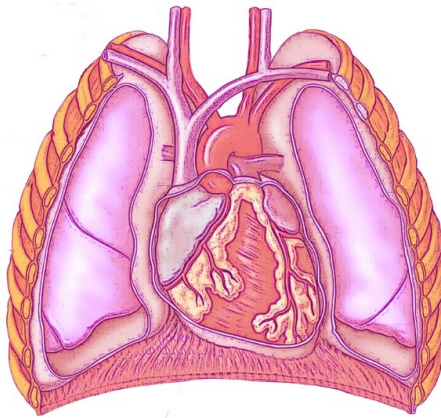




Internal Medicine



Topic : Cardiomyopathy Notes
Done by : Johainah Taha

Cardiomyopathy

Definition

Cardiomyopathy refers to a **disease of the cardiac muscle** of unknown cause. It is classified based on clinical presentation into:

1. Dilated Cardiomyopathy (DCM)
2. Hypertrophic Cardiomyopathy (HCM)
3. Restrictive Cardiomyopathy (RCM)
4. Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)
5. Other Rare Forms:
 - Conduction defects
 - Mitochondrial cardiomyopathy
 - Long QT Syndrome (LQTS)
 - Brugada Syndrome

Dilated Cardiomyopathy (DCM)

Definition & Pathophysiology

DCM is characterized by the dilatation and impaired **systolic function** of the left and/or right ventricle, in the absence of an underlying disease such as ischemic heart disease (IHD), hypertension, or valvular disease.

- The majority of cases are **idiopathic** (unknown cause).
- At least 25% of idiopathic cases are **familial**, involving **more than 20 abnormal chromosome** loci and genes.
- Most inherited cases follow an **autosomal dominant** pattern, though **X-linked** and **recessive** cases exist.

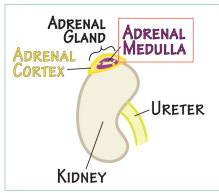
Causes of Secondary Cardiomyopathy

DCM can also be caused by various secondary factors:

Category	Examples
Infections	Viral: Influenza, Coronavirus, Coxsackie, Adenovirus, Erythroviruses, HIV Bacterial: Diphtheria, Fungal infections Parasitic: Toxoplasma, Trypanosoma cruzi (Chagas disease), Lyme disease
Toxins	Alcohol, heavy metals (cobalt, lead, mercury, arsenic), chemicals
Drugs	Chemotherapy agents, Cocaine
Autoimmune Diseases	Systemic lupus erythematosus (SLE), Dermatomyositis, Systemic sclerosis
Metabolic Disorders	Chronic kidney disease (CKD), Cardiorenal syndrome
Endocrine Disorders	Diabetes mellitus (DM), Thyroid disorders, Hyperparathyroidism, Acromegaly, Pheochromocytoma

Neuromuscular Diseases	Friedrich's ataxia, Muscular dystrophy, Myotonic dystrophy, Neurofibromatosis
Nutritional Deficiencies	Beriberi (Vitamin B1 deficiency), Pellagra (Niacin deficiency), Scurvy (Vitamin C deficiency), Kwashiorkor (Protein deficiency)
Inflammatory (Granulomatous) Disorders	Sarcoidosis
Infiltrative Disorders	Amyloidosis, Gaucher disease, Hurler's disease, Hunter's disease
Storage Disorders	Hemochromatosis, Glycogen storage disease (Type II)
Other Causes	Tachycardia-induced cardiomyopathy, Peripartum cardiomyopathy

Pheochromocytoma



CLINICAL PRESENTATION

Sx - Spont. Severe Anxiety
Sign - ↑Heart Rate & ↑BP

↑DOPAMINE

Agitation — Psychosis

↑NOREPI/EPINEPHRINE
"Fight or Flight"

BLOOD & URINE LABS
↑Catecholamines
↑Metanephrines

CATECHOLAMINE METABOLISM

↑DOPAMINE

↑3-MT & DOPAC

↑Homovanillic acid (HVA)

↑NOREPINEPHRINE
& EPINEPHRINE

↑Normetanephrine
& Metanephrine

↑Vanillylmandelic acid (VMA)

