Position statement of Polish Cardiac Society experts on cardiomyopathy

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DOI: 10.33963/v.phj.102977

Received:
September 30, 2024

Accepted:
October 3, 2024

Early publication date:
October 7, 2024

ABSTRACT

Cardiomyopathies (CMs) are a very broad group of diseases, including genetically determined and acquired, and their classification is based on phenotypic characteristics. There is always a need to search for the etiology (often also to try to identify the genetic cause), which may determine the appropriate choice of clinical management. The geographical distribution of genetic variants varies as does the prevalence across populations, ethnic groups, regions, and countries. The most reliable data on the distribution of individual genetic variants come from developed countries. The phenotypic classification includes 5 main types of CM, i.e., dilated CM, hypertrophic, restrictive, arrhythmogenic right ventricular CM, and non-dilated left ventricular (LV) CM. Individual CMs are characterized by a variety of causes and different phenotypic pictures, which affect their presentation, diagnosis, and response to treatment. Within each type of CM, there are both familial and sporadic (acquired) forms. The complex presentation of CM, as well as the limited availability of screening and diagnostic tests, causes CMs to be diagnosed late, often at an advanced stage of the disease. Therapeutic management of CM is strictly determined by its type and clinical picture. Diagnostics include the assessment of symptoms, the results of imaging and genetic tests, as well as morphological, functional, and often histological assessment. This allows for personalized and dedicated clinical management. To optimize the

diagnosis, treatment, and care of patients with CMs, an individualized, expert, systemic, coordinated, and often multidisciplinary structure of care is necessary. Hence, it is important to create multidisciplinary teams for CM management. We present examples of existing systemic solutions for the care of CM patients in Europe (France and Spain). Not all of these options are available to patients in our country. This article presents issues related to CM and may be the basis for developing a diagnostic and therapeutic model allowing for earlier detection of CM in Polish patients and their effective treatment.

Key words: cardiomyopathy, Polish Cardiac Society, position of experts

WHAT ONE SHOULD KNOW ABOUT CARDIOMYOPATHIES

Cardiomyopathies (CMs) are defined as diseases of the heart muscle associated with structural and functional abnormalities that cannot be attributed to the presence of hypertension, coronary artery disease, valvular heart disease, or congenital heart defects. Such a broad definition includes a variety of congenital (genetically determined) and acquired disease entities in which several pathological changes occur, such as hypertrophy of the ventricular muscles, dilation of the heart cavities, occurrence of localized scars, or other abnormalities in imaging tests. From the point of view of heart failure (HF), CMs can be classified according to the occurrence of systolic dysfunction (global and/or segmental) and/or LV diastolic dysfunction (restrictive profile) [1].

The complex etiology and multitude of clinical manifestations make it difficult to create a unified classification of CM. In light of the latest recommendations of the European Society of Cardiology (ESC), it is postulated that CM should be divided according to its phenotype, manifested by appropriate clinical symptoms, as well as structural and functional pathology found in imaging studies (Figure 1). The current approach emphasizes the need to search for the etiology responsible for the occurrence of a given phenotype, as it determines the appropriate path of clinical management.

The phenotypic classification includes 5 main types of CM, i.e., dilated CM (DCM), hypertrophic CM (HCM), restrictive CM, arrhythmogenic right ventricular CM (ARVC), and non-dilated LV CM (NDLVC) [1]. In familial forms, different phenotypes of CM can occur in members of the same family and progress from one to the other. Current definitions of CM and the basis for diagnosis are summarized in Table 1. It should be noted that within each of the 5 main CM types, there are familial forms in which the genetic background can be determined (e.g., 60% of HCM cases and 30% of DCM cases) and sporadic (acquired) forms. In the latter case, more and more data indicate the importance of genetic susceptibility, which is showed only after contact with the appropriate pathogen or toxic substance (second-blow theory) [1].

Limited availability of screening and diagnostic tests, as well as limitations of systemic health care solutions, mean that CMs are diagnosed late, often at an advanced stage of the disease. According to data from the ESC Cardiomyopathy Registry (2012–2016), the mean age of diagnosis of HCM in 2016 was 47 years, DCM 49 years, ARVC 39 years, and RCM 57 years.

