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Abstract

Cardiomyopathy is the most common cause of sudden cardiac death in young people, including trained athletes (Maron et al, 1996). Often the tragedy of sudden death is the first indication of its presence within a family. This article describes the four types of cardiomyopathy, diagnosis, treatment and management options, and discusses ways in which the new National Service Framework for Coronary Heart Disease (Department of Health, 2005) will help in planning and implementing services.

Cardiomyopathy is a disease of the heart muscle associated with cardiac dysfunction. There are four types: hypertrophic; dilated; arrhythmogenic right ventricular; and restrictive cardiomyopathy. All four types cause significant morbidity and mortality and are among the most common genetically inherited heart conditions. Early recognition is vital to help avert complications and prevent sudden cardiac death.

Hypertrophic cardiomyopathy
Hypertrophic cardiomyopathy (HCM) is characterised by unexplained myocardial hypertrophy and myocyte disarray. It is the most common type of cardiomyopathy, affecting around 1 in 500 people, both male and female, and all races (Sorajja et al, 2000).

HCM is predominately a familial disease with an autosomal dominant inheritance pattern, which means that each child of an affected parent has a 50 per cent chance of inheriting a particular HCM gene. The condition does not skip generations. However, there is variable penetrance, which means that some family members may be mildly affected while others may experience a severe form of the disease.

Research into the cause of HCM has so far identified 10 mutant genes affecting the sarcomeric contractile proteins (Seidman and Seidman, 2001). Other uncommon causes of HCM include Anderson-Fabry disease, Noonan’s syndrome, Leopard syndrome and Friedrich’s ataxia (Elliott and McKenna, 2004).

The main feature of HCM is an excessive thickening of the myocardium, usually the left ventricle, which can occur in any pattern (Fig 1). Asymmetric septal hypertrophy (ASH), where the interventricular septum is thicker than the posterior or left ventricular free wall, is the most common presentation and is seen in 50–60 per cent of cases.

Apical hypertrophy is present in approximately 10 per cent of patients. This type of hypertrophy is not usually associated with a heart murmur and gives a typical appearance on electrocardiogram (ECG) of giant negative (inverted) T-waves in the precordial leads. Concentric hypertrophy is when excessive muscle thickening occurs with equal severity throughout the whole ventricle.

In addition to the excessive thickening of the heart muscle people with HCM have myocardial disarray, which can affect up to 40 per cent of the heart muscle and can cause scarring in the septum, left ventricular free wall and occasionally the papillary muscles. Myocardial disarray is impossible to assess in life and it is thought to be responsible for electrical instability and diastolic dysfunction (McKenna et al, 1990).

Symptoms and diagnosis
HCM can present at any age. Although the patient has the genetic error, signs of HCM are not usually apparent until adolescence, and in fact an ECG recording and echo may be entirely normal in childhood. Many patients are asymptomatic and may be identified during screening or may complain of mild symptoms of the condition. Physical examination may be unremarkable.

Learning objectives
Each week Nursing Times publishes a guided learning article with reflection points to help you with your CPD. After reading the article you should be able to:

- Understand the differences between the four types of cardiomyopathy;
- Know the genetic inheritance of cardiomyopathy and the need to take a detailed family history;
- Be familiar with the mechanisms of cardiomyopathy and prevention of sudden cardiac death;
- Be aware of treatment options for cardiomyopathy.
Common symptoms of HCM include chest pain, dyspnoea, syncope, arrhythmias and lethargy. Some families present after a sudden cardiac death in the family.

Approximately 20–25 per cent of patients will have obstruction to the outflow tract, in which case the term HOCM (hypertrophic obstructive cardiomyopathy) is then used. This means that there is narrowing of the outflow tract that interferes with the normal ejection of blood from the heart. This pressure difference between the outflow tract and the aorta is measured in millimetres of mercury (mmHg) during echocardiography and is called a gradient.

The obstruction is not always obvious as it may only occur during exercise. Symptoms that may alert the cardiologist that the patient has obstruction include shortness of breath, chest pain, presyncope, syncope and exercise intolerance.

HCM is diagnosed by physical examination, family history and ECG, and echocardiography. Other tests performed to confirm the diagnosis include 48-hour ECG, exercise testing with VO₂ measurement and magnetic resonance imaging (MRI). Some patients will require a chest X-ray and coronary angiogram.

**Treatment of HCM**

Medical therapy aims to alleviate the symptoms of HCM, avoid complications and prevent sudden cardiac death. Many patients are asymptomatic and therefore do not require treatment. However, they should remain under regular consultant follow-up.

