CARDIOMYOPATHIES

1. DILATED CARDIOMYOPATHY

DCM is characterized by dilatation and systolic impairment of the LV, usually accompanied by dilatation of the RV and the atria.

Diagnostic findings

- LV dilatation—assessed by M-mode/2D/3D
- LVEDV > 112 mL/m2 corrected for age and body surface area
- EF < 45% or fractional shortening < 25%
- LV wall thinning is a common finding in DCM
- IHD, chronic pressure overload and volume overload states must be excluded before a diagnosis of DCM can be made
- A reduced LVEF and a LVIDd > 112% corrected for age and BSA is a diagnostic criterion for idiopathic DCM after exclusion of secondary causes.

Associated findings

- LV spherical remodelling: typical for DCM. long-axis diameter preserved and transverse diameter increased
- LV systolic dyssynchrony
- LV thrombus
- Dilated atria
- Dilated RV (not essential for diagnosis)
- Mitral regurgitations often due to dilated mitral annulus
- Pulmonary hypertension
- Diastolic dysfunction

Prognostic role of echocardiography

- Severity of systolic dysfunction
- LV filling pattern (LVDD)
- Coexistence of RV dysfunction
- Severity of LV dilatation
- Sphericity index: ratio of long to minor axis decreases (< 1.5 implies pathologic remodeling)
- Systolic dyssynchrony
- PA pressure
- LA volume

DCM can result from a number of conditions, including:

- Myocarditis (viral)
- Prolonged tachycardia (tachycardia-induced cardiomyopathy)
- Alcohol
- Drugs (e.g. anthracyclines)
- Peripartum cardiomyopathy.
- Familial DCM is also recognized, defined by the presence of DCM in two or more individuals in the same family. Asymptomatic first degree relatives of patients with idiopathic DCM should have an echocardiogram every 3-5 years or when they develop signs or symptoms
- DCM is also seen in X-linked diseases such as Becker and Duchenne muscular dystrophies.
- DCM without an identifiable cause is called idiopathic DCM.
- Apical ballooning or stress cardiomyopathy, also known as Takotsubo cardiomyopathy
- LV dilatation and impairment secondary to ischaemic, valvular or hypertensive heart disease is not usually classified as DCM; however, end stage ischemic LV dysfunction causes wall thinning and global reduction in systolic function similar to DCM. The same is true of end-stage RCM.

Echocardiographic role in CRT

- LVEF (2D Simpson's biplane, 3D)
- Dyssynchrony study—correct technique and further evidence needed
- Visual (apical rocking)
- Quantitative
  - interventricular dyssynchrony
  - atrioventricular dyssynchrony
  - intraventricular dyssynchrony
- Stress echo for viability and dyssynchrony assessment
- CRT optimization

ICD- is indicated in symptomatic patients (NYHA class II-III) with an ejection fraction ≤35%

2. HYPERTROPHIC CARDIOMYOPATHY

HCM is an autosomal dominant condition affecting 1 in 500 of the population and is a common cause of sudden cardiac death, particularly in the young. The generally accepted definition of hypertrophic cardiomyopathy (HCM) is LV hypertrophy associated with nondilated ventricular chambers in the absence of another cardiac or systemic disease that itself could produce such hypertrophy. The LV hypertrophy in HCM is usually asymmetrical (in contrast to the concentric LV hypertrophy seen in hypertension or aortic stenosis). Another common pattern is apical hypertrophy, which gives the LV cavity a characteristic 'ace of spades' appearance.

If the hypertrophy is located in the LV outflow tract (LVOT) it may obstruct the flow of blood out of the LV into the aorta – this is hypertrophic obstructive cardiomyopathy (HOCM). LVOT obstruction in HOCM is dynamic and varies with preload and afterload and, therefore, is dependant on many factors such as hydration, heart rate, squatting, etc.