Cardiomyopathies can be asymptomatic for a long time — most often in the case of carriers of mutations in familial forms at a young age, often in the case of HCM.

However, the majority of patients (85%–97%) present symptoms at the time of diagnosis [7]. The most common symptoms are those of HF, according to the British authors. This problem affects 66% of patients with CM, including 66% of patients with DCM, 62% of patients with RCM, 29% of patients with ARVC, and 27% of patients with HCM [8]. Other symptoms that CM patients may present include

- symptoms of LV outflow tract obstruction in the course of HCM — dyspnea due to exertion, dizziness, chest pain, presyncope, and syncope; HCM specific;
- symptoms of ischemic heart disease, secondary, e.g. to myocardial hypertrophy in the course of HCM — angina chest pain;
- symptoms of atrial and ventricular arrhythmias — palpitations, presyncope, or fainting;
- sudden cardiac arrest by ventricular tachycardia mechanism without pulse or ventricular fibrillation.

Analyzing the current classification, it should be noted that currently, the following diseases are not considered CM: channelopathies, LV non-compaction, peripartum CM, and stress-induced CM (takotsubo syndrome). LV non-compaction, also known as hypertrabeculation or elevated LV trabeculae, has been defined as a feature that may co-occur with various major phenotypes of CM or occur in an isolated form but does not constitute a separate type of CM [9].

HOW DO CARDIOMYOPATHIES AFFECT LIFE EXPECTANCY AND QUALITY OF LIFE?

CMs are characterized by a variety of pathophysiological causes and mechanisms, as well as different phenotypic expressions, which affects their presentation and response to treatment [10, 11]. Individual CMs can have different effects on life expectancy and quality of life. The worst prognosis is characterized by DCM and RCM, in these patients, HF symptoms can significantly worsen the quality of life.

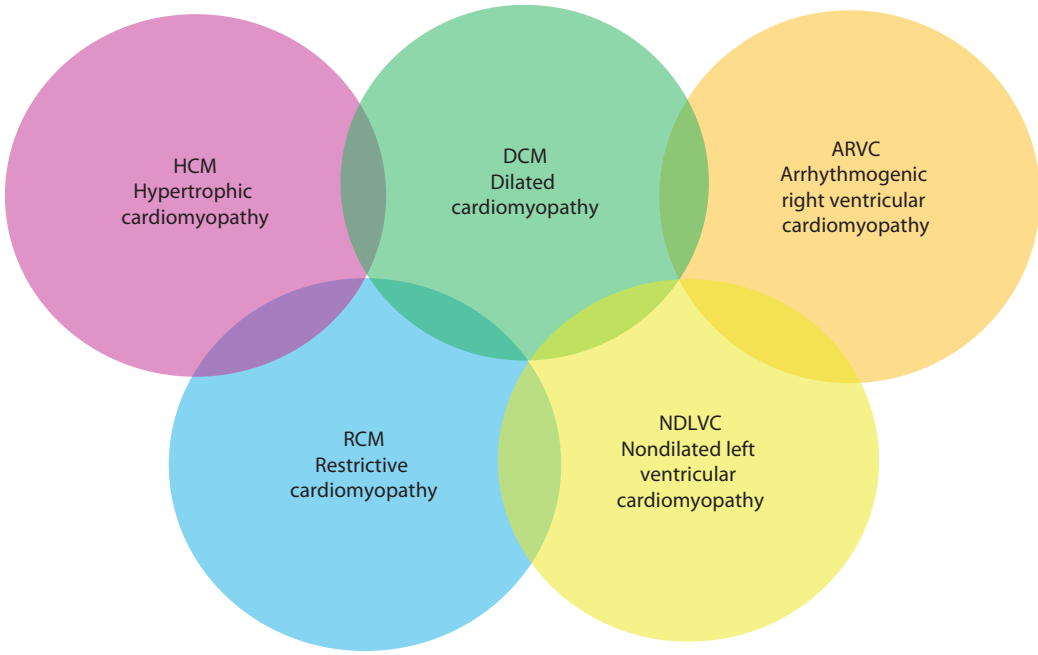
Dilated cardiomyopathy

DCM is characterized by progressive dilatation of the ventricular cavity of the heart with impaired contractility. Clinical presentation includes HF symptoms, ventricular

