

CARDIOMYOPATHIES

Relevant Guidelines

AHA/ACC 2024 guidelines on HCM.

2024 AHA/ACC/AMSSM/HRS/PACES/SCMR Guideline for the Management of Hypertrophic Cardiomyopathy: A Report of the American Heart Association/American College of Cardiology Joint Committee on Clinical Practice Guidelines. *Circulation*. 2024.

ACCF/AHA 2022 guideline on heart failure

2022 AHA/ACC/HFSA Guideline for the Management of Heart Failure. *Circulation* 2022

ESC 2014 guidelines on HCM

2014 ESC Guidelines on diagnosis and management of hypertrophic cardiomyopathy. *Eur Heart J*. 2014; 35:2733-79.

ESC 2023 guidelines on cardiomyopathies

2023 ESC Guidelines for the management of cardiomyopathies. *Eur Heart J*. 2023

ESC 2026 guidelines on heart failure

2026 ESC Guidelines for the management of heart failure

ESC 2022 Guidelines on ventricular arrhythmias and prevention of sudden cardiac death

2022 ESC Guidelines for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death. *Eur Heart J*. 2022.

AHA/ACC/HRS 2017 Guideline on ventricular arrhythmias and prevention of sudden cardiac death

2017 AHA/ACC/HRS Guideline for Management of Patients With Ventricular Arrhythmias and the Prevention of Sudden Cardiac Death. *JACC*. 2017

AHA/ACC 2024 Guideline on noncardiac surgery

2024 AHA/ACC/ACS/ASNC/HRS/SCA/SCCT/SCMR/SVM Guideline for Perioperative Cardiovascular Management for Noncardiac Surgery. *JACC* 2024

ESC 2022 Guidelines on cardio-oncology

2022 ESC Guidelines on cardio-oncology developed in collaboration with the European Hematology Association (EHA), the European Society for Therapeutic Radiology and Oncology (ESTRO) and the International Cardio-Oncology Society (IC-OS). *Eur Heart J*. 2022.

ESC 2025 guidelines on myocarditis and pericarditis

2025 ESC Guidelines for the management of myocarditis and pericarditis. *Eur Heart J*. 2025.

ESC 2026 Guidelines on Rehabilitation

2026 ESC Guidelines on cardiac rehabilitation. *Eur Heart J*. 2026.

ESC 2025 guidelines on pregnancy.

ESC Guidelines on the management of cardiovascular diseases and pregnancy. *Eur Heart J*. 2025.

Relevant Scientific Statements and Position Papers

AHA 2016 Scientific Statement on Dilated Cardiomyopathies

Current Diagnostic and Treatment Strategies for Specific Dilated Cardiomyopathies: A Scientific Statement From the American Heart Association. *Circulation* 2016; 134:e579-e646.

AHA 2017 Statement on Cardiac Involvement in Neuromuscular Diseases

Management of Cardiac Involvement Associated With Neuromuscular Diseases: A Scientific Statement From the American Heart Association. *Circulation*. 2017.

EHRA/HRS/APHRS/SOLAECE 2016 consensus on atrial cardiomyopathies

EHRA/HRS/APHRS/SOLAECE expert consensus on atrial cardiomyopathies: definition, characterization, and clinical implication. *Europace* 2016 E-Pub

AHA 2018 statement on Chagas cardiomyopathy

Chagas Cardiomyopathy: An Update of Current Clinical Knowledge and Management. A Scientific Statement From the American Heart Association. *Circulation* 2018.

HRS 2019 expert consensus statement on arrhythmogenic cardiomyopathy

2019 HRS expert consensus statement on evaluation, risk stratification, and management of arrhythmogenic cardiomyopathy. *Heart Rhythm*. 2019.

AHA 2020 Scientific Statement on Cardiac Amyloidosis

Cardiac Amyloidosis: Evolving Diagnosis and Management: A Scientific Statement From the American Heart Association. *Circulation*. 2020:

EHRA/HRS/APHRS/LAHRs 2022 Expert Consensus Statement on Genetic Testing

Expert Consensus Statement on the State of Genetic Testing for Cardiac Diseases. *Europace* 2022

AHA 2024 Statement on Cardiovascular Disease Risk After Adverse Pregnancy Outcomes

Opportunities in the Postpartum Period to Reduce Cardiovascular Disease Risk After Adverse Pregnancy Outcomes: A Scientific Statement From the American Heart Association. *Circulation*. 2024;149:e330-e346.

AHA 2020 Scientific Statement on Cardiac Sarcoidosis

Diagnosis and Management of Cardiac Sarcoidosis: A Scientific Statement From the American Heart Association. *Circulation*. 2024.

