Cardiomyopathies

Cardiomyopathies are a heterogeneous group of diseases affecting the heart muscle, leading to mechanical or electrical dysfunction and often progressing to heart failure (HF), arrhythmias, or sudden cardiac death (SCD). This pamphlet provides an overview of the major types of cardiomyopathies, their clinical presentation, causes, diagnostic workup, evaluation, consultants, complications, and treatment, with clinical scenarios for practical understanding.

Clinical Presentation

Dilated Cardiomyopathy (DCM):

- **Symptoms:** Heart failure (dyspnea, fatigue, edema), palpitations (arrhythmias), chest pain (ischemia-like).
- **Exam:** S3 gallop, JVD, crackles, peripheral edema, cardiomegaly, murmur (mitral/tricuspid regurgitation).

Hypertrophic Cardiomyopathy (HCM):

- **Symptoms:** Exertional dyspnea, chest pain, syncope (outflow obstruction), palpitations (arrhythmias).
- **Exam:** Systolic murmur (increases with Valsalva), S4 gallop, LVH signs (EKG), apical impulse displacement.

Restrictive Cardiomyopathy (RCM):

- **Symptoms:** Right-sided HF (edema, ascites), fatigue, dyspnea (diastolic dysfunction), palpitations.
- **Exam:** Elevated JVD, Kussmaul's sign (JVD increase on inspiration), hepatomegaly, edema, S3/S4 gallop.

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC):

- **Symptoms:** Palpitations, syncope (ventricular arrhythmias), exertional dyspnea, SCD risk.
- **Exam:** RV heave, irregular rhythm (VT), signs of RV failure (edema, JVD).

Other Types:

- **Takotsubo Cardiomyopathy:** Acute chest pain, dyspnea (stress-induced), mimics MI; often in postmenopausal women.
- **Peripartum Cardiomyopathy (PPCM):** HF symptoms late pregnancy/postpartum, fatigue, edema.

Causes

Dilated Cardiomyopathy (DCM):

- **Genetic:** 20-35% (e.g., TTN, LMNA mutations).
- **Acquired:** Ischemic (post-MI), toxins (alcohol, chemotherapy like doxorubicin), viral myocarditis, peripartum, metabolic (e.g., thiamine deficiency).

Hypertrophic Cardiomyopathy (HCM):

- **Genetic:** Sarcomere gene mutations (e.g., MYH7, MYBPC3); autosomal dominant.
- **Acquired:** Rarely secondary (e.g., hypertension, amyloidosis in older patients).

Restrictive Cardiomyopathy (RCM):

- **Primary:** Idiopathic, genetic (e.g., TTR mutations).
- **Secondary:** Amyloidosis (AL, ATTR), sarcoidosis, hemochromatosis, radiation, endomyocardial fibrosis.

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC):

- **Genetic:** Desmosomal gene mutations (e.g., PKP2, DSG2); autosomal dominant.
- **Acquired:** Rarely secondary (e.g., myocarditis mimicking ARVC).

Other Types:

- **Takotsubo:** Emotional/physical stress (catecholamine surge).
- **Peripartum:** Late pregnancy/postpartum; multifactorial (hormonal, immune, genetic).

Diagnostic Studies and Labs

Labs:

- **BNP/NT-proBNP:** Elevated in HF (all types).
- **Troponin:** Elevated in acute presentations (e.g., Takotsubo, myocarditis, ischemic DCM).
- **CMP:** Renal/hepatic function (HF complications), electrolytes (arrhythmia risk).
- **CBC:** Anemia (HF exacerbation), leukocytosis (infection in PPCM).
- **Specific Tests:**

o Genetic testing (HCM, ARVC, familial DCM).

o Serum ferritin, TIBC (hemochromatosis in RCM).

o Serum free light chains, SPEP (amyloidosis in RCM).

o TSH (thyroid dysfunction in DCM).

Diagnostic Studies:

- **EKG:**
  - **DCM:** LBBB, poor R-wave progression, arrhythmias.
  - **HCM:** LVH, Q waves, dagger-like ST-T changes.
  - **RCM:** Low voltage (amyloidosis), conduction abnormalities.
  - **ARVC:** Epsilon waves, T-wave inversions in V1-V3, VT.
- **ECHO:**
  - **DCM:** Dilated LV, reduced EF (<40%), global hypokinesis.
  - **HCM:** LVH (septal >15 mm), outflow obstruction, SAM (systolic anterior motion of mitral valve).
  - **RCM:** Normal/small LV, biatrial enlargement, restrictive filling pattern (E/A >2).
  - **ARVC:** RV dilation, hypokinesis, aneurysms.
- **Cardiac MRI:**
  - **DCM:** Late gadolinium enhancement (LGE) in non-ischemic pattern.
  - **HCM:** Asymmetric hypertrophy, LGE (fibrosis, SCD risk).
  - **RCM:** Amyloidosis (global LGE), sarcoidosis (patchy LGE).
  - **ARVC:** RV fatty infiltration, fibrosis, wall motion abnormalities.
- **Other:**
  - **Coronary Angiography:** Rule out ischemic cause in DCM.
  - **Endomyocardial Biopsy:** Amyloidosis, sarcoidosis in RCM; rarely needed.