Symptomatic management relies on beta-blockers and calcium antagonists. These reduce myocardial oxygen consumption and slow the heart rate, so relieving angina and improving diastolic filling. Patients with obstruction have a number of options, including the use of disopyramide, or surgical options, namely myectomy or alcohol septal reduction.

Arrhythmias are common in HCM and their origin should be investigated and treated as necessary. Atrial fibrillation (AF) affects approximately 10 per cent of patients. Treatment initially involves cardioversion but may require long-term use of amiodarone. Patients with AF or large atria will require anticoagulation. Those at high risk of sudden cardiac death should be offered an implantable cardioverter defibrillator. Risk stratification is important in all patients regardless of presentation or symptoms.

**Dilated cardiomyopathy**

Dilated cardiomyopathy (DCM) affects 1 in 2,500 people and is characterised by dilation and impaired contraction of the left or both ventricles. Although in many cases the aetiology is not known, many factors appear to be associated with its development. In 30–40 per cent of patients it is genetically inherited, and in those families who have inherited disease, as with HCM, there is a dominant pattern of inheritance.

**Symptoms and diagnosis**

Symptoms may appear at any age and usually include signs of pulmonary congestion and/or low cardiac output. There is often a history of fatigue and exertional symptoms for months or even years prior to diagnosis (Elliott, 2000).

Symptoms are often progressive and can necessitate cardiac transplantation. Arrhythmias, thromboembolism and sudden death are common and may occur at any stage. Symptoms include dyspnoea, palpitations, reduced exercise capacity, fatigue, orthopnoea, paroxysmal nocturnal dyspnoea, oedema and syncope.

An important part of the clinical assessment is the patient’s medical and family histories as these reveal specific causes of DCM. Investigations are similar to HCM and include ECG, echocardiogram, 48-hour ECG, exercise testing with VO₂ measurement, coronary angiogram and chest X-ray.

**Treatment of DCM**

Patients who are symptomatic should receive medical therapy, which aims to control their symptoms, reduce the left ventricular dimensions and prevent sudden cardiac death. Many patients are asymptomatic and therefore do not require treatment. However, they should remain under regular consultant follow-up.

Symptomatic management relies on beta-blockers and calcium antagonists. These reduce myocardial oxygen consumption and slow the heart rate, so relieving angina and improving diastolic filling. Patients with obstruction have a number of options, including the use of disopyramide, or surgical options, namely myectomy or alcohol septal reduction.

Arrhythmias are common in HCM and their origin should be investigated and treated as necessary. Atrial fibrillation (AF) affects approximately 10 per cent of patients. Treatment initially involves cardioversion but may require long-term use of amiodarone. Patients with AF or large atria will require anticoagulation. Those at high risk of sudden cardiac death should be offered an implantable cardioverter defibrillator. Risk stratification is important in all patients regardless of presentation or symptoms.

**REFERENCES**


Arrhythmogenic right ventricular cardiomyopathy

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a progressive, inherited heart muscle disease that is characterised by right ventricular myocardial atrophy and fibro-fatty replacement (where the heart muscle is gradually replaced by fatty tissue). Areas of fatty tissue may lead to weakness and bulges in the cardiac muscle wall. Over time the progressive nature of the condition may cause more diffuse right ventricular involvement and left ventricular involvement and may culminate in heart failure (Hamid, 2001).

The incidence of ARVC has been estimated as somewhere between 1 in 3,000 and 1 in 10,000 and predominantly presents in adolescents or young adults. In the older age group patients may present with symptoms of heart failure. ARVC may present in different ways and there are said to be four distinct phases:

- The concealed phase – patients are asymptomatic and may go undiagnosed;
- Symptomatic ventricular arrhythmia;
- Diffuse right ventricular disease – results in right-sided heart failure;
- The advanced stage – the patient may present with what appears to be dilated cardiomyopathy. The prognosis for ARVC is uncertain as there is a wide disease spectrum and presentation.

Symptoms and diagnosis

Symptoms of ARVC include palpitations, syncope and ventricular arrhythmias. Some patients may present with symptoms of heart failure. The diagnosis is made by ECG, echocardiography, 48-hour ECG, signal-averaged ECG and exercise testing with VO₂ measurement. Some patients will require an MRI scan and coronary angiogram to confirm the diagnosis.

Treatment

Anti-arrhythmic therapy with beta-blockers or amiodarone is used to control symptoms in patients with non-life-threatening ventricular arrhythmias. Due to impaired ventricular filling the patient with restrictive cardiomyopathy may present with symptoms of right and left heart failure and commonly have fairly advanced disease. Symptoms include exercise intolerance, lethargy, palpitations, oedema and syncope.