In an echo study for HCM look for the following features: (*ASH, LVDD, Dagger, SAM, MR, early closure of AV*)

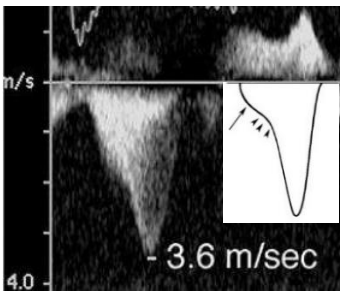
1) **LV hypertrophy**, which is usually asymmetrical (**ASH**), but is sometimes concentric. **LVH**: wall thickness (from one to all LV segments) ≥ 15 mm (or > 2SD for age, gender, and height), lower in first relatives of patients. **ASH**: IVS/PWT > 1.3 in normotensive patients (or > 1.5 in hypertensive patients)
2) **LV diastolic dysfunction** is common

   Global LV diastolic dysfunction (PW Doppler LV inflow profiles):
   - Classic patterns have poor correlation with LV filling pressures
   - Bizarre diastolic patterns (positive isovolumic relaxation flow, triphasic trans-mitral flow) → no prognostic impact

   LV subendocardial diastolic dysfunction
   - Doppler myocardial imaging (TDI): low e' velocities (e' < 7 cm/s; e'/a' < 1) in hypertrophic and non-hypertrophic segments, high heterogeneity of velocities
   - 2D speckle tracking: delayed LV untwist, occupying > 25% of diastole (normal: occurs in the first 25% of diastole)

   Increased LV filling pressures in HCM
   - E/e' lateral ≥ 10-12 (average E/e' ratio >14)
   - (Pulmonary vein a wave duration – mitral A wave duration) ≥ 30-35 ms
   - LA indexed volume≥ 34 ml/m2
   - PASP > 35 mmHg (TR velocity >2.8 m/s)

   N.B: LV systolic function is generally preserved until a late stage. LV EF is usually supernormal, but indexed stroke volume is often reduced due to the small LV cavity
3) **Evidence of obstruction (subaortic, midventricular)**: The dynamic LVOT obstruction profile on CW Doppler shows a **late**-peaking curve (dagger profile), usually with upward concavity in early systole, compared with the symmetric flow profile in valvular aortic stenosis and mitral regurgitation.



4) **Partial early/mid-systolic closure (notching) of AV** (non specific). The ejection dynamics of HOCM allow for relatively normal early left ventricular ejection. Obstruction occurs in mid- to late-systole at which point ejection transiently diminishes. The reduction in flow volume out the LVOT results in partial closure of the aortic valve, often with a secondary opening as final ejection occurs. This results in a single notch or occasionally several discrete high-amplitude notches of aortic valve motion.

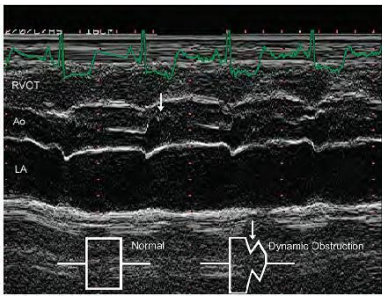


Fig. partial mid- systolic closure (notching) of the AV (arrow).

5) **Systolic anterior motion (SAM) of the anterior MV leaflet**: blood flowing through narrowed LVOT → a Venturi (suction) effect → drags the MV towards the septum during systole (recently SAM is thought to be due to abnormal geometric relationship of papillary muscles and the mitral supporting apparatus combined with hyperdynamic LV contraction)

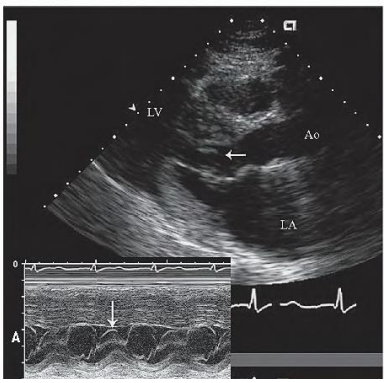


Fig. note the motion of the mitral valve into the LVOT (arrows). The M-mode echocardiogram (small inset) also demonstrates SAM of the MV (arrow).

