Cardiomyopathies

What is a cardiomyopathy?

Cardiomyopathies are a heterogeneous group of pathologies characterised by structural and functional alterations of the heart. The heart muscle is structurally and functionally abnormal without coronary artery disease, hypertension, valvular or congenital heart diseases.[1]

The degree of cardiac dysfunction ranges from lifelong symptomless forms to major health problems, such as progressive heart failure, arrhythmia, thromboembolism and sudden cardiac death.[2]

- Primary cardiomyopathies - idiopathic and not attributed to a specific cause.

- Secondary cardiomyopathies - often associated with:
    - Chronic kidney disease, cirrhosis, and obesity, which have all been shown to result in impaired left ventricular function.[3]
    - Multisystem diseases - eg, sarcoidosis, amyloidosis, systemic lupus erythematosus, systemic sclerosis and polyarteritis nodosa.
    - Endocrine and metabolic - eg, diabetes, thyroid disease, acromegaly, haemochromatosis.
    - Drugs and chemicals - eg, cocaine abuse, alcohol abuse, some chemotherapy drugs.
    - Infection: American trypanosomiasis and some viral infections.
    - Nutritional: malnutrition, obesity, deficiencies - eg, vitamin B1, selenium, calcium and magnesium.
    - Specific cardiac abnormalities - eg, chronic uncontrolled tachycardia, hypertension, ischaemic heart disease, valvular dysfunction or abnormalities of the pericardium.
    - Genetic: familial forms of cardiomyopathy; cardiomyopathy is also associated with Duchenne muscular dystrophy.[4]
    - Peripartum cardiomyopathy develops between the last month of pregnancy and 5-6 months after delivery. Echocardiography demonstrates an idiopathic dilated cardiomyopathy. There is high morbidity and mortality.[5]

Types of cardiomyopathies (classification)

The four major types of cardiomyopathy are (see links for separate articles):

- Dilated cardiomyopathy: the most common form; the left or both ventricles are dilated with impaired contraction. Causes include: ischaemic, alcoholic, toxic, thyroid disorders, valvular, familial/genetic and idiopathic.

- Hypertrophic cardiomyopathy: the second most common; estimated adult prevalence is 1:500, with left and/or right ventricular hypertrophy. Usually familial (autosomal dominant).
- Restrictive cardiomyopathy: rare; estimated prevalence between 1:1,000 and 1:5,000, with restrictive filling and reduced diastolic filling of one/both ventricles and normal or near-normal systolic function. Causes include: amyloidosis, endomyocardial fibrosis, and idiopathic.
- Arrhythmogenic right ventricular cardiomyopathy (ARVC): a progressive genetic cardiomyopathy characterised by progressive fatty and fibrous replacement of ventricular myocardium. The cause is unknown; the familial form is usually autosomal dominant with incomplete penetrance but can be recessive.

Classifications have also included an unclassified group consisting of causes with no typical features of the above - eg, endocardial fibroelastosis, non-compacted myocardium, systolic dysfunction with minimal dilatation, mitochondrial diseases.

Cardiomyopathy epidemiology

- In contrast to coronary heart disease having a higher incidence in the elderly, cardiomyopathies can occur at younger ages. Therefore, cardiomyopathy should be suspected in any young person presenting with a heart failure, arrhythmias or thromboembolism.
- However, paediatric cardiomyopathies are rare diseases with an annual incidence of 1.1 to 1.5 per 100,000.[6]
- Clinically overt hypertrophic cardiomyopathy is the most common cause of sudden unexpected death in childhood.
- Restrictive cardiomyopathy is rare in childhood and has a poor outcome once symptoms develop.[7]
- A familial cause has been shown in 50% of patients with hypertrophic cardiomyopathy, 35% with dilated, and 30-50% with arrhythmogenic right ventricular cardiomyopathy. Restrictive cardiomyopathy is usually not familial.

Differential diagnosis

- Coronary artery disease.
- Mitral valve disease.
- Athlete's heart: prolonged isometric training may produce heart changes resembling some features of hypertrophic cardiomyopathy.
- Amyloidosis.
- Hypertensive heart disease: severe, chronic systolic and diastolic hypertension.
- Valvular and subvalvular aortic stenosis.
- Infundibular pulmonary stenosis.
- Ventricular septal defect.
- Constrictive pericarditis.

Investigations

- Blood tests: FBC, ESR, renal function tests, electrolytes, LFTs, cardiac enzymes and TFTs.
- CXR.
- ECG: a normal ECG is uncommon in any form of cardiomyopathy.
- Transthoracic Doppler echocardiography: this can confirm the diagnosis of hypertrophic cardiomyopathy, help to distinguish between restrictive cardiomyopathy and constrictive pericarditis, exclude valvular heart disease, and assess the severity of ventricular dysfunction in dilated cardiomyopathies.
- Brain natriuretic peptide (BNP) has a potential role as a test for ventricular dysfunction.
- Non-invasive stress testing is recommended only for patients who have a high probability of underlying ischaemic heart disease, prior myocardial infarction, or extensive hibernating myocardium or for evaluation for possible heart transplantation.

- Cardiac catheterisation can help in excluding coronary artery disease as the cause of the dilated cardiomyopathy and in distinguishing restrictive cardiomyopathy from constrictive pericarditis.
- Right ventricular angiography is considered a very useful test to diagnose classic forms of ARVC and to evaluate right ventricular function.
- Magnetic resonance imaging: this may help to distinguish between constrictive disease and restrictive cardiomyopathy.
- Right ventricular endomyocardial biopsy is occasionally used to distinguish between myocarditis and idiopathic dilated cardiomyopathy. A normal result does not rule out cardiomyopathy.

Cardiomyopathy treatment and management

Management may include genetic testing of family members, therapy for heart failure, appropriate activity restriction, evaluation for implantable cardioverter-defibrillator placement, and consideration of heart transplantation in refractory cases.[8]

- Treatment options are symptomatic and mainly directed towards treatment of heart failure and prevention of thromboembolism and sudden death.
- Identification of patients with high risk for major arrhythmic events is important because implantable cardioverter defibrillators can prevent sudden death.
- All patients with cardiomyopathy require a thorough cardiological assessment of functional capacity, cardiac function and risk of serious arrhythmia.
- Surgical myectomy for hypertrophic cardiomyopathy has demonstrated excellent long-term efficacy and safety at selected high-volume centres.[9]
- Heart transplantation may be necessary in patients with refractory heart failure.

Authored by:	Peer Reviewed by: Dr Hayley Willacy, FRCGP	
Originally Published: 20/11/2023	Next review date: 18/11/2022	Document ID: doc_1913

View this article online at: patient.info/doctor/cardiomyopathies

Discuss Cardiomyopathies and find more trusted resources at Patient.