A diagnosis of RCM can be made using ECG and echocardiogram. An MRI scan may be useful to highlight the structure of the heart muscle. Some patients require endomyocardial biopsy to confirm the diagnosis.

Treatment of restrictive cardiomyopathy

Treatment of RCM again aims to alleviate symptoms, so will include beta-blockers, anti-arrhythmic drugs and diuretics. Some patients require anticoagulation,
and those with severe disease will be referred for cardiac transplantation. If familial disease is suspected then immediate family members should be offered cardiac screening.

**Risk stratification and prevention of sudden cardiac death**

Sudden and unexpected cardiac death is the most devastating and unpredictable complication of cardiomyopathy and occurs in all four types. Awareness of cardiomyopathy is important within the medical community because early diagnosis and detection of those at risk mean that prophylactic treatment can begin and young patients can be prevented from dying prematurely from arrhythmias and thromboembolisms.

All patients should be assessed to determine their risk of sudden cardiac death (Table 1). This involves an echocardiogram (looking at wall thickness), an exercise test (looking for ventricular arrhythmias and blood pressure response to exercise), a 48-hour ECG to detect any runs of ventricular beats. In addition, a detailed family history should be taken to determine if there is a family history of sudden cardiac death.

As a general rule if a patient has two or more risk factors for sudden cardiac death then they would be offered appropriate treatment, either in the form of an implantable cardioverter defibrillator or amiodarone if preferred. Patients who are at low risk can be reassured. However, patients who are at high risk should be strongly advised to avoid any strenuous exercise or competitive sports that require extreme physical exertion (McKenna and Behr, 2002).

**Screening families for signs of genetic inheritance**

In families where cardiomyopathy is inherited it is usually dominantly inherited. Therefore, screening of all first-degree relatives is vital – this includes brothers, sisters, children and parents of an affected individual.

Screening involves a physical examination, a detailed family history and a 12-lead ECG and echocardiogram. Signal-averaged ECG and exercise testing may also be included.

The expression of the disease can be age-related, occurring during rapid periods of growth, which is why intense cardiac screening is recommended during adolescence. Thereafter, if test results remain normal, screening should still be repeated at five-yearly intervals.

**Implementation of services**

A new chapter covering the subject of arrhythmias and sudden cardiac death was published in the *National Service Framework for Coronary Heart Disease* in 2005 (DoH, 2005). The NSF sets out a blueprint for how the NHS should respond to sudden cardiac death, giving quality standards for the diagnosis and treatment of arrhythmias and the care of patients with inherited heart diseases.

Included in the framework are suggested referral pathways for patients who have a history of collapse with unknown cause and how to evaluate families who have experienced sudden cardiac death.

The document states that first-degree relatives of victims of sudden cardiac death who are below the age of 40 should be referred to a heart rhythm expert, and that the evaluation of families should take place within a dedicated clinic and be carried out by staff who are trained in the diagnosis, management and support of the condition. It also says that genetic and other testing should be available.

**Nursing considerations in the management of cardiomyopathy**

Care of patients with cardiomyopathy presents the cardiac nurse with many challenges. The presentation within a family may range from the asymptomatic patient, who requires no treatment, to heart failure and in some cases, sudden cardiac death.

Cardiomyopathy is a chronic condition with extensive emotional and social ramifications. Anxiety levels are high, especially in symptomatic patients (Cox et al, 1997). Cardiomyopathy is genetically inherited in many cases, giving rise to fear of transmission and issues surrounding genetic counselling.

Parents may feel guilty for passing on the condition and it can change family dynamics. The nurse is in an ideal position to help guide the family through the difficult stages of diagnosis and treatment and support the family through the screening process.

Many patients will be looked after in their own locality in heart failure clinics. It is vital that each patient continues to receive regular follow-up care and that their families have regular screening.

**Patient information**

Providing cardiomyopathy patients with accurate and timely information will help to dispel any misconceptions and help them to adapt their lifestyle to cope with the illness.

Patients who receive detailed information on their condition and treatment options and have help with lifestyle issues seem to cope with cardiomyopathy more easily than those who do not (Cox et al, 1997).

A diagnosis of cardiomyopathy can be potentially life-threatening and some patients may perceive their levels of activity to be severely curtailed, placing restrictions on everyday tasks.

For support information contact the Cardiomyopathy Association on 0800 0181 024 or visit their website at www.cardiomyopathy.org.

**REFERENCES**

