

Cardiomyopathy

Cardiomyopathy is a term which indicates disease of the cardiac muscle of unknown cause.

Classification (based on clinical presentation)

- **DCM – Dilated cardiomyopathy**
- **HCM – Hypertrophic cardiomyopathy**
- **Restrictive cardiomyopathy**
- **Arrhythmogenic right ventricular cardiomyopathy**
- **Other rare cardiomyopathies:**
 - Conduction defect
 - Mitochondrial
 - LQTS
 - Brugada

Dilated Cardiomyopathy (DCM)

- DCM is characterized by dilation and impaired systolic function of the left and/or right ventricle, in the absence of underlying disease (e.g., IHD, hypertension, valve disease).
- The cause in the majority of cases is unknown (idiopathic).
- At least 25% of idiopathic cases are familial. More than 20 abnormal chromosome loci and genes are involved.
The majority are inherited as **autosomal dominant**, but **X-linked** or **recessive** cases can also be found.

Causes of Secondary Cardiomyopathy

- **Infections:** Viral (influenza, corona, coxsackie, adeno, erythro-viruses, HIV), bacterial (diphtheria), fungal, parasitic (toxoplasma, *T. cruzi*—Chagas disease, spirochaete—Lyme).
- **Toxins:** Alcohol, metals (cobalt, lead, mercury, arsenic), chemicals.
- **Drugs:** Chemotherapy, cocaine.
- **Autoimmune:** SLE, dermatomyositis, systemic sclerosis.
- **Metabolic:** CKD, cardiorenal syndrome.
- **Endocrine:** DM, thyroid heart disease, hyperparathyroidism, acromegaly, pheochromocytoma.
- **Neuromuscular:** Friedreich's ataxia, muscular dystrophy, myotonic dystrophy, neurofibromatosis.
- **Nutritional deficiency:** Beriberi, pellagra, scurvy, kwashiorkor.
- **Inflammatory (granulomatous):** Sarcoidosis.
- **Infiltrative:** Amyloidosis, Gaucher disease, Hurler's disease, Hunter disease.
- **Storage diseases:** Hemochromatosis, glycogen storage disease type II.
- **Other:** Tachycardia-induced cardiomyopathy, peripartum cardiomyopathy.

Clinical Features

Congestive heart failure; may also present with syncope, arrhythmia, conduction defect, heart block, systemic embolism (CVA), pulmonary embolism, or sudden death.

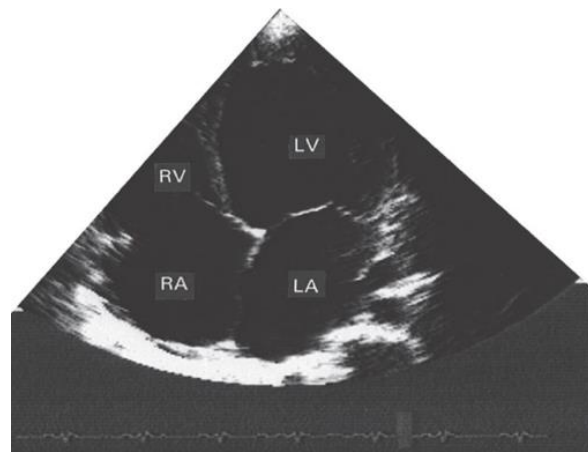
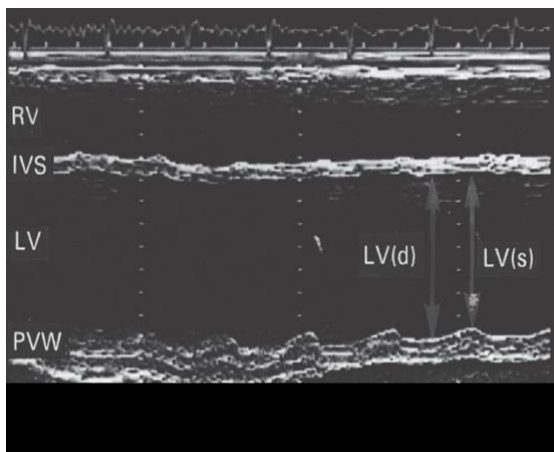
Investigations

- **Chest X-ray:** Enlarged cardiac shadow.
- **ECG:** Diffuse non-specific ST-T changes; may show sinus tachycardia, conduction defects, and arrhythmias (atrial fibrillation, ventricular premature beats, ventricular tachycardia).
- **Echocardiography:** Dilatation of the left and/or right ventricle.
- **Angiography (CATH):** Done to exclude coronary artery disease.
- **Biopsy.**

Treatment

Aim: relieve symptoms, retard disease progression, prevent HF complications.