Phenotype assessment				
Signs and symptoms:	**Family history:**	**TTE / CMR — Cardiac morphology assessment:**	**ECG/Holter ECG deviations:**	**Laboratory tests:**
• Dyspnea • Chest pain • Palpitations • Syncope/presyncope • Sudden cardiac arrest • Murmur over the heart • Signs and symptoms of heart failure	• Introduction to genetic diagnostics	• Hypertrophy • Dilatation • Features of systolic/diastolic dysfunction • Scar of non-ischemic etiology	• Ventricular arrhythmia • AF/AFl • Resting ECG: Ypsilon wave; RBBB; LBBB, AVB I st.	• Natriuretic peptides • Troponin • CK-MB

+

Assessment of etiology				
• Genetic evaluation of proband and relatives	• Non-cardiac pathology	• Detailed laboratory tests	• Detailed laboratory tests	• Histopathology – endomyocardial biopsy



REMARK! Currently, the following are not considered cardiomyopathies: channelopathies, left ventricular non-compaction, peripartum cardiomyopathy and stress-induced cardiomyopathy

Figure 1. Phenotype and etiology assessment with the current division of cardiomyopathies

Abbreviations: AF, atrial fibrillation; AFl, atrial flutter; AVB, atrioventricular block; CK-MB, cardiac creatine kinase fraction; CMR, cardiac magnetic resonance; LBBB, left bundle branch block; RBBB, right bundle branch block; TTE transthoracic echocardiography

arrhythmias, or sudden cardiac death (SCD). Clinical symptoms usually occur suddenly or intensify quite quickly within a dozen or so days and are the main reason for initial hospitalization and/or contact with a doctor.

The natural history of HF in DCM can be characterized by three distinct pathways:

- structural and functional reconstruction after an acute HF incident;
- remission of HF symptoms and improvement/stabilization of LV systolic function;
- progression to advanced HF leading to heart transplantation/mechanical support or death [1].

Prompt initiation of comprehensive treatment of HF is a key element in improving prognosis. Complete recovery of function and structure is rare and can occur if acute injury has not resulted in significant loss of myocytes, allowing

Table 1. Summary of criteria for diagnosis of the 5 main types of cardiomyopathy

Definitions of the basic types of primary cardiomyopathy				
Common element: The observed pathology is not secondary to the occurrence of: • Hypertension • Ischemic heart disease, including coronary artery disease • Valvular heart disease				
DCM [2]	**HCM [3]**	**RCM [4]**	**NDLVC [5]**	**ARVC [6]**
• With the presence of global or segmental contractility disorders that cannot be explained by abnormal filling conditions • LVEF <50 % based on TTE/CMR/SPECT + LVEDD >2 SD, i.e. z-score>2 of the predicted value for age, sex and BSA: LVEDD >58 mm, LVEDV 75 ml/m^2; for women – LVEDD >52 mm, LVEDV >62 ml/m^2	• Thickness of any LV segment ≥15 mm, which cannot be explained by incorrect filling conditions; preferred SAX projection • in first-degree relatives of a patient with confirmed HCM, LV hypertrophy ≥13 mm • in children, myocardial hypertrophy with LV thickness >2 SD (z-score >2)	• Presence of features of diastolic dysfunction with echocardiographic features of restriction • HFpEF-typical echocardiographic profile • Left ventricular muscle thickness within the normal range or benign hypertrophy	• Presence of non-ischemic scar or LV fatty infiltrates without LV dilation with/or without the presence of segmental/global contractility disorders or • Presence of LV systolic dysfunction with LVEF <50% without scarring	• Task Force criteria from 2010: — meeting 2 major criteria; — 1 large and 2 small or 4 small

Abbreviations: ARVC, arrhythmogenic right ventricular cardiomyopathy; BSA, body surface area; CMR, cardiac magnetic resonance; DCM, dilated cardiomyopathy; FS, shortening fraction; HCM, hypertrophic cardiomyopathy; HFpEF, heart failure with preserved left ventricular ejection fraction; LV, left ventricle; LVEDD, left ventricular end-diastolic dimension; LVEDV, left ventricular end-diastolic volume; LVEF, left ventricular ejection fraction; NDLVC, non-dilated left ventricular cardiomyopathy; RCM, restrictive cardiomyopathy; SD, standard deviation; SPECT, single-photon emission tomography; TTE, transthoracic echocardiography