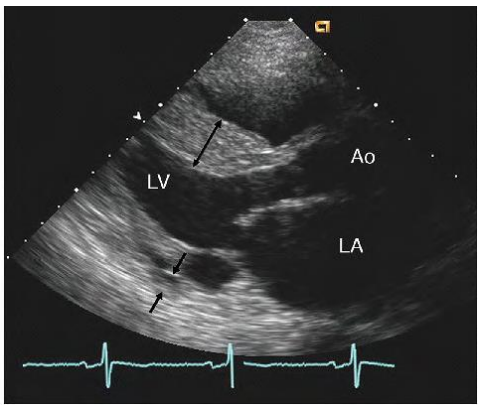
6) **Functional MR- due to SAM** that drags" the AML towards the septum (causing the leaflet tip to make contact with the septum) during systole. This opens the mitral valve, leading to an eccentric (**posteriorly directed**) jet of MR.

Treatment of HCM:

- Medical treatment
- Alcohol septal ablation: this essentially causes an infarction of the ventricular septum causing RWMA
- Surgical myectomy
- DDD pacing

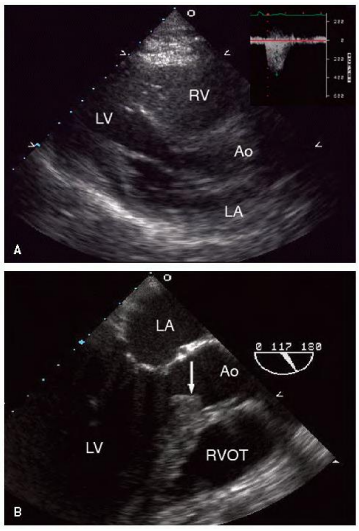
Conditions Which May Mimic Hypertrophic Cardiomyopathy- Any situation which results in relatively greater septal than posterior wall thickness potentially could be confused for isolated pathologic septal hypertrophy. The generally accepted definition of HCM is LVH associated with nondilated ventricular chambers in the absence of another cardiac or systemic disease that itself could produce such hypertrophy.

- Hypertensive heart disease (with inferior MI): Occasionally, one encounters a patient with LVH related to hypertension and concurrent inferior MI. The subsequent reduction in wall thickness of the posterior wall related to coronary disease, in conjunction with the hypertension-related hypertrophy of the remaining walls, creates a pattern which mimics classic HCM. By noting the akinesis and pathologic scarring of the posterior wall, as well as the clinical scenario, this situation should not be confused for a true HCM.



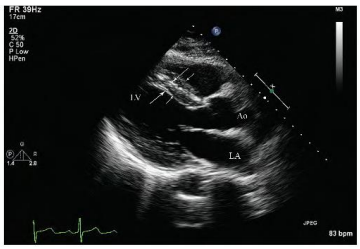
Patient with LVH related to hypertension in a previous inferior MI. Note the ASH with a septal to posterior wall ratio exceeding 1.3:1. In this instance, the finding is related to pathologic thinning of the posterior wall combined with hypertension-related hypertrophy of the septum and does not represent a true HCM.

- Subvalvular membrane: sometimes a discrete subvalvular membrane is difficult to visualize and the septal hypertrophy may progress to the edge of the membrane and further obscure it. Rarely, the septal hypertrophy may contribute a dynamic component to the obstruction. A valuable clue to the presence of a fixed subvalvular membrane is the presence of concurrent aortic insufficiency which is rare in hypertrophic cardiomyopathy but very common in patients with fixed outflow tract obstruction due to a discrete membrane. TOE is usually diagnostic.



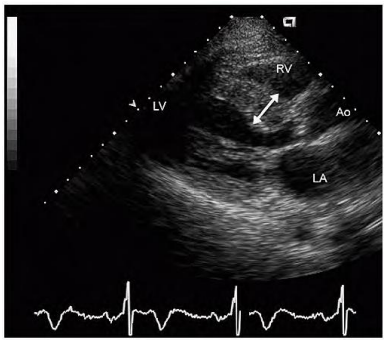
TTE parasternal (upper panel) and longitudinal view TOE (lower panel) in a patient with a fixed subvalvular obstruction mimicking HCM. ***A:*** *Note the ventricular hypertrophy with a greater degree of septal than posterior wall suggesting the presence of HCM. In the small inset, note the CW Doppler image recorded through the LVOT with a peak velocity of 4 m/sec, suggesting an outflow tract gradient of 64 mm Hg.* ***B:*** *Note the discrete fibromuscular ridge protruding into LVOT (arrow) which has resulted in a pattern mimicking typical HOCM.*

