Cardiomyopathies

Cardiomyopathies are disorders of heart muscle. There are 4 kinds:

1. **Dilated (or congestive) cardiomyopathy** Commonest. A dilated, flabby heart.
   Assocs: ischaemia, EtOH, drugs (phenytoin, heavy metals, cocaine, amphetamines), haemochromatosis, viral infection (e.g. HIV, CMV), autoimmune, peripartum, thyrotoxicosis, tachycardia-induced, cong.-Barth Syndrome (X-linked).
   Prevalence: 0.2%. Up to 50% familial. M>F. AfroAm>Causcasians.
   Presentation: Fatigue, dyspnoea, pulmonary oedema, RVF, emboli, AF, VT.
   Signs: ↑Pulse, ↓BP or ↑BP , ↑JVP , displaced, diffuse apex, S3 gallop, MR /TR (mitral or tricuspid regurgitation), pleural effusion, oedema, jaundice, hepatomegaly, ascites.
   Investigations:
   - CXR: cardiomegaly, pulmonary oedema.
   - ECG: tachycardia, nonspecific T wave changes, poor R wave progression, LVH, LAE.
   - Echo: globally dilated hypokinetic heart + low ej frac. MR, TR , LV mural thrombus.

   Management: As for heart failure. Bed rest, diuretics, digoxin, ACE inhibitor, anticoagulation. Consider pacemaker, cardiac transplantation.
   Mortality: Variable, e.g. 40-80% in 5yrs.

2. **Hypertrophic cardiomyopathy** Hypertrophic obstructive cardiomyopathy (HOCM) LV outflow tract (LVOT) obstruction from asymmetric septal hypertrophy.
   Prevalence: 0.2%. Auto dom., but 50% are sporadic. 70% have mutations in genes for ß-myosin, α-tropomyosin and troponin T. Any age. Ask about FamHx or sudden death.
   The patient: Angina; dyspnoea; palpitation; syncope; sudden death (VF is amenable to implantable defibrillators). Jerky pulse; a wave in JVP ; double apex beat; systolic thrill at lower left sternal edge; harsh ejection systolic murmur.
   Investigations:
   - ECG : LVH; LAE; deep, narrow 'septal' Q waves in ant, lat & inf leads with upright T's; AF; WPW syndrome; ventricular ectopics; progressive T wave inversion; VT.
   - Echo: asymmetrical septal hypertrophy; small LV cavity with hypercontractile posterior wall; mid-systolic closure of AV; systolic anterior movement of MV.
   - Cardiac catheterization may provoke VT. It helps assess: severity of gradient; coronary artery disease or mitral regurgitation.
   - MRI, Biopsy, Electrophysiological studies (WPW syndrome) may be needed.

   Management: ß-blockers or verapamil for symptoms. Amiodarone 100-200mg/day for arrhythmias (AF, VT). Anticoagulate for paroxysmal AF or systemic emboli.
   HOCM in childhood = poor prognosis, but improved by high dose ß-blocker therapy. Dual chamber pacing when symptoms resistant to Rx. Septal myomectomy (surgical, or chemical, with EtOH, to ↓LVOT gradient) reserved for severe symptoms.
   Mortality: 5.9%/yr if <14yrs; 2.5%/yr if >14yrs. Poor prognostic factors: age <14yrs or syncope at presentation; family history of HOCM /sudden death. Genetic testing for some types of HOCM is available.

3. **Restrictive cardiomyopathy** Rare. Not usually familial.
   Prevalence: 0.02-0.1%.
   Causes: Amyloidosis; haemochromatosis; sarcoidosis; scleroderma; Löffler's eosinophilic endocarditis, endomyocardial fibrosis.glycogen storage diseases, idiopathic
   Presentation is like constrictive pericarditis. Features of RVF predominate: ↑JVP, with prominent x and y descents; loud S3, pulm oedema, hepatomegaly; oedema; ascites.
   Diagnosis: Cardiac catheterization. Biopsy in ddition to usual ECG, CXR, bloods.
   Management: Treat heart failure. Anticoagulants, amiodarone, +/- pacemaker/ICD, consider transplantation.
   Prognosis: Variable. Particularly poor when assoc with amyloidosis.

4. **Arrhythmogenic Right Ventricular Cardiomyopathy** Formerly called arrhythmogenic right ventricular dysplasia. Characterised by progressive fibro-fatty replacement of right ventricular myocardium with progressive effects on the right ventricle, a strong familial transmission, and presentation with symptomatic arhythmias or sudden death.
   Epidemiology: 30% familial. M>F. Young adults.
   Causes: unknown
   Presentation:
   - *Concealed phase* – may have minor ventricular arrhythmias. Sudden death in sport
   - *Overt electrical disorder* - Symptomatic RV arrhythmias + functional and structural abnormalities. Usually presents with palpitations or syncope. Arrhythmias and sudden death are common.
   - *Right ventricular failure* - extension of disease to whole RV causes dysfunction.
   - *Biventricular pump failure* - end stage. LV involvement leads to heart failure and may mimic dilated CM.

   Diagnosis: Cardiac catheterization. Biopsy in addition to usual ECG, CXR, bloods.
   Management: Treat heart failure. Anticoagulants, amiodarone, +/- pacemaker/ICD, consider transplantation.
   Prognosis: Variable. Particularly poor when assoc with amyloidosis.

Tako-tsubo Syndrome or CM *aka transient apical ballooning, stress-induced cardiomyopathy, broken heart syndrome.* A rare non-ischaemic CM with sudden weakening of the myocardium.
Assocs: post menopausal F, emotional or clinical stress in >60%. Pathogenesis not clear- ?inflammatory, ?regional myocarditis, ?catecholamine-induced microvascular spasm.
Inv: Echo - LV has norm/hyperkinetic base & dilated/hypokinetic mid/apex → sim to a Japanese octopus trap (tako-tsubo). ECG may show changes of ant. AMI (↑ST V1-V3, ↓T V2-V6) with modest ↑Trop but coronary arteries appear normal on angio.
Mx: supportive with recovery of LV fn in >90% initial survivors after couple of months.

Specific heart muscle diseases (cause known) mostly sim to dilated CM. Amyloid and carcinoid may be restrictive; amyloid and and cardiac involvement in Friedreich's ataxia mimic HOCM.
The chief causes: Ischaemic heart disease and ↑BP (may present in failure with normal BP: examine fundi to reveal signs of earlier hypertension).
Other causes: Infection, EtOH, post-partum, smoking, connective tissue diseases, DM, hyper- and hypothyroidism, acromegaly, Addison's, phaeochromocytoma, haemochromatosis, sarcoid, Duchenne muscular dystrophy, myotonic dystrophy, irradiation, cytotoxics, storage diseases.