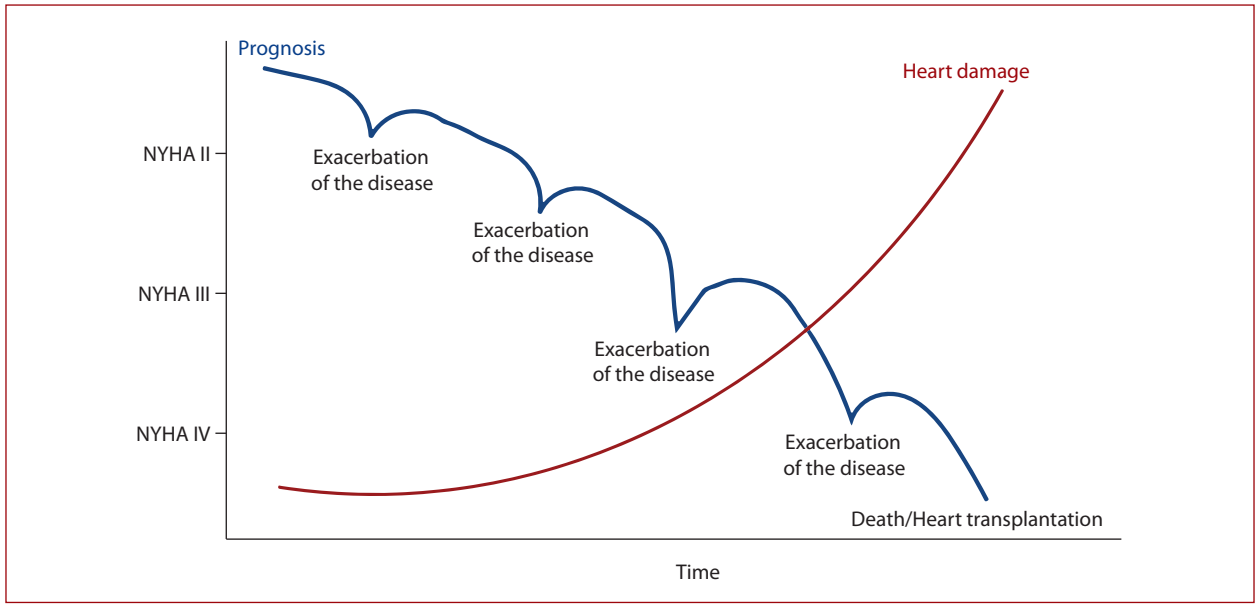


Figure 2. Natural history of patients with dilated cardiomyopathy

Abbreviations: NYHA, New York Heart Association

for normalization of LV function. Most often, in DCM patients, there is a gradual deterioration of LV systolic function accompanied by progressive impairment of physical performance, interspersed with successive exacerbations of HF symptoms (Figure 2). The most common effect of treatment is to slow down or stop the progression of the disease, to prolong the periods between exacerbations of the disease, to maintain LV systolic function at a stably impaired level (in rare cases normal systolic function returns), and, if possible, to ensure physical capacity at a level that allows the patient to function normally. Observational data from before the era of pharmacological treatment of HF indicate that a significant clinical improvement occurred in fewer than 20% of patients, while 77% died within 2 years of diagnosis, mainly due to progressive HF [12, 13]. Over the past few decades, following the introduction of modern pharmacological treatment, cardiac implantable electronic devices, and advanced cardiac surgery techniques, including ventricular support and heart transplantation, the prognosis in DCM patients has significantly improved. In the cohort of patients with DCM included in the study in the years 1982–1989, i.e., before the era of drug treatment, the 5- and 10-year survival rates were 61% and 35%, respectively [14]. The introduction of classic treatment with angiotensin-converting enzyme inhibitors (ACEis) and beta-blockers (LBAs) allowed for an improvement in 5- and 10-year survival rates to 81% and 65%, respectively [15]. Further progress in pharmacological treatment and the results of randomized trials allowed an increase in transplant-free survival at 1, 2, and 4 years of follow-up in

94%, 92%, and 88% of patients, respectively [16]. During the same period, hospitalization-free survival for HF was 88%, 82%, and 78%, respectively [8, 9]. Despite these advances, DCM treatment is still associated with a significant risk of mortality, higher than in the case of ischemic HF or valvular disease. Advanced HF remains the most common cause of death in DCM, while arrhythmic death is responsible for more than 30% [17,18].

