Clinical review - cardiomyopathy

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The cardiomyopathies are defined as a group of diseases in which the primary abnormality is one of the heart muscle. They are idiopathic by definition, and must be distinguished from the specific heart muscle diseases that have an identifiable cause, such as systemic hypertension.

The essentials
♥ Cardiomyopathy is an important cause of sudden death in young people
♥ Hypertrophic cardiomyopathy prevalence is 1 in 500 in young adults
♥ All types of cardiomyopathy tend to present insidiously
♥ Early recognition of cardiomyopathies can avert complications and death

Categories of cardiomyopathy
The cardiomyopathies are categorised according to their morphologic and haemodynamic characteristics. There are four groups: dilated, arrhythmogenic right ventricular, hypertrophic and restrictive cardiomyopathies. They all cause significant morbidity and mortality, and are among the most common genetically transmitted cardiac disorders. Early recognition is vital, as they can affect relatively young adults, have significant morbidity and mortality, and a high familial tendency.

1 Dilated cardiomyopathy

Characteristics
Dilated cardiomyopathy (DCM) is characterised by ventricular dilation and impairment of systolic function. It presents with heart failure, and to make the diagnosis, all other causes of ventricular dysfunction, particularly coronary artery disease and hypertension, must be excluded. The annual incidence is between five and eight cases per 100,000. In most cases the aetiology is unknown, but it can be associated with heavy alcohol consumption, pregnancy, cytotoxic agents and viral myocarditis. Thirty five per cent of cases of DCM are familial, so identifying affected relatives is important.

Symptoms and diagnosis
Effort-related fatigue and dyspnoea occur with heart failure, and chest pain occurs in about one third of cases. Examinations can be normal, or show heart failure and myocardial dysfunction. Right-sided heart failure occurs at the end stage and has a poor prognosis. The ECG is usually abnormal, but changes are non-specific, and a chest X-ray may show cardiac enlargement. An echocardiogram provides much more useful information.

Management of DCM
Whatever the cause of heart failure, the aim is to control symptoms, reduce left ventricle dimensions and prevent complications. Weight loss, stopping smoking, reducing alcohol intake and regular moderate exercise is important. Competitive sport is not advisable. Symptomatic therapy relies on digoxin, diuretics, and the early use of ACE inhibitors and beta-blockers.
In severe left ventricular dysfunction and those at risk of developing atrial fibrillation, anticoagulants can prevent systemic and pulmonary emboli. Patients at high risk of sudden cardiac death should be considered for an implantable cardioverter defibrillator. With progressive, non-responsive disease, a cardiac transplant may prove the ultimate lifeline.

2 Arrhythmogenic right ventricular cardiomyopathy

Characteristics
Arrhythmogenic right ventricular cardiomyopathy (ARVC) is associated with fibrofatty replacement of the right ventricular myocardium and/or left ventricle.

It has only been recognised relatively recently, and its incidence has not yet been established. But a study of sudden death in young people and athletes in northern Italy showed that previously undiagnosed ARVC seemed to be responsible in about 20 per cent of cases. About 50 per cent are familial and ARVC shows an autosomal dominant inheritance with incomplete penetrance.

In the absence of sustained ventricular arrhythmias, most patients will be asymptomatic and sudden death may be the first and only manifestation of the condition.

Symptoms and diagnosis
Symptoms include palpitations, syncope, ventricular arrhythmias and sudden death. The ECG may show inverted T-waves in the right praecordial leads and ventricular post-excitation (epilson) waves due to delayed right ventricular depolarisation. Echocardiography or magnetic resonance imaging show a dilated, poorly contractile right ventricle, with either a normal left ventricle or some degree of bilateral dysfunction. Histological confirmation of the diagnosis would be ideal, but this can be difficult because endomycardial biopsies are usually taken from the septal region for safety reasons, and this area is often unaffected in ARVC.

Management
Anti-arrhythmic therapy with beta-blockers or amiodarone is used to control the symptoms of patients with non life-threatening ventricular tachycardias. If these drugs do not help, then catheter ablation of the arrhythmogenic focus or surgery may help.

