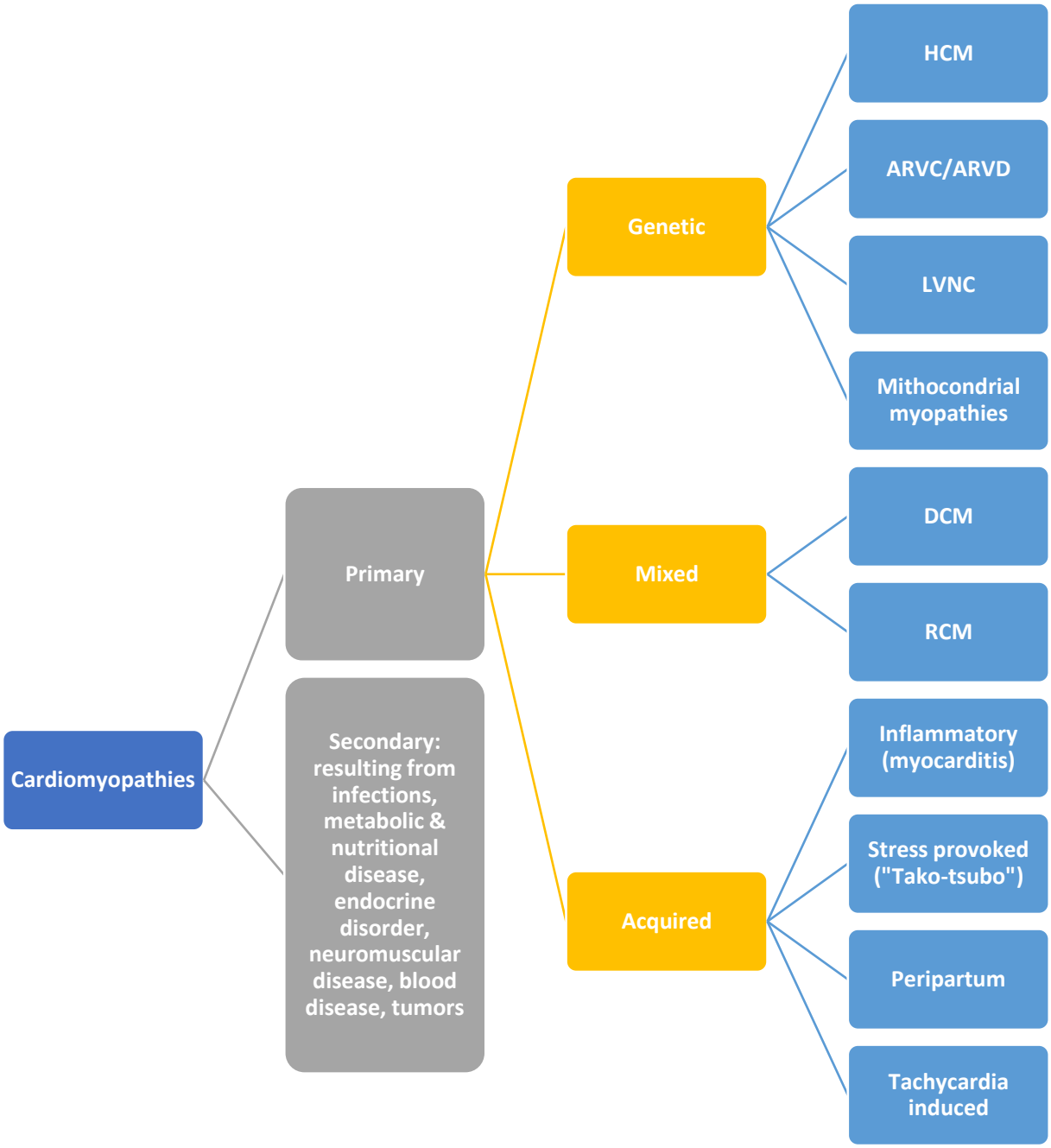
Cardiomyopathies

Definition: Cardiomyopathy implies that the myocardium is structurally and functionally altered, and the pathological changes are not explained by coronary heart disease, hypertension, valvular disease or congenital heart disease, inflammation in myocardium (these conditions should be excluded). Cardiomyopathy leads to impaired cardiac function and heart failure.

Types

Cardiomyopathies	Other names	Leads to
Dilated Cardiomyopathy (DCMP)	Congestive Cardiomyopathy	Impaired LV Diastolic Function
Hypertrophic Cardiomyopathy (HCM)	**Obstructive variant**: – Idiopathic Hypertrophic Subaortic Stenosis (IHSS) – Asymmetrical Septal Hypertrophy (ASH) **Non-Obstructive variant** – Apical hypertrophic cardiomyopathy (AHCM or ApHCM) also called Yamaguchi syndrome ("**Spade Shaped Configuration**")	Impaired LV Diastolic Function
Restrictive Cardiomyopathy (RCM)	Infiltrative Cardiomyopathy	Impaired LV Diastolic Function
Takotsubo Cardiomyopathy (TTC)	Stress Induced Cardiomyopathy; Broken Heart Syndrome; Apical Ballooning Syndrome	Impaired LV Systolic Function
Non-compaction cardiomyopathy (NCCM)	Left Ventricular Noncompaction (LVNC)	Most Commonly Asymptomatic; may leads to LV dysfunction (systolic & diastolic), SCD, thromboembolic phenomenon & malignant arrhythmia
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	Arrhythmogenic Right Ventricular Dysplasia (ARVD)	Impaired RV systolic Function

ACC/AHA Classification



Prevalence

	Children	Adults (19-64 years)
DCMP	Uncommon	1:250/500
HCM	Uncommon	1:250/500
RCM	Uncommon	Uncommon

