic diseases that indirectly impair cardiac function. The frequency of coronary artery disease (CAD) and ischaemic cardiomyopathy (ICM) is increasing, while patients with congenital heart disease (CHD) now live well into adulthood and have children of their own. Health professionals need to understand cardiac conditions, since they are likely to encounter them in all settings.

How does the heart work?
The heart acts as a pump and works in conjunction with blood vessels to transport blood around the body (see part 1: Bit.ly/NTCardiacSOL).

Any component of this complex system can become compromised for a variety of reasons and impair normal function.

Congenital heart disease
CHD refers to a range of conditions that normally manifest in childhood, although sometimes they are not identified until adulthood. CHD affects up to nine in 1,000 newborns in the UK; thanks to recent advances in treatment, 80% will survive into adulthood (NHS England, 2015).

To understand why some CHDs occur, one needs to understand foetal circulation. In foetal life, most oxygenated blood comes from the placenta; this has a low vascular resistance, while the lungs in the foetus are fluid filled (and have high resistance) so only 10% of the blood passes to the lungs (Fernandes, 2017). In the foetus, blood is diverted away from the lungs by two right-to-left shunts. The first is the foramen ovale, an opening in the
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Systems of life

Interratial septum, so that in foetal life blood is shunted from the right to left atrium. Atrial septal defects (ASDs) can occur when the foramen ovale does not close after birth, as it should. As a result, there is a left-to-right shunt and oxygen-rich blood in the left side of the heart mixes with oxygen-poor blood in the right side. This reduces oxygen saturation levels in the blood delivered to the rest of the body. The larger the shunt, the higher the risk of long-term complications such as atrial fibrillation and pulmonary hypertension.

“In cardiomyopathies, the heart becomes structurally and functionally abnormal”

A second important heart structure during foetal life is the ductus arteriosus, which connects the pulmonary artery to the arch of the aorta and diverts any blood that passes through the right ventricle to the lungs back into the aorta (another right-to-left shunt). If it does not close after birth as it is supposed to, the ensuing defect, known as a patent ductus arteriosus (PDA), allows oxygenated blood from the aorta to mix with deoxygenated blood in the pulmonary artery and increase the lung pressures.

The most common congenital heart defects are ventricular septal defects (VSDs), which affect one in 500 live births. VSDs cause an abnormal communication between the right and left ventricles. Oxygenated blood flows through the VSD from the left into the right ventricle (Fig 1). Again, there is a left-to-right shunt. Clinical severity depends on the size of the defect and in some cases, VSDs can lead to heart failure.

Other CHD defects include: a transposition of the great arteries resulting in the aorta arising from the right ventricle (instead of the left) and the pulmonary artery arising from the left ventricle (instead of the right); a tetralogy of Fallot, which combines a VSD, pulmonary valve stenosis, right ventricular hypertrophy and an overriding aorta where the aorta is positioned directly over a VSD; and a narrowing of the aorta called coarctation.

Some defects, including ASD, VSD and coarctation of the aorta, may be associated with genetic diseases such as trisomy 21 (Down’s syndrome) or Turner syndrome.

Diseases of the heart valves

The cardiac valves are key in maintaining a smooth and unidirectional blood flow through the heart. They are anchored to the endocardium by tendons called chordae tendineae, attached to the papillary muscles. Any injury or abnormality affecting the valves, chordae tendineae or papillary muscles, as well as any changes in the myocardium (such as dilation), can alter valvular function. Damage may culminate in the valves either becoming regurgitant (leaky valves), which means blood flows backwards, or exhibiting stenosis (tightened valves), reducing outflow. Cardiac valve disease may be congenital or acquired as a result of rheumatic fever or bacterial endocarditis (Box 1).

Rheumatic fever

Rheumatic fever is an autoimmune inflammatory disorder that develops after an untreated pharyngeal infection (often caused by group A streptococcus bacteria) (LeMone et al, 2010). It can affect any of the heart layers, and up to 10% of affected patients develop cardiac valve disease. Valve leaflets may become rigid, or the chordae tendineae may undergo fibrosis and shortening. It is mostly the mitral and aortic valves that are affected, becoming regurgitant or stenotic.

Bacterial endocarditis

Bacterial endocarditis is an infection of the endocardium and usually affects the cardiac valves, on which lesions called vegetations, made up of platelets and fibrin, develop. Initially sterile, these become colonised by micro-organisms and cause scarring and deformities on the valves, resulting in regurgitation or stenosis. They may also break off and travel in the bloodstream to other organs (embolise), causing infarction and infection. Patients most at risk of bacterial endocarditis include those with previous heart damage or surgery, those with pre-existing valve disease or valve replacement, and those with tooth abscesses or undergoing dental procedures.

Diseases of the coronary vessels

Diseases of the coronary arteries are a major cause of death and disability in developed countries (Wang et al, 2016). The heart receives its own blood supply from the left and right coronary arteries and their branches. A narrowing or blockage of the arteries will reduce that supply and starve the myocardium of oxygen (ischaemia). A complete loss of blood supply to a region of the myocardium can lead to irreversible damage, muscle cell death (infarction) and remodeling of the heart.

The most common cause of CAD in the Western world is atherosclerosis, with its main risk factors being hypertension, hypercholesterolaemia, diabetes, smoking and a family history of ischaemic heart disease. Atherosclerosis is an inflammatory state where a lipid-rich plaque forms on the inner lining of blood vessels. Over time, the plaque progresses, causing a narrowing of the artery.