ESC Working Groups and EHRA 2024 Statement on Cardiac Sarcoidosis

Management of cardiac sarcoidosis: A clinical consensus statement of the Heart Failure Association, the European Association of Cardiovascular Imaging, the ESC Working Group on Myocardial & Pericardial Diseases, and the European Heart Rhythm Association of the ESC *Eur Heart J*. 2024;45:2697-2726.

AHA 2025 Policy Statement on Genetic and Genomic Testing

Genetic and Genomic Testing in Cardiovascular Disease: A Policy Statement From the American Heart Association. *Circulation*. 2025 Dec 16;152(24):e474-e489.

ESC HFA 2026 Statement on amyloidosis

Non-amyloid specific treatment for transthyretin cardiac amyloidosis: a clinical consensus statement of the ESC Heart Failure Association. *Eur Heart J*. 2026;47:22-36.

ESC Working Groups 2026 Statement on DCM

Genetic counselling implementation in dilated cardiomyopathy. *Eur Heart J*. 2026;47:3040-3051.

ESC Groups 2026 Statement on arrhythmias in myocarditis

Workup and management of rhythm disorders in myocarditis and inflammatory cardiomyopathy: a clinical consensus statement of the European Heart Rhythm Association and the Heart Failure Association of the ESC, the ESC Working Group on Myocardial & Pericardial Diseases, the European Association of Preventive Cardiology of the ESC, the Heart Rhythm Society, the Asian Pacific Heart Rhythm Society, and the Latin American Heart Rhythm Society. *Europace*. 2026;28.

CLASSIFICATION DIAGNOSIS and GENERAL PRINCIPLES OF THERAPY

Classification

Classification schemes have been proposed by WHO (1995), the AHA (2006), and the ESC (2008). Recently, the MOGE(S) scheme attempted a comprehensive classification.

American Heart Association

Definition

A heterogeneous group of diseases of the myocardium associated with mechanical and/or electrical dysfunction that usually (but not invariably) exhibit inappropriate ventricular hypertrophy or dilatation and are due to a variety of causes that frequently are genetic. Cardiomyopathies either are confined to the heart or are part of generalized systemic disorders, often leading to cardiovascular death or progressive heart failure–related disability.¹

Classification

Primary cardiomyopathies are those solely or predominantly confined to heart muscle.

Secondary cardiomyopathies show pathological myocardial involvement as part of a large number and variety of generalized systemic (multiorgan) disorders. Ion channelopathies are considered forms of primary genetic cardiomyopathy.²

European Society of Cardiology

Definition

Myocardial disorders in which the heart muscle is structurally and functionally abnormal, in the absence of coronary artery disease, hypertension, valvular disease and congenital heart disease sufficient to cause the observed myocardial abnormality.³

Classification

Cardiomyopathies are classified into the conventional phenotypes of ***hypertrophic cardiomyopathy, dilated cardiomyopathy, non-dilated left ventricular cardiomyopathy, arrhythmogenic right ventricular dysplasia, and other traits and syndromes*** (such as ***left ventricular non-compaction, and Takotsubo syndrome***).

Aetiology

CMPs can be either inherited (genetic/familial) and/or acquired (Table 1). Genes associated with monogenic, non-syndromic cardiomyopathies are presented in Appendix 2. They can also be accelerated by disease modifiers.

Diagnosis

A diagnostic workflow is presented in Figure 1, and Tables 2 and 3. Morphological traits and features that should raise the suspicion of a cardiomyopathy are presented in Tables 4-8.⁴

Atrial cardiomyopathies may occur in several clinical settings, and have a significant impact on atrial function and arrhythmogenesis (Table 9).⁵ P wave terminal force (PTF $\geq 5000 \mu\text{V}$ calculated as the duration x amplitude of the negative part of the P-wave in lead V1), prolonged P-wave ($>120 \text{ ms}$), LA dilation ($\text{LAVI}_{\text{Max}} > 60 \text{ mL/m}^2$ or $\text{LAVI}_{\text{Min}} > 30 \text{ mL/m}^2$), and LA mechanical dysfunction (LA emptying fraction $< 45\%$), are atrial cardiomyopathy markers associated with AFm heart failure and stroke.⁶ Atrial failure is a new clinical entity that has been proposed as a consequence of atrial dysfunction.⁷ Evidence for a genetic component in left atrial morphology and function (on 18 genetic loci associated with left atrial volume and function) supports the concept of atrial cardiomyopathy as an intrinsic disease entity.^{8, 9}

Laboratory tests

An overview and recommendations are presented in Tables 10 and 11.