The limited symptoms of scleroderma are referred to as **CREST**

Calcinosis- calcium deposits in the skin

Raynaud's phenomenon-
spasm of blood vessels in
response to cold or stress

Esophageal dysfunction- acid reflux and
decrease in motility of esophagus

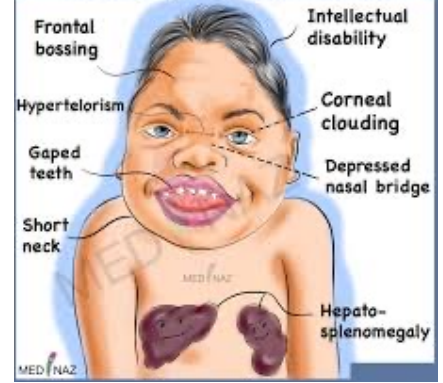
Sclerodactyly- thickening and tightening
of the skin on the fingers and hands

Telangiectasias- dilation of
capillaries causing red marks
on surface of skin



ADAM

HURLER SYNDROME



Signs & Symptoms Of Hunter Syndrome

- Nose becomes broad
- Tongue is enlarged
- Cheeks become enlarged and rounded
- Lips thicken
- Enlarged head
- Hearing loss
- Heart valve issues
- Stiffness in joints
- Growth is restricted
- Compressed and damaged spinal cord



Picture A: Coarse facies including frontal bossing, broad nose, heavy eyebrows and thick lips



Picture B: Stooped posture and toe-walking are signs of skeletal dysostosis and contractures

Hemochromatosis: Genes Involved in Iron Overload

Causes of Hemochromatosis

- Genetic Mutations
- Excessive blood transfusions

HFE Mutations

HFE Gene
Chromosome 6

C282Y
H63D
S65C

High risk

Lower Risk

C282Y/C282Y
C282Y/H63D
C282Y/-
H63D/H63D
H63D/S65C
H63D/-

HFE Gene

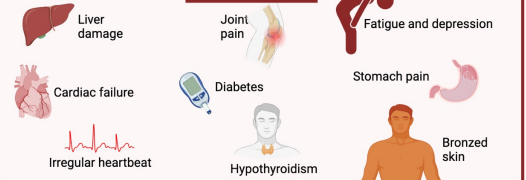
Controls iron absorption in the
intestines
Mutations lead to excessive iron
absorption

Building Up Iron

Iron is essential for oxygen
transport and needed at the
right level.

Excess iron gets stored in
organs and can cause
inflammation and cell death.

Health Effects



Genetic Lifehacks
Learn. Experiment. Optimize.

Symptoms of Chronic Determinate Chagas Disease

Chagas Heart Disease

fatigue and
syncope

shortness of breath

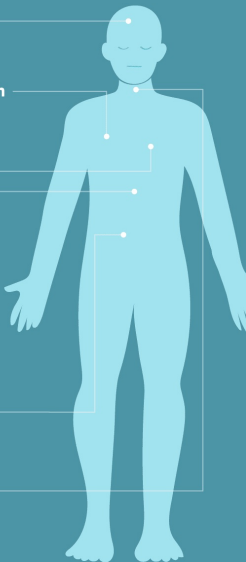
palpitations

Chagas GI Disease

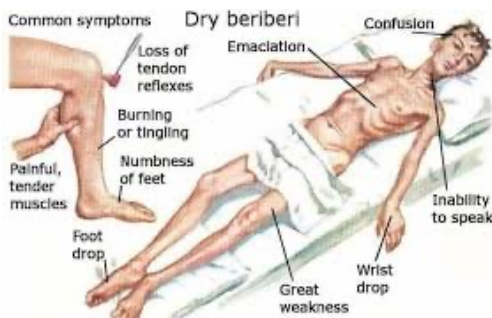
GI reflux

severe
constipation

difficulty and
pain swallowing



verywell



Hemochromatosis - Arthropathy

Clinical Features

Patients may present with congestive heart failure symptoms and additional complications such as:

- Syncope
- Arrhythmias
- Conduction defects (heart block)
- Systemic embolism (e.g., stroke)
- Pulmonary embolism
- Sudden cardiac death