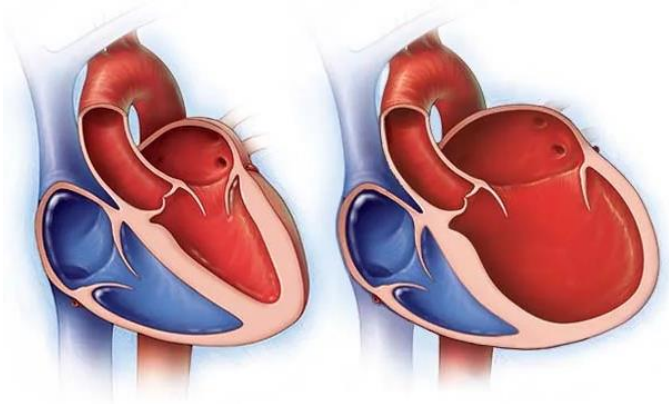
- Global Prevalence: It's estimated that around 6 million people worldwide are affected by various types of cardiomyopathies.
- The hospital-based prevalence of cardiomyopathy is approximately 809 per million inhabitants per year.

Dilated cardiomyopathy (DCM)

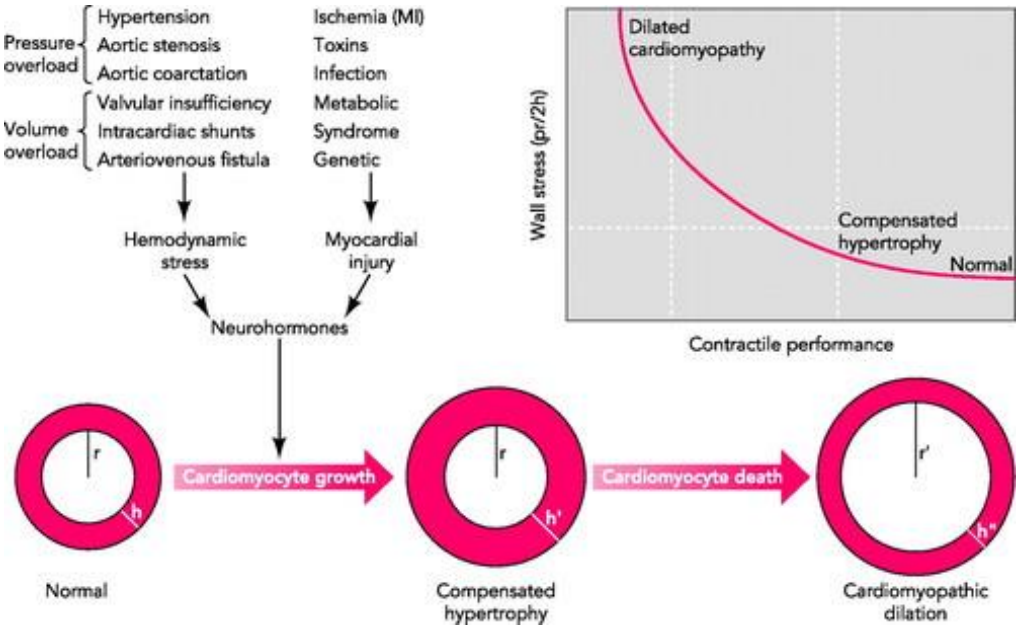
Dilated cardiomyopathy (DCM) is primary myocardial disease & is defined as the dilation of one or both ventricles. Dilation of the left ventricle is virtually always accompanied by impaired left ventricular systolic function, globally hypokinetic.

Key Features:

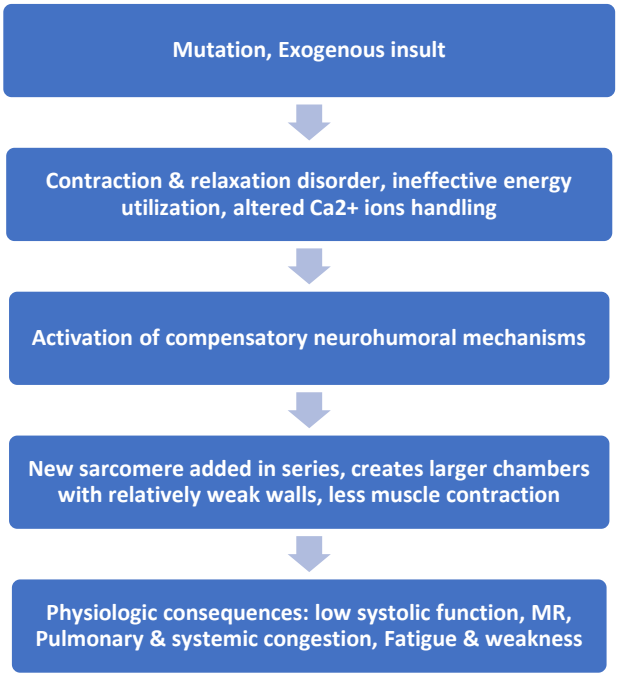
- Biventricular Failure
- Pathologically Dilated both Chambers (Eccentric Hypertrophy, Balloon like cavity)
- Global Impairment in Contraction



Pathogenesis



Pathophysiology



Prevalence

- Most patients present in age group 20 to 60 years (1:250/500) even it can be found in infants (uncommon)