- Prominent muscle bundle or trabeculation, lying along the RV side of the anterior IVS. With either M-mode echocardiography or isolated parasternal long-axis imaging the overlying trabeculation may be confused with an intrinsic portion of the ventricular septum. Similarly, any entity resulting in right ventricular hypertrophy will also result in septal hypertrophy. It is quite common to see disproportionate septal hypertrophy meeting the classic criteria of the septal to posterior wall thickness of 1.3:1 in patients with pulmonary hypertension.



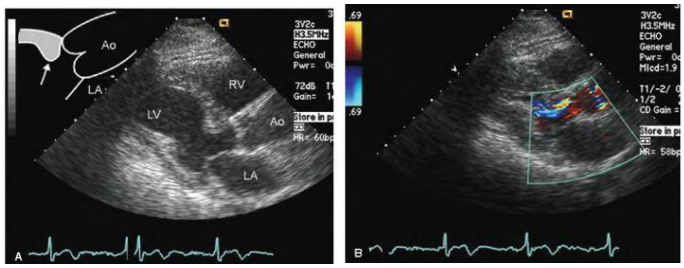
Normal patient in whom a prominent right-sided trabeculation has resulted in the appearance of septal hypertrophy, mimicking a HCM.

- Acquired hypertrophic cardiomyopathy of the hypertensive elderly. This is a variation of hypertensive cardiovascular disease in which there has been a relatively greater degree of hypertrophy of the ventricular septum, which when combined with the normal increase in septal angulation seen in the elderly results in a variable degree of outflow tract obstruction. SAM of the MV can result in secondary MR. The diagnosis is established clinically when one encounters the anatomic appearance of an obstructive hypertrophic cardiomyopathy in an elderly patient with longstanding hypertension, but no family history or other features consistent with true hypertrophic cardiomyopathy.



Elderly hypertensive patient with "hypertensive HCM of the elderly." The combination of septal angulation and disproportionate proximal septal hypertrophy results in an anatomic pattern, mimicking classic HCM. SAM of the MV and varying degrees of outflow tract obstruction may also be encountered.

- Discrete upper septal thickening (DUST), also known as "sigmoid septum", "septal bulge" or "septal knuckle"- is a common anatomical variant in elderly, characterized by angulation between the ascending aorta and LVOT often in conjunction with basal septal hypertrophy. The hypertrophy may be quite focal and result in a localized area of turbulence in the outflow tract that may be the source of the ejection murmur often heard in elderly patients.



87-year-old patient with a systolic murmur. A: Note the angulated septum with proximal septal hypertrophy (arrow in the schematic) and the mild thickening of the aortic valve. B: The image was recorded with colour flow Doppler imaging and reveals the marked acceleration of flow around the sigmoid septum, which is the cause of a systolic murmur in this patient.

- On occasion, cardiac amyloid may also be confused for hypertrophic cardiomyopathy when the distribution of amyloid infiltration is not uniform. TDI may detect markedly reduced annular velocities which, while not specific, may point in the direction of an infiltrative rather than hypertrophic cardiomyopathy.

- Highly trained athletes may develop a pattern of ventricular hypertrophy, which may include chamber dilation as well as increased wall thickness. The “athlete's heart” can be confused for HCM. The increase in wall thickness in athlete's heart is usually ≤13 mm. In the athlete's heart there will be no evidence of outflow tract obstruction. Doppler tissue profiles will reveal higher systolic and diastolic annular and wall velocities in the athlete's heart than in HCM.