- ACE inhibitors, ARB, ARNI, SGLT2 inhibitors
- Spironolactone (MRA), ivabradine, beta-blocker
- Anti-arrhythmic therapy
- Pacemaker, CRT, ICD, anticoagulant therapy
- LV/RV mechanical assist device
- Cardiac transplantation



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Hypertrophic Cardiomyopathy (HCM / HOCM)

HCM is characterized by **variable myocardial hypertrophy**, mostly involving the **interventricular septum**, with **disorganization (disarray) of cardiac myocytes and myofibrils**, in the **absence of clear causes** such as aortic stenosis (AS) or hypertension (HTN).

The majority of cases are **familial, autosomal dominant**, caused by **mutations in genes encoding sarcomeric proteins**.

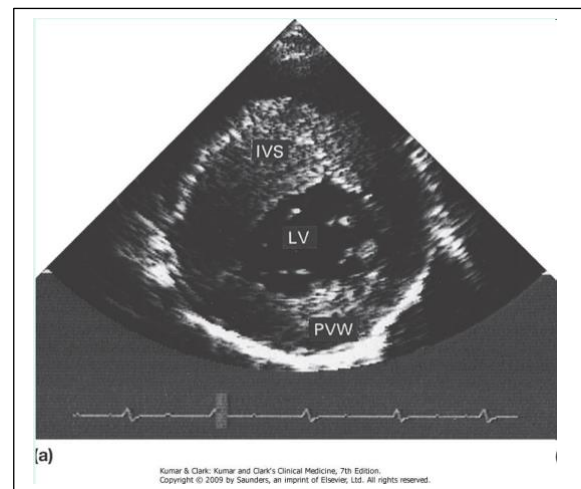
Clinical Features

Patients may present with:

- Asymptomatic
- Chest pain
- Dyspnea
- Syncope
- Cardiac arrhythmia
- Sudden death

Risk factors for sudden death:

1. IVS thickness > 3 cm
2. Syncope with EF ~50%
3. Arrhythmia
4. Age < 30 years
5. Family history of sudden death



Classic physical findings:

- Double apical pulsation
- Jerky carotid pulse
- Rapid ejection with LV outflow obstruction
- Fourth heart sound (S4)
- Ejection systolic murmur
- Pansystolic murmur of mitral regurgitation

Investigations

- **Chest X-ray:** Usually unremarkable
- **ECG:** LV hypertrophy, ST-T changes, Q waves in inferolateral leads
- **Echocardiography (diagnostic):**
 - Thick IVS
 - SAM (systolic anterior motion of anterior mitral leaflet)
 - Asymmetric LV hypertrophy
 - High pressure gradient across LV outflow tract
- **Pedigree analysis**
- **Exercise testing**

Treatment of Hypertrophic Cardiomyopathy (HCM / HOCM)

Medical Management

- **Chest pain and dyspnea:** Beta-blockers, Verapamil
- **Pre-syncope, syncope, arrhythmia:** Anti-arrhythmic drugs (Amiodarone, Flecainide, Propafenone)

Interventional / Surgical Management

- **ICD (Implantable Cardioverter Defibrillator):** Prevent sudden death
- **Dual-chamber pacing**
- **Alcohol septal ablation:** Induce MI in coronary artery to reduce IVS thickness
- **Surgical myectomy:** Relieve LV outflow obstruction

Note: Vasodilators should be **avoided**.

Restrictive Cardiomyopathy (RCM)

- No ventricular dilatation or muscular hypertrophy.
- Characterized by **resistance to diastolic ventricular filling**, resulting in heart failure (HF).

Secondary Causes

- Amyloidosis
- Sarcoidosis
- Loeffler's endocarditis
- Endomyocardial fibrosis

Clinical Presentation

- Dyspnea, fatigue
- Thromboembolic events
- Anasarca: elevated venous pressure, hepatomegaly, ascites, generalized edema
- Signs similar to **chronic constrictive pericarditis**

Investigations

- Chest X-ray
- ECG
- Echocardiography
- Cardiac catheterization: to distinguish from chronic constrictive pericarditis

Treatment

- No specific treatment; mainly **symptomatic**
- Cardiac transplantation in selected cases
- **Amyloidosis:** Melphalan + Prednisolone ± Colchicine