- **Holter Monitor:** Arrhythmias in ARVC, HCM.

Evaluation

Stepwise Approach:

- History and Exam:
  - **Symptom onset:** Acute (Takotsubo, PPCM) vs. chronic (HCM, DCM).
  - **Family history:** SCD, cardiomyopathy (HCM, ARVC, familial DCM).
  - **Precipitants:** Stress (Takotsubo), pregnancy (PPCM), toxins (DCM).
  - **Exam:** Signs of HF (JVD, crackles), murmurs (HCM), RV failure (ARVC).
- Initial Workup:
  - EKG, ECHO, BNP, troponin to assess HF, ischemia, arrhythmias.
  - CMP, CBC to evaluate organ function, anemia, infection.
- Advanced Testing:
  - Cardiac MRI for tissue characterization (e.g., amyloidosis, ARVC).
  - Genetic testing if familial history or young onset.
  - Coronary angiography if ischemic etiology suspected (DCM).
- Identify Cause:
  - Rule out secondary causes (e.g., amyloidosis, hemochromatosis in RCM).
  - Assess for precipitants (e.g., infection in PPCM, stress in Takotsubo).

Consultants

- **Cardiology:** Primary consultant for all cardiomyopathies; manages HF, arrhythmic risk, device therapy (ICD, CRT).
- **Electrophysiology (EP):** For ARVC, HCM (SCD risk, ICD implantation), DCM (VT ablation, CRT).
- **Heart Failure/Transplant:** For advanced HF in DCM, PPCM (LVAD, transplant evaluation).
- **Genetics:** For HCM, ARVC, familial DCM (genetic counseling, testing).
- **Rheumatology:** For RCM (e.g., sarcoidosis, amyloidosis workup).
- **Hematology:** For amyloidosis in RCM (AL amyloidosis, chemotherapy).
- **Obstetrics:** For PPCM (peripartum management, future pregnancy risks).

Complications

- **Heart Failure:** Common in DCM, RCM, PPCM; presents with dyspnea, edema, reduced EF or diastolic dysfunction.

- **Arrhythmias:** VT/VF in ARVC, HCM, DCM; atrial arrhythmias (AF) in all types; increases SCD risk.
- **Sudden Cardiac Death (SCD):** High risk in HCM (LVH, syncope), ARVC (VT), DCM (EF <35%).
- **Thromboembolism:** Intracardiac thrombus in DCM (dilated LV, low EF), AF-related stroke in all types.
- **Pulmonary Hypertension:** Secondary to LV dysfunction in DCM, RCM; presents with RV failure.
- **Cardiogenic Shock:** Severe cases of DCM, PPCM, Takotsubo; presents with hypotension, organ hypoperfusion.

Treatment

General Management:

- **Lifestyle:** Salt restriction (<2 g/day), fluid restriction in HF, avoid alcohol/toxins (DCM).
- **HF Therapy:**
  - **Beta-Blockers:** Metoprolol succinate 25-200 mg PO daily (all types except acute Takotsubo).
  - **ACEi/ARBs/ARNIs:** Lisinopril 5-40 mg PO daily or sacubitril/valsartan 49/51 mg PO BID (DCM, PPCM, RCM).
  - **Diuretics:** Furosemide 20-80 mg IV/PO daily for volume overload (all types).
  - **Aldosterone Antagonists:** Spironolactone 25-50 mg PO daily (DCM, PPCM, RCM with EF <35%).
- **Arrhythmia/SCD Prevention:**
  - **ICD:** For HCM (SCD risk: syncope, LVH >30 mm), ARVC (VT), DCM (EF <35%, NYHA II-III).
  - **Amiodarone:** 200 mg PO daily for VT (ARVC, DCM).

Specific Treatments:

- **DCM:**
  - HF therapy (above), anticoagulation if AF or thrombus (e.g., apixaban 5 mg PO BID).
  - CRT if EF <35%, LBBB, QRS >150 ms.
  - **Treat cause:** E.g., alcohol cessation, chemotherapy modification.
- **HCM:**
  - **Beta-Blockers:** Metoprolol 50-200 mg PO daily (first-line for symptoms).

    - **Non-Dihydropyridine CCBs:** Verapamil 120-360 mg PO daily (if beta-blockers fail).
    - **Septal Reduction:** Myectomy or alcohol ablation if outflow obstruction (gradient >50 mmHg).
    - **Avoid:** Inotropes, vasodilators (worsen obstruction).
- **RCM:**
    - Diuretics for volume overload, cautious use (low preload dependence).
    - **Treat cause:** E.g., chemotherapy for amyloidosis, steroids for sarcoidosis.
    - **Avoid:** Beta-blockers, CCBs (negative inotropy worsens diastolic dysfunction).
- **ARVC:**
    - **Beta-Blockers:** Metoprolol 25-100 mg PO daily (arrhythmia control).
    - ICD for VT/SCD risk.
    - **Avoid:** Competitive sports (increases VT risk).
- **Takotsubo:**
    - **Supportive:** Beta-blockers, ACEi for transient LV dysfunction.
    - **Avoid:** Inotropes (catecholamine surge worsens).
- **PPCM:**
    - HF therapy (above), bromocriptine 2.5 mg PO BID x 2 weeks (prolactin inhibition).
    - **Anticoagulation:** Warfarin or LMWH (high thrombosis risk in pregnancy/postpartum).