Presentation

- Incidental finding
- Symptomatic
- Survivors of SCD

Etiology/Causes

- **A**lcoholism (Regular alcohol consumption >80 g/day for more than 5 years confers a high risk of developing dilated cardiomyopathy. There are large individual variations regarding the amount of alcohol required to cause cardiomyopathy), **A**denovirus, Obstructive sleep **A**pnea, **A**cromegaly, **A**utoimmunity (AHA- Anti Heart Antibodies)
- **B**eriberi (B1 deficiency)
- **C**ocaine, **C**oxsackie B, **C**hagas (American trypanosomiasis, is a **tropical parasitic disease caused by Trypanosoma cruzi)**, **C**irrhosis, **C**ardiomyopathies (other types of cardiomyopathies like ischemic, Alcohol, Diabetic, Takotsubo, Tachycardia induced, non-compaction, peripartum cardiomyopathy)

- **D**oxorubicin
- **E**cho virus
- **F**amilial (genetic): *TTN mutation → defective Titin (sarcomere protein). Approximately 40% of all cases of DCM are genetic.
- **G**lue sniffers: The chemicals in the substance can sensitize the heart to adrenaline. This can result in a very fast heartbeat that causes heart failure
- **H**emochromatosis, **H**yper/**H**ypothyroidism, **H**IV, **H**HV-6
- **I**schemic heart disease: *Ischemic cardiomyopathy should be suspected if there is significant stenosis (>75% luminal obstruction) of the left main coronary artery (LAD) or >2 epicardial coronary arteries,* **I**diopathic dilated cardiomyopathy
- **M**ELES syndrome: mitochondrial myopathy, encephalopathy, lactic acidosis & stroke
- **P**regnancy (**P**eripartum cardiomyopathy), **P**ost myocarditis
- **S**arcoidosis, **S**LE, **S**VTs
- **T**B, **T**oxoplasmosis, **T**richinosis

Signs & Symptoms

Symptoms are similar to congestive heart failure

- Right sided heart failure due to DCMP
  - Raised Jugular Venous Pressure
  - Cardiac Cirrhosis – Hepatomegaly (congestion of liver)
  - Lower limb oedema
- Left sided heart failure due to DCMP
  - Pulmonary congestion due to pulmonary oedema
  - Morning Cough
  - Shortness of breath (Dyspnoea, orthopnoea, paroxysmal nocturnal dyspnoea)
  - Sometimes haemoptysis & even repeated chest infections
- Cardiomegaly
- Decreased Pulse Pressure
- Myocardial Dilation leads to valvular regurgitation (mitral & tricuspid regurgitation) → S3 Heart Sound
- Ventricular arrhythmias: stretching muscles irritates conduction system.
- Decreased Cardiac Output (Dec. EF) → Dec. BP
- Reduced capillary refill, cold clammy skin

Diagnose

- **Clinically**
  - **Auscultation**
    - ~ Pansystolic/ Holosystolic murmur (mitral regurgitation during systole)
    - ~ S3 sound (blood rushing into, dilated ventricular wall during diastole)
    - ~ Wet crackles

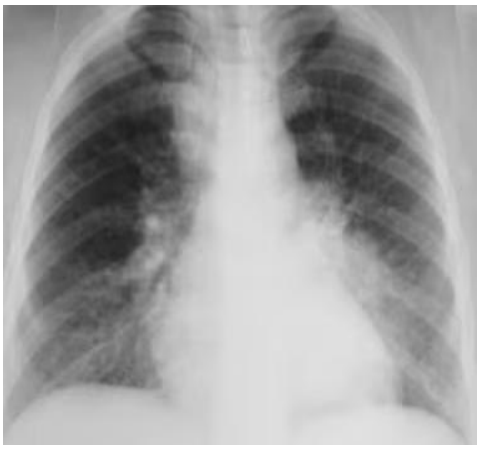
- **ECG**: There are no specific features unique to DCM, however the ECG is usually not normal
  - Left Atrial Enlargement (may progress to AF) or Bi atrial enlargement
  - Sings of left ventricular hypertrophy or Bi ventricular hypertrophy
  - LBBB or RBBB occurs due to cardiac dilation
  - Left Axis Deviation
  - Poor R wave progression with QRS complexes in V1-V4 ("pseudo-infarction" pattern)
  - Diffuse myocardial fibrosis may lead to reduced voltage QRS complexes, particularly in the limb leads.
  - Ventricular dysrhythmias (VF/VT)
  - Frequent ventricular ectopic (seen with severe DCM)
  - Non-specific IVCD
  - Non-specific ST-T changes

- **ECHO**
  - **In dilated cardiomyopathy (DCM), both ventricles are dilated**
  - **Left ventricular systolic function is impaired** (i.e., ejection fraction is reduced). Myocardial **contractile function is globally impaired (i.e., general hypokinesia exists)**. Regional wall motion abnormality can be seen in the septum in the setting of left bundle branch block.
  - **Left ventricular diastolic function may be normal or exhibit a restrictive pattern** (increased E/A ratio and rapid deceleration time; see Diastolic function).
  - Left ventricular wall thickness may be normal, but since the ventricle is enlarged, **the ventricular mass & volume is always increased**. LV mass is uniformly increased

- Although the ejection fraction is reduced in DCM, **stroke volume may be normal due to the large ventricular volume** (SV may be preserved despite of reduced EF)**.** Symptoms may, therefore, not manifest until advanced stages of cardiomyopathy, when stroke volume is declining.
- Ventricular dilation leads to dilation of the mitral annulus and tricuspid annulus, resulting in **mitral regurgitation** (annular dilation leads to incomplete MV coaptation & papillary muscles are apically displaced "Functional MR") and **tricuspid regurgitation**.
- Pronounced ventricular dilation and impaired contractility lead to slow blood flow in the ventricular cavity. This may result in **spontaneous echo contrast and the appearance of thrombi** in the ventricle.