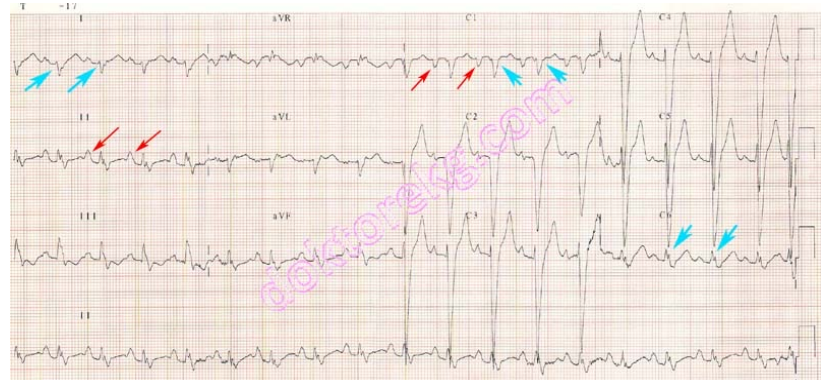
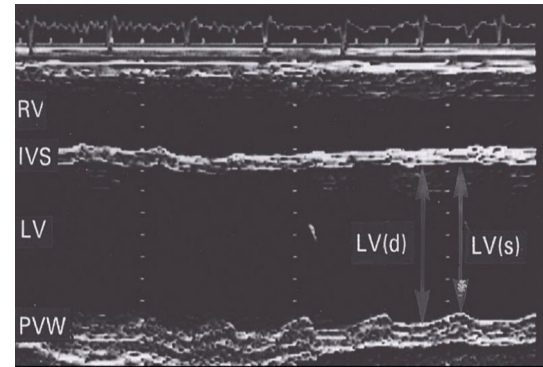
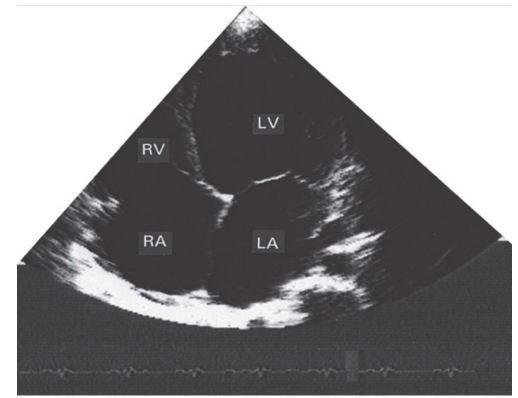
Investigations

Test	Findings
Chest X-ray	Enlarged cardiac silhouette (cardiomegaly)
ECG	Diffuse, non-specific ST-segment and T-wave changes, sinus tachycardia, conduction defects, arrhythmias (atrial fibrillation, ventricular premature beats, ventricular tachycardia)
Echocardiography	Left and/or right ventricular dilatation
Coronary Angiography (Catheterization)	Done to exclude coronary artery disease
Endomyocardial Biopsy	Occasionally performed for further evaluation

Treatment Goals

- Relieve symptoms
- Slow disease progression
- Prevent complications

Treatment Type	Examples
Heart Failure Medications	ACE inhibitors, ARBs, ARNI (Sacubitril/Valsartan), SGLT2 inhibitors
Aldosterone Antagonists	Spironolactone, Mineralocorticoid Receptor Antagonists (MRAs)
Heart Rate Control	Ivabradine, Beta-blockers
Anti-arrhythmic Therapy	Used in specific cases
Device Therapy	Pacemaker, CRT (Cardiac Resynchronization Therapy), ICD (Implantable Cardioverter-Defibrillator)
Advanced Therapies	LVAD (Left Ventricular Assist Device), RVAD (Right Ventricular Assist Device), Cardiac transplantation



Hypertrophic Cardiomyopathy (HCM or HOCM)

Definition & Pathophysiology

HCM is characterized by:

- Myocardial hypertrophy (thickening of the heart muscle), predominantly involving the interventricular septum.
- Disorganization (disarray) of cardiac myocytes and myofibrils.
- Absence of secondary causes like aortic stenosis (AS) or hypertension (HTN).
- Mostly **familial, autosomal dominant** inheritance due to mutations in genes encoding **sarcomeric proteins**.