Imaging

Multimodality imaging in clinical suspicion of cardiomyopathy is presented in Figure 2. Recommendations and comments on echocardiography are presented in Table 12, on cardiac magnetic resonance (CMR) in Table 13 and Figure 3, on CT and nuclear imaging in Table 14, and on endomyocardial biopsy in Table 15.

Genetic Testing

Genes implicated in non-syndromic cardiomyopathies are summarized in Appendix 2. The list is continually expanding. Familial forms of cardiomyopathies show diverse modes of inheritance. Mendelian (monogenic) disease genes that most commonly display autosomal dominant inheritance, although other inheritance patterns including autosomal recessive, X-linked, and mitochondrial (matrilineal) are also observed.³

Genetic testing is now essential in the management of patients with cardiomyopathy (Tables 16-19).¹⁰ Family screening and follow-up are presented in Figure 4, and Table 18. Considerations for children are presented in Figures 5 and 6.

General Principles of Therapy

Patients with subtle structural abnormalities with cardiomyopathy may remain asymptomatic and have a normal lifespan. Other patients may develop symptoms, often many years after the appearance of ECG or imaging evidence of disease.³ Identification and management of risk factors and concomitant diseases is recommended as an integral part of the management of cardiomyopathy patients (Table 20). An overview of the proposed management of patients by the ESC is provided in Figure 7.

Heart Failure

Although recommendations of heart failure in general apply to patients with cardiomyopathy, However, therapy should be individualized according to specific genetic causes as described in specific disease entities. Recommendations for mechanical assistance are provided in Table 21.

Atrial fibrillation

Atrial fibrillation is the most common arrhythmia in all subtypes of cardiomyopathies and is associated with an increased risk of cardio-embolic events, heart failure, and death.³ Oral anticoagulation is recommended according to the CHA₂DS₂-VAS_c score in both AF and atrial flutter (Tables 22 and 23).

Ventricular Arrhythmias

Ventricular arrhythmias are responsible for a significant increase in mortality of patients, especially with arrhythmogenic cardiomyopathy. Specific recommendations are provided in Chapters on nonspecific cardiomyopathies. General recommendations on ICD therapy are presented in Table 24.

Sports

An individualized risk assessment for exercise prescription is mandatory. However, regular low- to moderate-intensity exercise is recommended in all able individuals with cardiomyopathy (Table 25). Specific instructions are provided in individual Chapters.

Follow-up and Psychological Support

Recommendations for routine follow-up are presented in Table 26, and for psychological support in Tables 27 and 28. Recommendations for reproductive issues and non-cardiac surgery are presented in Tables 29 and 30. General guidance for daily activity is presented in Table 31.

Pregnancy

Recommendations are presented in Table 32.

References

1. Maron BJ, et al. Contemporary definitions and classification of the cardiomyopathies: an American Heart Association Scientific Statement from the Council on Clinical Cardiology, Heart Failure and Transplantation Committee; Quality of Care and Outcomes Research and Functional Genomics and Translational Biology Interdisciplinary Working Groups; and Council on Epidemiology and Prevention. *Circulation*. 2006;113:1807-16.
2. Lehnart SE, et al. Inherited arrhythmias: a National Heart, Lung, and Blood Institute and Office of Rare Diseases workshop consensus report about the diagnosis, phenotyping, molecular mechanisms, and therapeutic approaches for primary cardiomyopathies of gene mutations affecting ion channel function. *Circulation*. 2007;116:2325-45.
3. Arbelo E, et al. 2023 ESC Guidelines for the management of cardiomyopathies. *Eur Heart J*. 2023.
4. Rapezzi C, et al. Diagnostic work-up in cardiomyopathies: bridging the gap between clinical phenotypes and final diagnosis. A position statement from the ESC Working Group on Myocardial and Pericardial Diseases. *Eur Heart J*. 2013;34:1448-58.
5. Goette A, et al. EHRA/HRS/APHRS/SOLAECE expert consensus on atrial cardiomyopathies: definition, characterization, and clinical implication. *Europace*. 2016;18:1455-1490.
6. Vad OB, et al. Atrial cardiomyopathy: markers and outcomes. *Eur Heart J*. 2026;47:235-246.
7. Fisbal F and al. e. Atrial Failure as a Clinical Entity. *JACC*. 2020.
8. Ahlberg G, et al. Genome-wide association study identifies 18 novel loci associated with left atrial volume and function. *Eur Heart J*. 2021.
9. Camen S and Schnabel RB. Genetics, atrial cardiomyopathy, and stroke: enough components for a sufficient cause? *Eur Heart J*. 2021.
10. Elliott P, et al. Integration of genetic testing into diagnostic pathways for cardiomyopathies: a clinical consensus statement by the ESC Council on Cardiovascular Genomics. *Eur Heart J*. 2024.