- **Chest X-ray**

  - **Cardiomegaly**: This is the most prominent feature and refers to an enlarged heart. The heart's silhouette appears enlarged due to the dilation of the ventricles.
  - **Pulmonary Congestion**: ***cephalization of vasculature*** Increased vascular markings and pulmonary oedema can be seen, particularly in the upper lobes, due to elevated pressure in the pulmonary circulation.
  - **Pleural Effusion**: Fluid may accumulate in the pleural spaces, often seen at the lung bases.
  - **Kerley B Lines**: These are horizontal lines seen at the lung periphery, indicating interstitial oedema.
  - **Prominent Pulmonary Arteries**: Due to increased pressure and flow, the pulmonary arteries may appear enlarged.
  - **Left Ventricular Enlargement**: Specifically, the left ventricle may appear more prominent compared to the right side.



- **Troponin:** could be raised

Differential Diagnosis

Many of the symptoms of DCM are also common to other end-organ diseases such as:

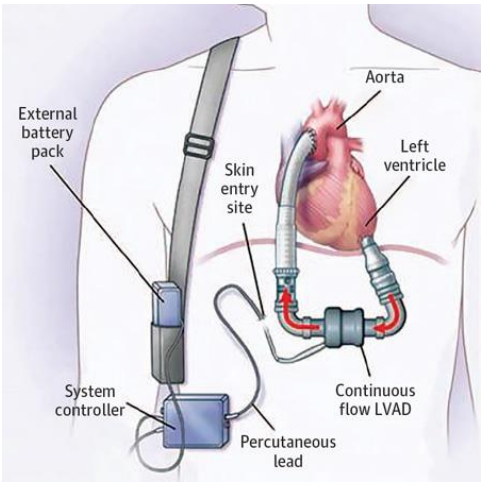
- Dysphonia (lung disease)
- Ascites, Peripheral Oedema (Cirrhosis)
- Volume Overload (Renal Failure)
- Fatigue (Hypothyroidism)
- Also, the diagnosis of DCM in young patients is often delayed because of new onset asthma or chronic bronchitis/pneumonia – either of which results in dyspnoea & fatigue as the main presenting symptoms

Treatment

Treatment of underlying causes

- **Diet:** low Na+, Exercise
- **Medical**:
  - Beta-blockers (metoprolol), ACEi (lisinopril), ARBs
  - Aldosterone inhibitors (spironolactone), SGLT2 inhibitors (Dapagliflozin)
  - Digoxin
  - Hydralazine, Nitrates
- **Surgical**
  - **Biventricular pacemaker (CRT):** this device is for people who have heart failure & irregular heartbeats. It stimulates both of the lower heart chambers to make the heart beat better.
  - **Implantable cardioverter defibrillator (ICD):** an ICD doesn't treat cardiomyopathy itself. It monitors the heart rhythm & delivers electrical shocks if an irregular heartbeat (Arrhythmia) is detected. Cardiomyopathy can cause dangerous arrhythmias, including those that cause the heart to stop. When LVEF 35% or less. The timing of insertion is different depending on the Etiology: >40 days post-MI or after revascularization for patients for ICM, & >3 months for patients with nonischemic DCM on optimal therapy.

- **Left Ventricular Assist Device (LVAD):** this mechanical device helps a weakened heart pump better. A LVAD usually is considered after less invasive approaches are unsuccessful. It can be used as a long-term treatment or as a short-term treatment while waiting for heart transplant.
- If medications & other treatments for DCM no longer work, a **heart transplant** may be needed.

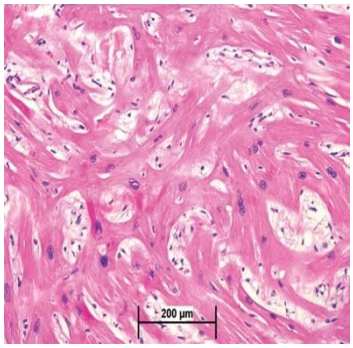


Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy is characterised by the presence of an asymmetrical increase in **left ventricular wall thickness**, not solely explained by **abnormal loading conditions** (commonly hypertension and aortic stenosis where LV hypertrophy is symmetrical).

Key Features:

- **Dynamic LVOT Obstruction** (intermittent LVOT obstruction)
- **Asymmetrical hypertrophy** especially upper part of IVS is grossly hypertrophic then other part which bulges during systole to obstruct LVOT
- **Hyperkinetic/Hyperdynamic left ventricle**
- Pathological features include
  - **Myocardial fibrosis/disarray** (see fig.)
  - **Abnormal coronary microcirculatory** function.
- Systolic anterior motion: **SAM** of the mitral valve (When the septum bulges into the LVOT, hemodynamics change in the outflow tract, which leads to the anterior leaflet of the mitral valve being sucked into the LVOT)