- In patients with intravascular volume depletion, especially if concurrently on inotropic agents. Often, there is a history of hypertension, and the relatively low intravascular volumes with augmented contractility result in hyperdynamic motion of the ventricle with an acquired dynamic outflow tract obstruction. The acquired dynamic outflow tract obstruction and SAM of the MV can occasionally result in significant degrees of MR and detection of clinically significant murmurs. Detection of a small hyperdynamic ventricle with outflow tract obstruction in this setting is an indication for volume resuscitation and discontinuation or decrease in inotropic support

- An additional entity which can mimic obstructive cardiomyopathy is a patient with ischemia in the LAD coronary artery distribution. This can occur either as a consequence of an acute coronary syndrome or be provoked at the time of DSE. The distal ischemia results in an exaggerated angulation of the anterior septum which, when combined with hyperdynamic contractility at the base of the heart, may result in dynamic outflow tract obstruction with SAM of the MV and, on occasion, mitral regurgitation. A similar phenomenon has, on occasion, been noted in the apical ballooning syndrome (Tako-Tsubo)

- Spontaneously closed perimembranous VSD

- Fabry’s disease (an x-linked lysosomal storage disorder) causes ventricular hypertrophy and septal thickening similar to HCM

- Aortic stenosis

- Mitochondrial disease

- Friedreich’s ataxia

- Danon disease

- Noonan syndrome

- Pompe disease

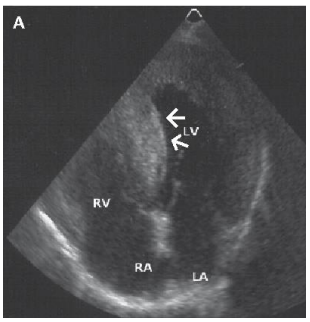
3. RESTRICTIVE CARDIOMYOPATHY

The underlying abnormality in restrictive cardiomyopathy is primary thickening & "stiffening" of the left ventricular myocardium and subsequent congestive heart failure due purely to diastolic dysfunction. In many of the restrictive cardiomyopathies, however, especially later in their course, a component of systolic dysfunction may be present. Restrictive cardiomyopathy most commonly results from:

- **Myocardial infiltration**, as seen in amyloidosis, haemochromatosis or glycogen storage disease (Fabry's disease)
- **Endomyocardial fibrosis**
- **Sarcoidosis**

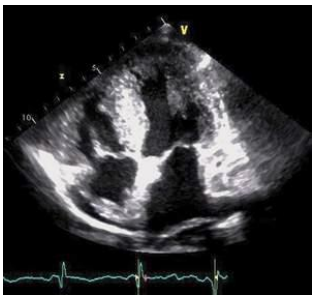
Echo features of restrictive cardiomyopathy include:

- LVH (usually concentric)
- Restrictive cardiomyopathy may also involve the RV.
- In the pure, isolated restrictive cardiomyopathy, the internal dimensions of the LV and RV are normal and there is secondary dilation of both atria (due to ↑ventricular filling pressure). This secondary atrial dilation is commonly associated with AF and stasis of blood. Secondary pulmonary hypertension is common.
- Systolic and/or diastolic dysfunction
- Valvular regurgitation (most commonly mitral)
- **Sarcoidosis** can cause RWMA and conduction disturbances
- **Fabry's disease** → binary appearance of the endocardial border due to compartmentalisation of glycosphingolipids. Fabry's disease (also known as Anderson–Fabry's disease) is an X-linked lysosomal storage disorder in which glycosphingolipids accumulate in and damage various organs, including the heart. The diagnosis of Fabry's disease should be considered in patients with unexplained LV hypertrophy. Because enzyme replacement therapy is effective in Fabry's disease, it is very important to differentiate this condition from HCM by measuring the blood level of alpha-galactosidase A. Systolic function is generally preserved in Fabry's disease.



Cardiac Fabry's disease: concentric LVH and the binary appearance of the left ventricular endocardial border

- **Amyloid infiltration** → echo-reflective myocardium (bright) (speckling) (sparkling). This abnormal myocardial texture was initially described using early generation scanners. It should be emphasized that when scanning with modern scanners in a tissue harmonic mode, myocardial intensity is enhanced and that appearance of a bright myocardial signature is not specific for amyloid infiltration. In addition to cardiac amyloid, hypertrophic cardiomyopathy and hypertrophy seen in end-stage renal disease often have a similar appearance. Apical sparing, pericardial effusion and IAS hypertrophy are frequent in cardiac amyloidosis.



