

Recognising cardiomyopathy

Cardiomyopathy may be defined as a 'myocardial disorder in which the heart muscle is structurally and functionally abnormal, in the absence of coronary artery disease, hypertension, valvular disease and congenital heart disease sufficient to cause the observed myocardial abnormality'¹.

Subtypes of cardiomyopathy are defined by their characteristic pattern of muscle morphology and consequent impact on structure and function.

Current prevalence figures suggest that at least 1 in every 500 of the population has cardiomyopathy, although many are asymptomatic and go undiagnosed.

Patients with cardiomyopathy may present with a range of symptoms, or with a relevant family history pointing towards a genetic causation (see over page). In addition, cardiomyopathy may be an incidental finding whilst undergoing tests or procedures for other presenting conditions.

Accurate diagnosis and timely initiation of appropriate therapies is essential to manage symptoms and reduce disease burden, and to reduce potential sequelae such as arrhythmias, blood clots and consequent elevated stroke risk, heart block and heart failure. The risk of sudden cardiac death should also be considered.

Further reading

European Society of Cardiology practice guidelines: Ventricular Arrhythmias and the Prevention of Sudden Cardiac Death (2015), Diagnosis and management of hypertrophic cardiomyopathy (2014), Atrial Fibrillation (Management of) 2010 and Focused Update (2012).

NICE pathways: heart defects (structural), heart failure (acute and chronic), heart rhythm conditions, atrial fibrillation, structural heart defects at <http://pathways.nice.org.uk/>

NICE guidance and quality standards: cardiovascular conditions: general and other, heart failure, heart rhythm conditions, and structural heart defects at www.nice.org.uk/guidance

Further viewing

Watch Cardiomyopathy UK's videos for GPs on recognising the symptoms of cardiomyopathy (www.cardiomyopathy.org/recognise-symptoms) and supporting patients with cardiomyopathy (www.cardiomyopathy.org/support-patients)

Reference

1. Elliott P, et al. Classification of the cardiomyopathies: a position statement from the European Society Of Cardiology Working Group on Myocardial and Pericardial Diseases. Eur Heart J 2008;29:270–276

Take 2 minutes to assess risk factors for cardiomyopathy

Any patients positive for the following clinical symptoms, or with a relevant family history indicative of genetic cardiomyopathy, should be referred to cardiology services for evaluation and diagnosis.

Clinical symptoms

Symptoms include, but are not limited to: fatigue, breathlessness (particularly at rest, with minimal exertion or when lying prone), signs of pulmonary oedema on chest auscultation, oedema in ankles and abdomen, palpitations, and syncope (or near syncope) where these are not explained by other causes. Patients may also experience chest pain or tightness in the chest.

Family history

As cardiomyopathy is predominantly (although not exclusively) a genetic condition, investigation of the first-degree family history may identify the disease in family members. A lack of family history does not preclude a de novo mutation in the presenting patient, which should be considered. The five key questions to ask when taking a family history are as follows.

Are both parents still alive?

This determines any history of cardiac death in the parents. Previously occult cardiomyopathy may also become symptomatic in adulthood, so parental medical history is important.

Have there been any unexplained sudden deaths in family members under the age of 35?

Some cardiomyopathies carry a risk of sudden cardiac death, which often occurs at a young age (under 35).

Have there been any cot deaths?

As above, this question identifies any sudden cardiac death in the family.

Have there been any accidental deaths, such as by drowning or car accidents?

Some deaths recorded as accidental may mask a cardiac event which caused a loss of consciousness, leading to accidental death.

Has any family member been fitted with a pacemaker or are they being treated for any arrhythmia?

This is a positive indication of a potential genetic heart condition such as cardiomyopathy.