Key Pearls

- **Presentation:** DCM (HF), HCM (exertional syncope), RCM (right-sided HF), ARVC (palpitations, VT), Takotsubo (stress-induced), PPCM (peripartum HF).
- **Causes:** Genetic (HCM, ARVC), acquired (DCM: toxins, myocarditis; RCM: amyloidosis).
- **Diagnosis:** EKG (arrhythmias, LVH), ECHO (chamber size, function), MRI (tissue characterization).
- **Evaluation:** History/exam, EKG/ECHO, MRI, genetic testing, rule out secondary causes.
- **Consultants:** Cardiology (all), EP (arrhythmias), HF/transplant (advanced HF), genetics (familial).
- **Complications:** HF, arrhythmias, SCD, thromboembolism, pulmonary HTN, cardiogenic shock.
- **Treatment:** HF therapy (beta-blockers, ACEi), ICD (SCD risk), treat cause (e.g., bromocriptine for PPCM).

References

- **UpToDate:** “Cardiomyopathies: Diagnosis and Management” (2025). UpToDate Cardiomyopathy
- **AHA:** “Guidelines for the Management of Cardiomyopathies” (2024). AHA Guidelines
- **ESC:** “Diagnosis and Treatment of Hypertrophic Cardiomyopathy” (2023). ESC Guidelines
- **NEJM:** “Advances in the Management of Peripartum Cardiomyopathy” (2024). NEJM PPCM

Case Scenarios

Case 1: A 40-Year-Old Male with Dilated Cardiomyopathy and HF

- Presentation:A 40-year-old male with a history of alcohol use presents with 2 months of dyspnea, fatigue, and leg swelling. Exam shows T 37°C, BP 110/70 mmHg, HR 90 bpm, JVD, S3 gallop, crackles, 2+ edema.
- Labs/Studies:**BNP 800 pg/mL, troponin normal, EKG:** LBBB, ECHO: EF 30%, dilated LV, global hypokinesis, MRI: Non-ischemic LGE.
- Diagnosis: Dilated Cardiomyopathy (Alcohol-Induced) → HF symptoms, reduced EF, alcohol history.
- Management: Admit for HF management. Start furosemide 40 mg IV BID, lisinopril 5 mg PO daily, metoprolol succinate 25 mg PO daily, spironolactone 25 mg PO daily. Consult cardiology (EF <35%, NYHA III). Holter monitor: No VT. Alcohol cessation counseling. Monitor for arrhythmias (none noted). EF improves to 35% at 1 month. Discharge with HF meds, ICD consideration, follow-up with cardiology.

Case 2: A 25-Year-Old Male with Hypertrophic Cardiomyopathy and Syncope

- Presentation: A 25-year-old male presents with exertional syncope. Family history of SCD. Exam shows T 37°C, BP 120/80 mmHg, HR 70 bpm, systolic murmur (increases with Valsalva), S4 gallop.
- Labs/Studies: **EKG:** LVH, Q waves in inferior leads, ECHO: Septal thickness 20 mm, outflow gradient 60 mmHg, MRI: LGE in septum (SCD risk).
- Diagnosis: Hypertrophic Cardiomyopathy → Exertional syncope, LVH, outflow obstruction, family history.

- Management: Admit for evaluation. Start metoprolol 50 mg PO daily (symptom control). Consult cardiology/EP: High SCD risk (syncope, LGE, family history). ICD placed. Avoid competitive sports. Genetic testing (MYH7 mutation). Monitor for VT (none noted). BP stable, syncope resolves. Discharge with metoprolol 100 mg PO daily, cardiology follow-up.

Case 3: A 50-Year-Old Female with Restrictive Cardiomyopathy and Amyloidosis

- Presentation: A 50-year-old female presents with 3 months of edema, ascites, and fatigue. Exam shows T 37°C, BP 100/65 mmHg, HR 80 bpm, elevated JVD, Kussmaul's sign, hepatomegaly, 3+ edema.
- Labs/Studies: **BNP 600 pg/mL, EKG:** Low voltage, ECHO: Normal LV, biatrial enlargement, restrictive filling (E/A >2), MRI: Global LGE (amyloidosis), biopsy: AL amyloidosis confirmed.
- Diagnosis: Restrictive Cardiomyopathy (AL Amyloidosis) → Right-sided HF, restrictive pattern, amyloidosis on biopsy.
- Management: Admit for HF management. Start furosemide 20 mg IV BID (cautious, low preload). Consult cardiology, hematology: Chemotherapy for AL amyloidosis (cyclophosphamide, bortezomib, dexamethasone). Avoid beta-blockers (diastolic dysfunction). Monitor for AF (none noted). BP stable, edema improves. Discharge with diuretics, hematology follow-up for amyloidosis treatment.