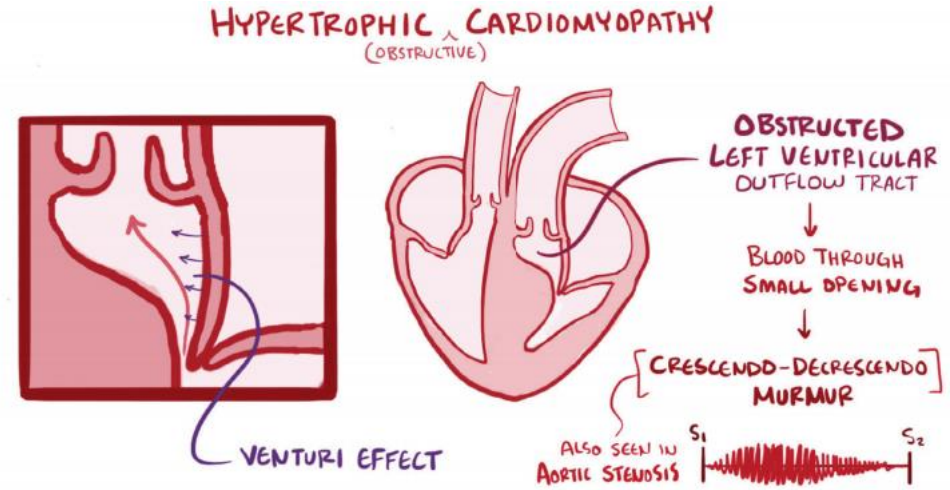
- In hypertrophic obstructive cardiomyopathy (HOCM), **the left ventricle (LV) can exhibit** a unique shape on imaging. Specifically, the LV silhouette may resemble a **kidney or even a banana**. This distinctive appearance is due to the hypertrophied myocardium and the presence of LV outflow tract (LVOT) obstruction.
- Hypertrophic cardiomyopathy has a **bimodal peak occurrence** most commonly presenting in the third decade of life.
  - **In adolescence and young adulthood** (often during the third decade of life, which is ages 20-29)
  - **Later in life** (commonly between ages 50-60)
- Most Common Cause of **Sudden Cardiac Death**

Prevalence

- Hypertrophic cardiomyopathy has an estimated prevalence of 1 in 500 and most commonly presents in young adults.
- Among athletes, hypertrophic cardiomyopathy is the most common cause of sudden cardiac death.

Risk factors

The only risk factor for hypertrophic cardiomyopathy (given it is a mostly inherited condition) is **family history**.



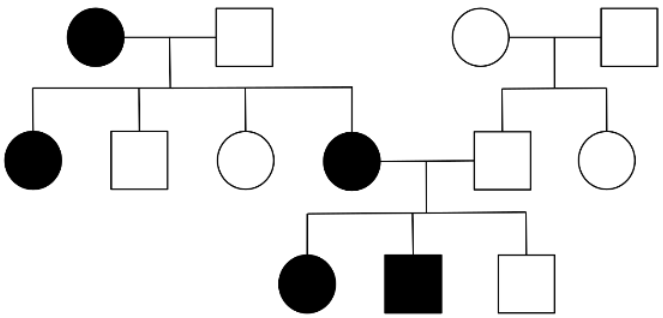
Causes

Hereditary (Genetic)

- Typically inherited via **autosomal dominant pattern** with mutations in **cardiac sarcomere protein** genes (e.g., beta-myosin heavy chain, myosin-binding protein C & cardiac troponin C).
  100% genetic defect where 50% is sporadic (new mutations), 50% family history (inherited mutations)

Others:

- Fabry's, Pompe's, Mitochondrial disease, Noonan/LEOPARD syndromes (rasopathies), Friedrich ataxia.
- Hypertrophy is either secondary to functional and structural defects in myocytes or increased storage of materials, such as glycogen



A *typical pedigree for autosomal dominant inheritance.*

Signs & Symptoms

Most affected individuals do not experience substantial symptoms and remain undiagnosed.

Typical **symptoms** of hypertrophic cardiomyopathy include:

- **Dyspnoea (90%):** due to left ventricular diastolic dysfunction and resultant pulmonary oedema.
- **Syncope and presyncope**: due to inadequate cardiac output (especially on exertion) or arrhythmias. **Patients with syncope are at high risk of sudden cardiac death**.
- **Chest pain (Angina):** due to microvascular complications of the disease or mismatch between increased oxygen requirement of the hypertrophied myocardium and reduced perfusion of coronary arteries due to impaired diastolic relaxation & LVOT obstruction.
- **Palpitations:** due to arrhythmias, both supraventricular and ventricular in origin.
- Symptoms mimics Congestive Heart Failure
    - Right Sided Heart Failure:
        - Raised Jugular venous pressure
        - Hepatomegaly
        - Lower limb oedema
    - Left Sided Heart Failure
        - Pulmonary Oedema
        - Cough, Orthopnoea
        - Haemoptysis

Diagnoses

History

- Family history: sudden cardiac death, unexplained heart failure, cardiac transplantation or implantable cardioverter-defibrillator (ICD) insertion.

Clinical Examination

In patients with suspected hypertrophic cardiomyopathy, a thorough cardiovascular examination is required.

Examination is often unremarkable and findings can be unspecific, however, typical **clinical findings** in hypertrophic cardiomyopathy include:

- **Harsh systolic crescendo decrescendo murmur** loudest between the apex and left sternal border: indicative of left ventricular outflow tract obstruction (accentuated with Valsalva manoeuvre).
- Fourth heart sound (S4) of atrial systole against a non-compliant ventricle – **S4 gallop**.
- **Paradoxical splitting** of the **S2** (second heart sound).
- Holosystolic murmur loudest at the apex or axilla: indicative of mitral regurgitation.
- "**Triple Riple** (Atrial contraction, early rapid ejection, late slow ejection)" Triple or Double apical beat of ventricular contraction and left atrial contraction against hypertrophic ventricle.
- Lateral displacement of the apical pulse.
- Pulses Bisferiens
- Prominent *a* wave: indicative of reduced right ventricular compliance with massive left ventricular hypertrophy.

Laboratory Investigations

- Complete blood count: to look for anaemia.
- Thyroid function tests: thyroid disease can exacerbate left ventricular dysfunction.
- NT-proBNP: quantify the degree, if any, of heart failure.