Patients with DCM often report limitations in quality of life and psychological well-being. Differences in emotional distress and perceived limitations due to the disease are not dependent on demographic or clinical characteristics, suggesting that the limitations can only be partially explained by the symptoms and severity of the underlying disease. It was also emphasized that poor adjustment to CM was not related to quality of life but predicted poor physical performance, mental health, and emotional distress. This suggests that the Adjustment Scale may prove useful as a screening tool to identify patients whose deterioration in quality of life and emotional distress are greater than would be expected from cardiac measurements [19].

Hypertrophic cardiomyopathy

The development of HCM is caused by a mutation of sarcomere proteins, causing uncontrolled hypertrophy of the muscle, mainly of the LV, along with a disturbance in the spatial organization of cardiomyocytes and muscle fiber energetics. The prognosis in the group of patients with HCM varies, as in about 40% of patients, the presence of HCM does not directly affect the prognosis, while in the remaining patients, it significantly worsens it [20]. The main causes of poor prognosis include progression to severe HF with ventricular dilation and impaired systolic function (HCM) and SCD due to dangerous ventricular arrhythmias [21].

In more than 30% of patients, resting LV outflow obstruction can be observed, while provocation tests double this group [22]. In this group of patients, the severity of HF symptoms and prognosis depend mainly on the degree of obstruction. This is evidenced by data showing that a gradient of ≥ 30 mm Hg at rest was an independent predictor of HF progression and increased mortality. Population studies of patients with left ventricular outflow tract obstruction (LVOTO) show that progression to functional class New York Heart Association (NYHA) III–IV occurs at an annual rate of 3.2%–7.4%, depending on the degree of narrowing. As a result, severe HF develops in about 30% of patients within 6 years [23].

Intraventricular obstruction occurs in approximately 10% of patients. These patients have severe symptoms of HF and worse prognosis. Similarly, severe diastolic dysfunction can be found in approximately 9% of patients, usually in cases of severe myocardial hypertrophy and severe fibrosis, with or without LV outflow obstruction.

In patients without obstruction in LVOT, the disease usually has a mild and stable course, and most remain free of HF or have mild symptoms due to diastolic dysfunction. However, in 7%–10% of patients with non-obstructive HCM, the disease may progress through the development of systolic HF with extensive fibrosis (Figure 3). As a consequence, about 3% of patients develop severe HF with significant risk of death [24, 25].

Sudden cardiac death is the most devastating complication of HCM; it is the most common cause of death in HCM and often affects young and often asymptomatic patients. The annual incidence of SCD is <1%, but there are subgroups with a much higher risk in the general HCM population. The most common cause of SCD is ventricular fibrillation, which can be preceded by ventricular tachycardia, rapid atrial fibrillation (AF), or accelerated atrioventricular conduction [26].

The quality of life in HCM patients remains moderately reduced and mainly depends on HF symptoms. No differences were found between patients with LV outflow stenosis and no restriction with similar clinical symptoms. Similarly, patients with implantable cardioverter-defibrillators (ICD) did not report a lower level of quality of life than patients without an implanted device despite their fear of ICD shock [27].

Arrhythmogenic right ventricular cardiomyopathy

The natural history of ARVC is mainly associated with electrical instability, which can lead to arrhythmic sudden death, especially in young athletes. In the later stages of the disease, progressive RV impairment and LV involvement can lead to right and/or LV failure. The total mortality rate estimated in these studies ranges from 0.08% to 3.6% per year [28]. In population-based studies that provide real-world data, the annual mortality rate is <1%. Risk stratification remains a major clinical challenge, and antiarrhythmic drugs, transcatheter ablation, and implantable cardioverter-defibrillators are the currently available therapeutic tools. Disqualification from competitive and high-intensity sports prevents cases of sudden death, as exertion can cause not only electrical instability but also lead to disease progression [29].