Amyloid heart disease: sparkling myocardial LVH + pericardial effusion + interatrial septum hypertrophy

- **Endomyocardial fibrosis** → The endocardium is echo-reflective
- **Hypereosinophilia** due to eosinophilic leukaemia, tropical hypereosinophilia, or idiopathic eosinophilia results in characteristic abnormalities detected with echocardiography. The most classic abnormality is obliteration of the left or right ventricular apex by laminar thrombus. Pathologically, the thrombus is composed of inflammatory tissue, thrombus, and eosinophilic infiltrates. It results in a reduction of ventricular chamber size and increasing stiffness, resulting in a restrictive cardiomyopathic picture. Additionally, hypereosinophilic syndrome has a propensity to involve the posterior left ventricular wall and posterior mitral valve leaflet and result in MR. DCM has also been reported with hypereosinophilic syndrome

Separation of constrictive pericarditis from restrictive cardiomyopathy

	Constriction	Restriction
Atrial size	Normal	Dilated
Pericardial appearance	Thick/bright/calcified	Normal
Septal motion	Abnormal (2nd reverberation)	Normal
Septal position	Varies with respiration (inspiratory bounce)	Normal
Mitral E/A	Increased (≥2.0)	Increased (≥2.0)
Deceleration time	Short (≤160 ms)	Short (≤160 ms)
Annular e′ (LV diastolic function)	Normal	Reduced (≤10 cm/sec)
Pulmonary hypertension	Rare	Frequent
LV size/function	Normal	Normal
LV dimensions	Normal	LVH
Mitral/tricuspid regurgitation	Infrequent	Frequent (TR > MR)
Isovolumic relaxation time	Varies with respiration	Stable with respiration
Respiratory variation of mitral E velocity	Exaggerated (≥25%)	Normal
Hepatic vein flow	Expiratory **diastolic** reversal	Inspiratory **diastolic** reversal
Colour M-mode mitral valve Vp	Increased (≥55 cm/sec)	Reduced

4. HYPERTENSIVE HEART DISEASE

Poorly controlled hypertension results in hypertensive cardiovascular disease and, when long-standing, the appearance of a dilated cardiomyopathy. In this instance, LVH typically persists in the presence of chamber dilation and global dysfunction. This combination of hypertrophy with moderate degrees of dilation and global dysfunction is fairly typical of hypertensive cardiovascular disease with subsequent LV dysfunction but also could be mimicked by a variety of infiltrative diseases.

Although LVH associated with hypertension is typically concentric, but it can be associated with LVOT obstruction and significant mid-cavity gradients similar to that seen in idiopathic hypertrophic cardiomyopathy

5. ARRHYTHMOGENIC RV CARDIOMYOPATHY

ARVC is one of the most common and under-diagnosed causes of cardiac sudden death in a young person. ARVC causes fatty infiltration of the RV (and sometimes the LV) resulting in hypokinesis and thinning of the RV free wall, often with localised aneurysms.

Major echocardiographic criteria for ARVC:

Regional RV Dyskinesia or Aneurysm

And one of the following

PLAX RVOT ≥ 32mm (corrected for body size [PLAX/BSA] ≥ 19mm/m2)

PSAX RVOT ≥ 36mm (corrected for body size [PLAX/BSA] ≥ 21mm/m2)

Or

Fractional Area Change ≤ 33%

Minor echocardiographic criteria for ARVC

Regional RV Akinesia or Dyskinesia

And one of the following

PLAX RVOT ≥ 29 to < 32mm (corrected for body size [PLAX/BSA] ≥ 16 to < 19mm/m2)

PSAX RVOT ≥ 32 to < 36mm (corrected for body size [PLAX/BSA] ≥ 18 to 21mm/m2)

Or

Fractional Area Change > 33 to < 40%

Hints:

ARVC is a rare cause of RWMA; ischaemic heart disease, PEs and bundle branch block are more common causes.