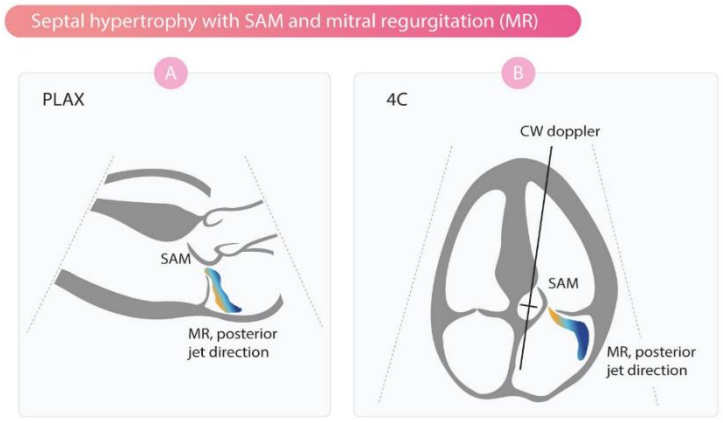
ECG

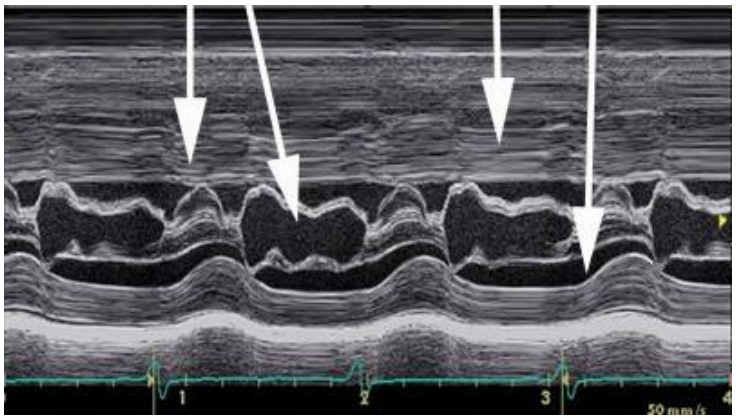
- **LVH** with increased precordial voltages & non-specific ST segment & T wave abnormalities
- **Deep, narrow ("dagger like") Q waves** in lateral leads (I, aVL, V5, V6) $^{+}/_{-}$ inferior leads (II, III, aVF) leads. Lateral Q waves are more common than inferior Q waves in HCM.
  - *Infarction Q waves are typically >40 ms duration*
  - *Septal Q waves in HCM are <40 ms*
- **Other associated features may include:**
  - **P-mitral** left atrial enlargement due to LV diastolic dysfunction
  - Signs of WPW (short PR, delta wave)
  - Dysrhythmias: AF & SVT are common; PACs, PVCs, VT

- **Apical Hypertrophy:** relatively uncommon form of HCM is seen most frequently in Japanese patients. There is localized hypertrophy of LV apex, causing a "**spade shaped**" configuration of the LV cavity.
  - The classical ECG findings in apical HCM is **Giant TWI (T wave inversions)** in the precordial leads.

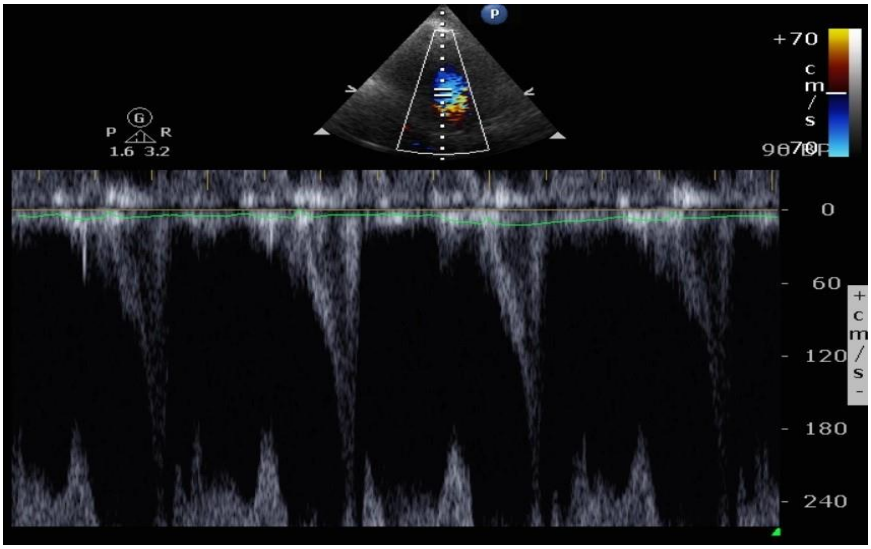
ECHO

- LV cavity is crescent-shaped, kidney or even a banana shaped.
- LV systolic function is usually normal to supranormal (also termed hyperdynamic).
  - LVEF is often >70%. The reason for this hypercontractile state is unclear.
- LV hypertrophy (LV mass) is typically asymmetric.
  - Wall thickness >1.5 cm, (1.3-1.5 cm is considered borderline), typically in the basal anteroseptum.
  - ASH: ratio of septum to posterior wall of 1.3:1
- SAM of the anterior mitral leaflet towards the septum
  - Best seen in PLAX 2D or M-Mode
    - ~ May see mid-systolic contact of anterior leaflet with the septum in severe obstruction.