Some patients with CAD present with stable angina (chest pain that occurs
predictably upon physical exertion or emotional stress and lasts <10 minutes) or unstable angina (new-onset angina or abrupt deterioration of previously stable angina) (National Institute for Health and Care Excellence, 2017). Both are due to a mismatch between supply and demand of blood flow to the heart (Ford et al, 2017).

Rupture of an atherosclerotic plaque in the wall of a coronary artery affects the blood supply to the region supplied by that artery, and the cells are starved of oxygen, leading to cell necrosis. In that scenario, timely medical intervention is key to preserve the myocardium (Jarvis and Saman, 2017a).

Diseases of the myocardium

For an effective cardiac output, the myocardium needs to generate large forces, so anything that impairs its ability to do so will affect cardiac output and may ultimately lead to heart failure. In cardiomyopathies (diseases of the myocardium), the heart becomes structurally and functionally abnormal (Cooper, 2017). There are various types of cardiomyopathies (Fig 2), including dilated cardiomyopathy (DCM), hypertrophic cardiomyopathy (HCM), restrictive cardiomyopathy (RCM), arrhythmogenic right ventricular cardiomyopathy (ARVC) and ischaemic cardiomyopathy (ICM).

DCM, the most common type, is characterised by an enlarged and poorly contracting left ventricle. It occurs due to genetic or acquired causes such as hypertension, inflammation or infection, valve disease, metabolic disease and toxins (McNally and Mestroni, 2017). Reversible causes include alcohol intake, cocaine intake and pregnancy. Due to the progressive nature of DCM, the prognosis is poor and around 50% of patients die within five years of diagnosis (Cooper, 2017; McNally and Mestroni, 2017).

HCM has a completely different pathology: asymmetrical hypertrophy of the left ventricle leads to reduced ventricular compliance, reduced filling and reduced cardiac output. A large proportion of cases are due to an underlying genetic disease, and around 50% of patients have a family history of HCM or sudden cardiac death.

In RCM, the least common type, there is increased myocardial stiffness leading to impaired ventricular filling and reduced cardiac output. It accounts for around for 5% of cases and may be due to a primary cause (endomyocardial fibrosis, idiopathic cause), systemic diseases that cause an infiltrative cardiomyopathy (such as sarcoidosis and amyloidosis) or genetic storage diseases (such as haemochromatosis, glycogen storage, Fabry’s disease) (LeMone et al, 2010).

In ARVC the ventricles have a scarred appearance and their muscles are replaced by fibrous or fibro-fatty tissues. Patients are at risk of ventricular arrhythmias and sudden cardiac death, and need urgent intervention.

Incidence of ICM is rising because more patients presenting to hospital with acute myocardial infarction are successfully treated and survive but develop ventricular remodelling and chronic dysfunction of the myocardium (Jarvis and Saman, 2017b; Briceno et al, 2016).

Diseases of the conduction system

Each heart beat is initiated by an electrical impulse from the sinotrial node (SAN) that passes to the atrioventricular node (AVN) and, in response, the atria and ventricles beat sequentially. In normal circumstances, the SAN is the pacemaker, but if there is a problem with it, the AVN or ventricles can assume that role. This is referred to as an escape rhythm (Newby et al, 2014).

Fig 2. Different types of cardiomyopathy

<table>
<thead>
<tr>
<th>Hypertrophic cardiomyopathy</th>
<th>Dilated cardiomyopathy</th>
<th>Restrictive cardiomyopathy</th>
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<tbody>
<tr>
<td>Hypertrophy of the ventricular mass</td>
<td>Dilated heart chambers</td>
<td>Rigidity of the ventricle walls, which alters filling ability</td>
</tr>
<tr>
<td>Small LV volume</td>
<td>Impaired pump action</td>
<td>Secondary to systemic diseases</td>
</tr>
<tr>
<td>Dilated LA occurs</td>
<td>Increased volumes of end diastolic and end systolic residual blood in chambers</td>
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RV – right ventricle; LV – left ventricle

Hypertrophy of ventricular mass

Small LV volume

Dilated LA occurs

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An arrhythmia is defined as a heart rhythm that is not normal sinus rhythm. During normal sinus rhythm, the heart rate should be 60-100 beats per minute (bpm). Tachycardia occurs when the heart rate is >100bpm, while bradycardia is when the heart rate is <60bpm. An arrhythmia can be paroxysmal (that is, begin and stop abruptly) or continuous.

Cardiac arrhythmias can be broadly divided into (Newby et al, 2014):
- Sinus arrhythmias (for example, sinus bradycardia and sinus tachycardia);
- Atrial arrhythmias;
- Ventricular arrhythmias (for example, ventricular tachycardia);
- Atrioventricular block (a delay or interruption of the impulse transmitted from the atria to the ventricles).

Box 2 lists common arrhythmias.

### Diseases of the pericardium

The pericardium covers the heart and anchors it in the thorax. A number of conditions can lead to inflammation of its membranes (pericarditis), which causes chest pain. Pericarditis may be a primary disorder or be secondary to another systemic disease (such as renal failure, connective tissues disease, infection and myocardial injury). Complications include pericardial effusion (the presence of fluid between the pericardial layers), constriction pericarditis, and cardiac tamponade (where compression of the heart, if left untreated, causes cardiac arrest).

**Box 2. Common arrhythmias**
- Atrial premature beats
- Ventricular premature beats
- Bradycardia (including sinus bradycardia)
- Ventricular tachycardia
- Atrial fibrillation and atrial flutter
- Supraventricular tachycardia
- Atrioventricular block
- Non-sustained ventricular tachycardia

Source: Lévy and Olshansky (2017)

### References


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