ARVC patients report a lower quality of life compared to the rest of the healthy population; however, they report a better quality of life compared to patients with other cardiac diseases. Younger patients with ARVC, women, and those who have experienced at least one implantable ICD discharge are at risk of developing psychosocial problems, including poorer quality of life [30].

Restrictive cardiomyopathy

A characteristic feature of RCM is reduced compliance of the myocardial walls, impairing ventricular filling. In primary RCM, abnormal ventricular stiffness is attributed to increased calcium sensitivity, increased collagen deposition, and mutant protein aggregates such as desmin or filamin

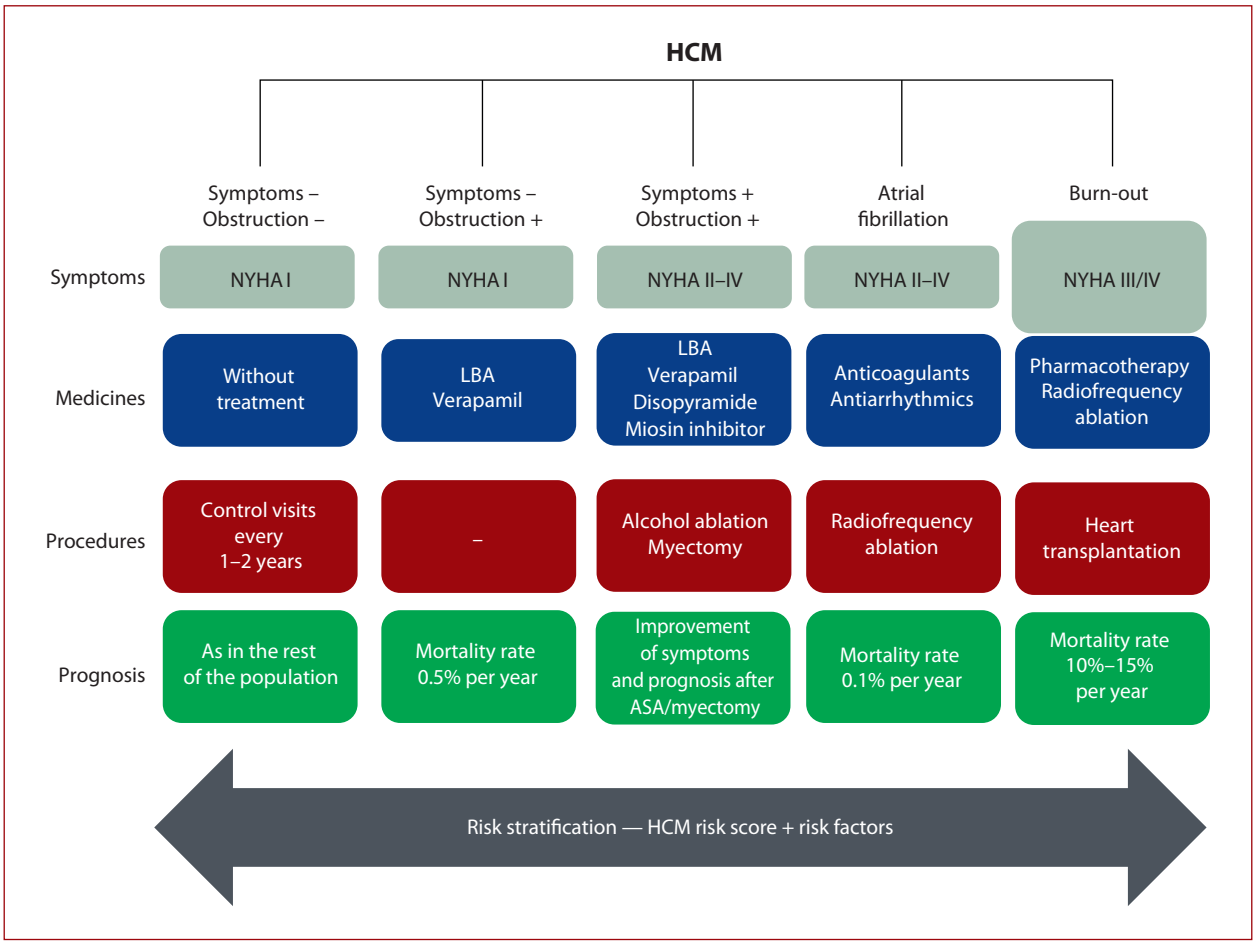


Figure 3. Summary of hypertrophic cardiomyopathy (HCM) subtypes with recommended treatment regimen and prognosis assessment
Abbreviations: LBA, beta-blockers; other — see Figure 2