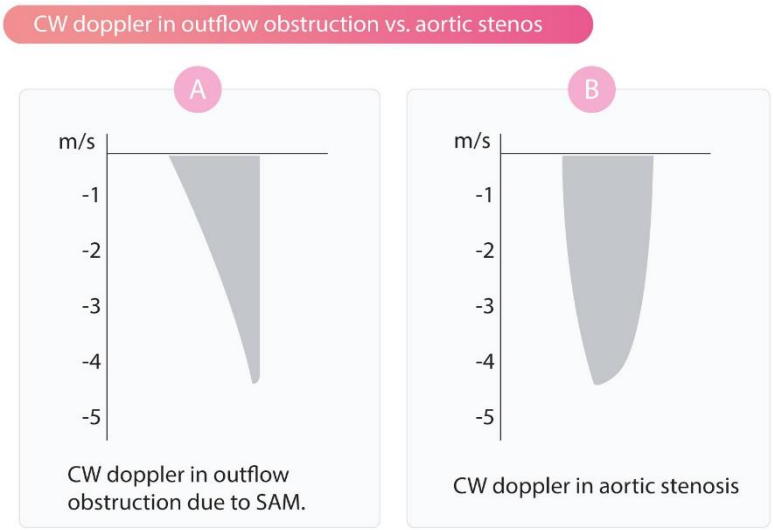
- Eccentric, posteriorly directed MR jet secondary to SAM (causing incomplete leaflet apposition), as the AML is pulled away from the PML the MR is increases. It also appears mid-to-late peaking (may confused with dynamic LVOT gradient)



Mid-to-late peaking appearance of MR

- Dynamic LVOT obstruction due to bulging basal anteroseptum may lead to mid-systolic closure of the aortic valve (seen in 2D & M-Mode)
  Note that this finding can also be seen with fixed LVOT obstruction

  - CW Doppler will show late-peaking Doppler envelope ("broad blade dagger" shape).
  - Dynamic LVOT gradient ≥30 mmHg (velocity ≥2.7 m/sec)
  - Intracavitary LV gradients resulting from hyperdynamic LV function & small LV cavity.



Intracavitary "scythe-shaped", late-peaking gradient

Apical Variant of HCM

Accounts 15-25% of HCM in Japan; much less prevalent in western populations

ECHO Features

- Apical hypertrophy
  - Results in the typically "spade-shaped" configuration of the LV at end-diastole
  - Best seen in A2C or A3C view
  - Contrast enhancement may be necessary to determine the full extent of the apical hypertrophy
- Typically, there is no LVOT obstruction

Chest X-ray

Chest radiographic findings may be normal or reflect nonspecific features

- **Cardiomegaly**: Enlarged heart silhouette, although it may not be as pronounced as in other cardiac conditions because the thickening is often more localized.
- **Pulmonary Congestion**: In cases where there is associated heart failure, signs of pulmonary congestion or oedema might be present.

MRI

Cardiac MRI is superior to echocardiography in identifying areas of segmental hypertrophy not reliably visualized or underestimated by echocardiography. Visualized features may include:

- left ventricular systolic dysfunction
- left ventricular hypertrophy
  - with or without right ventricular hypertrophy
  - predilection (preferably) for the basal interventricular septum
- systolic anterior motion (SAM) of the mitral valve
  - mitral regurgitation
- left ventricular apical aneurysms
- morphologic variations involving the mitral valvular apparatus (e.g. papillary muscles)

Cardiac MRI also has a role in asymptomatic HCM mutation carriers by identifying phenotypic markers of HCM in the absence of left ventricular hypertrophy including:

- myocardial crypts

- elongated mitral valve leaflets
- late gadolinium enhancement (LGE)
    - patchy/streaky intramyocardial patterns at the right ventricular insertion sites within the hypertrophied myocardium suggest fibrosis

Lab Results

- **Genetic testing**
    - Cardiomegaly-implicated gene mutations

Differential diagnoses

It is important to differentiate hypertrophic cardiomyopathy from other causes of left ventricular hypertrophy including hypertension, aortic stenosis, athletic heart, and cardiac amyloidosis.

A **detailed history** is essential for making this distinction as well as a thorough review of past medical and family history. Imaging, typically echocardiography, is also important for the exclusion of other differential diagnoses.

Other **less common** differential diagnoses include:

- Metabolic disorders (e.g. Anderson-Fabry disease, Pompe disease)
- Primary mitochondrial disease
- Neuromuscular disease (e.g. Friedreich's ataxia)
- Malformation syndromes (e.g. Noonan syndrome, LEOPARD syndrome)

Factors that Dec. LVOT Obstruction (Anything that Inc. LV preload)	Factors that Inc. LVOT Obstruction (Anything that Dec. LV preload
ECF volume expansion (e.g., saline infusion)	ECF Volume depletion
Squatting	Standing
Hand grip exercise	Valsalva
Bradycardia	Tachycardia
Beta-blockers, CCBs (Non-Dihydropyridine), Disopyramide	Beta-agonists, Digoxin, Dopamine
Vasoconstrictors (e.g., alpha 1 agonists), phenylephrine	Vasodilators (e.g., Nitrates), Dihydropyridine CCBs

	Most Murmurs	Murmurs in HOCM
	More Blood → More Murmur Less Blood → Less Murmur	More Blood → Less Murmur Less Blood → More Murmur
Squatting	↑Blood → ↑murmur	↑Blood → ↓murmur
Hand Grip	↑Blood → ↑murmur	↑Blood → ↓murmur
Leg Raising	↑Blood → ↑murmur	↑Blood → ↓murmur
Valsalva	↓VR → ↓murmur	↓VR → ↑murmur
Standing	↓VR → ↓murmur	↓VR → ↑murmur

Treatment/Management

- **Medication**
  - **Beta-blockers**
    - Dec. heart rate, contractile force
  - **CCBs (non-dihydropyridine)**
    - If Beta blockers not tolerated
    - Slows down heart rate
  - **Disopyramide**
    - Can be used for its negative inotropic properties
  - **Avoid Volume Depletion**