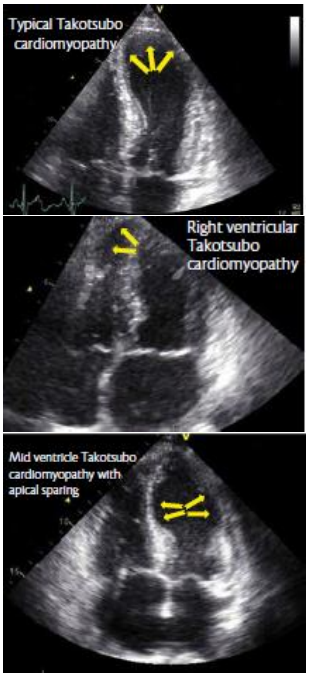
Basal RV:LV ratio at end diastole may be used to demonstrate RV dilatation; however, there are no specific values for diagnosis of ARVC. RV:LV ratio > 0.66 is abnormal

6. TAKOTSUBO CARDIOMYOPATHY

Takotsubo cardiomyopathy (Broken heart syndrome or stress cardiomyopathy)- is a rare form of cardiomyopathy, first described in Japan, named after the characteristic shape of the LV seen in this condition. This cardiomyopathy is most commonly seen in post-menopausal women and is triggered by extreme emotional or clinical stress, which causes a ballooning of the LV apex. Patients present with chest pain and/or heart failure with ECG changes suggestive of an anterior myocardial infarction (but in the absence of coronary disease). In most cases LV function recovers within 2 months of the presenting episode.

Diagnostic findings:

- **Hypokinesia/akinesia** which does not follow a coronary territory of distribution
- **No scar** in the myocardium
- **No coronary artery lesions nor plaque rupture**
- Typical Takotsubo: **hypokinetic apex with hyperkinetic basal segments** (apical ballooning, light bulb-like LV)
- Or **apical RV**
- Or **Variant form—sparing apex**
- **Typical complete recovery** in a few weeks



7. LEFT VENTRICULAR NON-COMPACTION (LVNC)

LVNC- previously "spongy heart syndrome" is a rare congenital form of cardiomyopathy in which the apical portion of the LV (and sometimes RV) is involved due to failure of embryogenic involution of LV trabeculae. Involved myocardium is characterized by a spongy appearance with prominent trabeculations and deep intertrabecular recesses. The ratio between the thickness of the non-compacted (N) and compacted (C) layers should be measured at end-systole, and is characteristically > 2. Colour flow imaging will demonstrate flow within these spongiform recesses (manifest as ridging of the endocardium), creating a "Swiss cheese-like" appearance. LV function can be preserved or severely decrease. Isolated ventricular non-compaction (IVNC) can cause systolic and/or diastolic dysfunction, and can predispose the patient to thromboembolism and arrhythmias. Some (but not all) cases are familial.

OTHER SYSTEMIC DISEASES ASSOCIATED WITH CARDIOVASCULAR DYSFUNCTION

- **Diabetes mellitus-** associated with heart failure with normal ejection fraction and premature CAD.
- **Renal failure-** associated with cardiac failure (due to accumulation of metabolic by-products, including metalloproteinases, or more commonly due to associated hypertension, DM and premature CAD. Also associated with pericarditis, pericardial effusion, mitral annular calcification and LVH due to hypertension and an abnormal texture of the hypertrophied myocardium that mimics that seen in cardiac amyloid
- **Thyrotoxicosis-** associated with high-output cardiac failure
- **Psoriasis-** associated with high-output cardiac failure
- **SLE-** associated with LV dysfunction and RWMA due to coronary vasculitis. More commonly associated with non-infective endocarditis (Libman-Sacks vegetation on the atrial side of the leaflet)
- **Hypereosinophilic syndrome**- is a rare cause of LV thrombus in the absence of regional wall motion defects.
- **Chagas' disease**- can cause apical LV aneurysm (that may contain thrombus) and is exceptional in that it has almost no involvement of the ventricular septum.