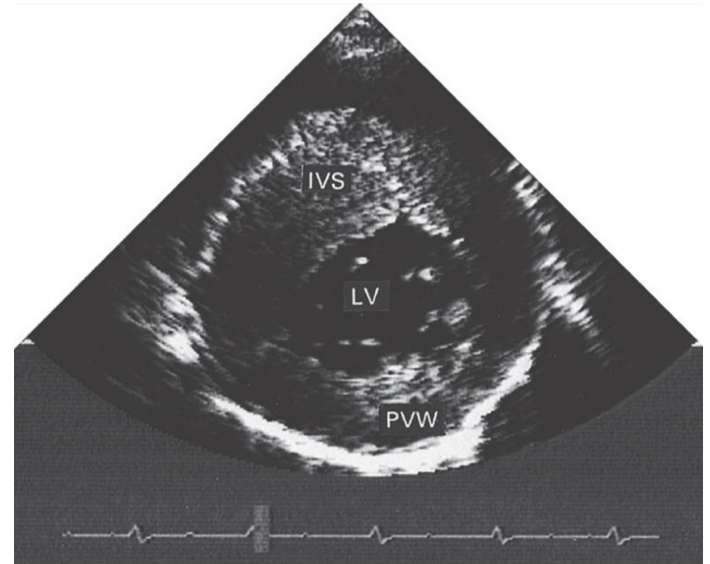
Clinical Features

Patients may present with:

- Asymptomatic cases
- Chest pain
- Dyspnea
- Syncope
- Arrhythmias
- **Sudden cardiac death (SCD), especially in:**

1. IVS thickness >3 cm
2. History of syncope
3. Presence of arrhythmias
4. Young age (<30 years)
5. Family history of SCD

*Very important
for history taking*



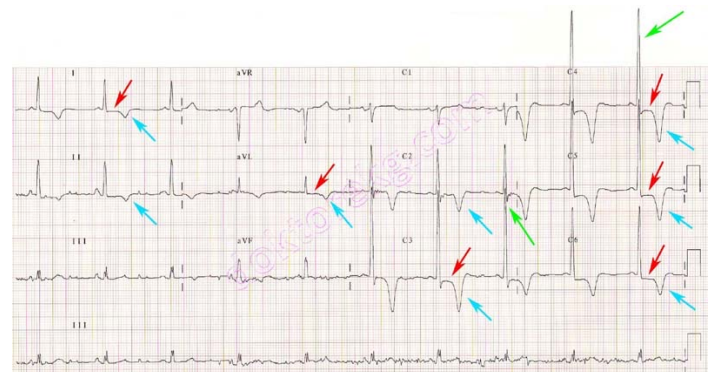
a)

Physical Examination Findings

- Double apical impulse
- Jerky carotid pulse
- Rapid ejection and sudden left ventricular outflow obstruction
- Fourth heart sound (S4)
- Ejection systolic murmur
- Pansystolic murmur of mitral regurgitation

Investigations

Test	Findings
Chest X-ray	Usually normal (<i>unremarkable</i>)
ECG	Left ventricular hypertrophy (LVH), ST and T wave changes, Q waves in inferolateral leads
Echocardiography	Diagnostic shows thickened IVS, systolic anterior motion (SAM) of the anterior mitral leaflet, asymmetric LV hypertrophy, and high-pressure gradient across LV outflow tract
Other Tests	Pedigree analysis, Exercise testing



Treatment

Symptom	Treatment
Chest pain, dyspnea	Beta-blockers, Verapamil
Pre-syncope, syncope, arrhythmias	Anti-arrhythmic drugs (Amiodarone, Flecainide, Propafenone)
Sudden cardiac death prevention	ICD implantation
Other Interventions	Dual-chamber pacing, Alcohol ablation (to induce controlled MI and reduce IVS thickness), Surgery for severe cases
Avoid	Vasodilators

Restrictive Cardiomyopathy (RCM)

Definition

RCM is characterized by impaired diastolic ventricular filling due to increased myocardial stiffness, without ventricular dilatation or hypertrophy. *leading to HF.*

Secondary Causes

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

Clinical Features

- Dyspnea
- Fatigue
- Thromboembolic events
- Anasarca (generalized edema)
- Signs similar to chronic constrictive pericarditis (elevated venous pressure, hepatomegaly, ascites)

Investigations & Treatment

Investigation	Purpose
Chest X-ray, ECG, Echocardiogram	Initial evaluation
Cardiac Catheterization	Differentiates RCM from constrictive pericarditis

Treatment is mainly symptomatic. In some cases:

- Cardiac transplantation may be considered.
- Amyloidosis treatment: Melphalan + Prednisolone ± Colchicine.