- **Surgery**
  - **Implantable cardioverter-defibrillator (ICD)**
  - **Surgical septal myectomy**
    - Involves removing portion of IVS, decrease obstruction
  - **Septal ablation**
    - Chemical myomectomy to partially ablate septum
  - **Heart Transplant**
    - If unresponsive to all other forms of treatment

- **Life style changes**
  - Avoid Alcohol
  - Avoid strenuous exercise

Restrictive Cardiomyopathy

In the RCM, the muscles of heart's lower chamber stiffens & can't fill with blood. This can cause heart failure, which increase pressure on heart & may cause fluid buildup in lungs

Causes

- **Primary (idiopathic)**
  - Mutations in gene encoding for cardiac troponin I
- **Secondary (to another disease)**
  - **Sarcoidosis**
    - Immune cell collections form granulomas in heart tissue
  - **Amyloidosis**
    - Amyloids are misfolded proteins → insoluble → deposit in tissue, organs → organs less compliant
    - Familial amyloid cardiomyopathy
    - Mutant transthyretin (TTR) protein; misfolded deposits preferentially in heart tissue
    - Senile cardiac amyloidosis; TTR protein/wild type TTR deposits in heart over time.
  - **Hemochromatosis**
    - Iron deposits in heart tissue, contributes to restricted tissue
  - **Loefler syndrome**
    - Eosinophils accumulate in lung tissue
    - Loeffler endocarditis/Loeffler endomyocarditis: eosinophils also accumulate in endocardial layer of heart tissue → inflammation, endocardial fibrosis → restrict heart tissue
  - **Cancer (primary or metastatic)**
  - **Fibrosis (endocardial fibroelastosis)**
    - Fibrosis develops in endocardium (inner lining of heart) & subendocardium (layer underneath endocardium)
    - Post cardiac surgery, or transplant
  - **Heart tissue Radiation**
    Radiation generates reactive oxygen species → inflammation over time → myocardial fibrosis → tissue stiff, restrict
  - Systemic sclerosis
  - Systemic lupus

- Doxorubicin, busulfan, ergotamine, methysergides
- Diabetes mellites
- Pompe's & Fabry's
- Infiltrative disease, storage disease, endomyocardial disease

Signs & Symptoms

Congestion & low output symptoms

- **Right Sided Heart Failure**
  - Raised jugular venous pressure
  - Hepatomegaly
  - Lower limb oedema
  - Ascites
- **Left Sided Heart Failure**
  - Pulmonary oedema
  - Cough
  - Haemoptysis
  - Dyspnea
  - Paroxysmal Nocturnal Dyspnea
  - Orthopnea
- Low cardiac output symptoms
  - Syncope
  - Dizziness
  - Fatigue
  - Angina: if coronary arteries involved

Right sided heart failure is more prominent then left sided

Diagnosis

Physical examination

- $^{+}/_{-}$ Kussmaul's sign is occasionally present
- Paradoxical pulses are absent
- Apical pulses are prominent

Lab Findings

- Raised BNP level

Auscultation

- Stiff ventricle → S4 Heart sound

- Regurgitant Av valves murmur is common
- Crackles in chest

ECG

- Low voltage QRS complexes
- Non-specific ST segment/ T wave changes
- BBB (bundle branch block)
- AV block (3rd degree AV block may occur in sarcoidosis)
- Pathological "pseudo-infarction" Q waves.
- Atrial & ventricular dysrhythmias.

- **Pathophysiology:** RCM is the least common form of cardiomyopathy. It occurs in the advanced stages of myocardial infiltrative disease – e.g., due to haemochromatosis, amyloidosis or sarcoidosis

    - ~ *Diffuse myocardial infiltration leads to low voltage QRS complexes.*
    - ~ *AF may occur due to atrial enlargement; ventricular arrhythmias are also common.*
    - ~ *Infiltration of cardiac conduction system (e.g., due to septal granuloma formation in sarcoidosis) may lead to conduction disturbances -e.g., BBB & AV Block*
    - ~ *Healing granuloma in sarcoidosis may produce" pseudo-infarction" Q waves.*

ECHO

- Restrictive diastolic filling, bi-atrial enlargement, normal systolic function
- Restrictive mitral filling because of noncompliant LV
    - E >100cm/sec
    - E/A >2
    - Deceleration Time (DT) ≤160msec
    - Septal e' <7 cm/sec, lateral e' <10 cm/sec
    - Average E/e' ratio >14
- Markedly **reduced tissue doppler velocities**, & **hepatic doppler flow reversals** are most marked during inspiration
- Myocardium may have an abnormal **echogenic, sparkling appearance** suggestive of infiltrative disease

- **“Binary” appearance of LV myocardium** in Fabry’s disease with bright endocardium & myocardium with “clearing” of intervening sub endocardium related to compartmentalization & accumulation of glycosphingolipids in certain layers of the wall.
- Cardiac involvement in hyper eosinophilic syndrome involves eosinophilic Infiltration of the myocardium causing necrosis & obliteration of the ventricular apex with thrombus. Endomyocardial fibrosis eventually leads to RCM.

Chest-X ray

- Enlarged atria
- Pulmonary oedema at times

CT/MRI

- LA enlargement
- LV hypertrophy
- Thickened atrial septum

Treatment/Management

- **Medications**
  - **Loop Diuretics**
    - Dec. systemic, pulmonary congestion
  - **ACEi**
  - **Anticoagulants**
- **Surgery**
  - Left Ventricular assist device
  - Heart Transplant
- **Lifestyle Changes**
  - Salt restriction