In patients at high risk of sudden death, an implantable cardioverter defibrillator should be considered. This is the treatment of choice in patients successfully resuscitated from a cardiac arrest. It is important to try to identify patients at risk. Asymptomatic patients should undergo baseline assessments, including Holter monitoring and exercise testing, to detect occult arrhythmias.

3 Hypertrophic cardiomyopathy

Characteristics
Hypertrophic cardiomyopathy (HCM) is characterised anatomically by unexplained myocardial hypertrophy, and histologically by myocyte disarray. Physiologically, impaired diastolic relaxation leads to an increase in the left ventricular diastolic pressure, with pulmonary congestion and dyspnoea.
The prevalence is about 1 in 500 young adults. More than 50 per cent are familial, with an autosomal dominant pattern of inheritance, and a high degree of penetrance. The history might suggest that about half the family have the disease, but ECG and echo studies show a familial prevalence closer to 90 per cent.

Genetic counselling should be offered at diagnosis, and relatives should be screened. Prognosis is generally good, but sudden death may occur in up to four per cent of adults and four to six per cent of children.

**Symptoms and diagnosis**

Common symptoms are dyspnoea and angina, fatigue, syncopy and palpitations. Some patients develop symptoms of congestive heart failure. Many patients are asymptomatic or have mild symptoms. They are often identified during screening of relatives. It is important to screen children who have relatives with HCM, as the mortality rate is higher in younger patients, and death is often sudden and unexpected.

Consider HCM in any child who presents with shortness of breath or unexplained syncope.

The echocardiogram is diagnostic, with left ventricular hypertrophy and a diminished ventricular cavity. The ECG is rarely normal, but is not typical. So any bizarre ECG in a young person should raise the possibility of HCM. Chest X-ray may show enlargement of the atria or the ventricle.

**Management**

Symptomatic management is reliant on beta-blockers and calcium antagonists. These reduce myocardial oxygen consumption and slow heart rate, so relieving angina and improving diastolic filling. A quarter of patients have left ventricular outflow tract obstruction caused by contact between the mitral valve and interventricular septum in systole. If outflow obstruction is severe and there is resistance to medical treatment, surgery or alcohol ablation of the interventricular septum may provide symptomatic relief. Atrial fibrillation should be treated with anticoagulants, and either a beta-blocker or verapamil to control the ventricular rate.

Patients who survive a cardiac arrest, or have episodes of sustained ventricular tachycardia, and young patients with a family history of multiple premature sudden deaths are at risk of sudden death. This is also true for those with recurrent syncope, an abnormal blood pressure response to exercise or severe left ventricular hypertrophy. Treatment with an implantable cardioverter defibrillator is required. Risk assessment is important in all patients, regardless of symptoms or lack of them, or the apparent severity of their disease based on morphology or haemodynamics.

4 **Restrictive cardiomyopathy**

**Characteristics**

Restrictive cardiomyopathy (RCM) is the least common of the cardiomyopathies. It is characterised by abnormal diastolic function in which the walls of the ventricles become excessively rigid and impede ventricular filling.

RCM is also thought to have a familial tendency, and research suggests the disease may be caused by the same genetic abnormalities that result in the more common hypertrophic cardiomyopathy.
Symptoms and diagnosis
Patients with RCM present with the signs and symptoms of heart failure such as fatigue, shortness of breath, oedema and abdominal distension. Up to one third may present with an embolic complication.

Arrhythmias and palpitations are also common. RCM diagnosis is usually based on a combination of physical examination, ECG findings and an echocardiogram. MRI can be useful to provide additional information about the structure of the heart. In some patients a precise diagnosis may require an endomyocardial biopsy.

As the onset of symptoms of RCM is often insidious, the diagnosis may be made late in the course of the disease. There is no specific treatment but medical therapy aims to improve the symptoms of heart failure, treat cardiac arrhythmias, and prevent thromboembolism.

Patients with bradycardia or heart block will need a pacemaker, and the most severe cases may require a cardiac transplant.

In any situation where familial disease is suspected, patients should be counselled and family screening offered.

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