C [31]. The prognosis of HF in RCM is poor, regardless of the cause of the disease. The main mechanism limiting survival is severe HF, refractory to standard pharmacological treatment. Life-threatening ventricular arrhythmias, bleeding, and thromboembolic complications may also occur [32]. It should be remembered that RCM is often the result of systemic infiltrative or storage diseases that can lead to renal failure or neuropathy. In adult RCM patients, the 5-year survival rate was 56%, and the main cause of death was HF [33].

The quality of life in patients with restrictive CM is significantly reduced and is strongly associated with measures reflecting physical capacity and daily activity level, but not with an increase in N-terminal pro B-type natriuretic peptide levels or with previous hospitalization due to HF. This is worst in young, obese, and diabetic patients [33].

The latest 2023 guidelines introduced the term "non-dilated LV CM" (NDLVC), which includes patients whose conditions were previously classified as DCM without ventricular dilation, arrhythmogenic DCM (without criteria for ARVC), arrhythmogenic LV CM (ALVC), and left-dominant ARVC. There are no epidemiological data for NDLVC (previously, patients were classified as DCM or ARVC). As it is a new phenotypic category, genes have not been evaluated in terms of their relationship to this phenotype.

Recommendations for conduct are also based mainly on the commonalities of guidelines for DCM and ARVC [1].

EPIDEMIOLOGY

Cardiomyopathies, in many cases, are heart diseases with an identifiable or suspected genetic cause, with variable and often incomplete expression throughout life. The geographical distribution of genetic variants affects the estimated prevalence in different populations, ethnic groups, regions, and countries [34]. It should be emphasized that reliable data on the incidence of CM, collected on the basis of established diagnostic criteria, come mainly from developed countries.

Table 2 summarizes the incidence of CMs classified according to the International Classification of Diseases (ICD-10): HCM, DCM, ARVC, and RCM. Data from large European registries report the relative incidence of RCM as a rare CM, occurring 26 times less often than HCM [7]. Currently, there are no reliable data on the epidemiology of NDLVC.

Based on a pooled analysis of eight studies, the prevalence of HCM is estimated to be 0.2% (95% confidence interval [CI], 1.44–2.71) or 1/460 of the population. Interestingly, data from the analysis of cardiac magnetic resonance imaging in a large cohort of adults (> 45 years) suggest that the incidence may be much higher than the results from

Table 2. Prevalence of cardiomyopathy

Cardiomyopathy	Prevalence	
	Adults	Children
HCM	0.2%	0.029%
DCM	0.036%–0.4%	0.026%
ARVC	0.078%	Very rare
RCM	Rare	0.0003%
NDLVC	No data	No data

Abbreviations: see Table 1

early studies based on echocardiographic studies and may be as high as 1.4% [1].

The incidence of DCM has traditionally been estimated to be 0.036% (95% CI, 0.023–0.050). However, the latest ESC guidelines emphasize a much higher frequency (up to 10 times) of DCM diagnosis with less stringent diagnostic criteria.

The prevalence of ARVC based on a pooled analysis of 3 relatively large studies is estimated to be 0.078% (95% CI, 0.077–0.078) or 1/1290. It should be noted that there is a lack of robust and reliable epidemiological data, partly due to the complexity of the diagnostic process [1].

Taking into account the data presented earlier on the prevalence of CM, the estimated number of cases of CM in Poland should be, in the case of DCM, about 75 000–150 000 patients (1:250–500), similarly HCM about 75 000–150 000 (1:250–500), ARVC about 7600–19 000 patients (1:2000–5000). Deepening the diagnostics, especially genetic testing, may contribute to the better classification of this patient population.

It is difficult to compare these data to real information on the occurrence of CMs in Poland and patient care. Epidemiological studies in Poland are scarce. A report prepared as part of the project "Maps of health needs in the field of cardiology" used data from the National Health Fund registered in 2016–2020. In the analysis of this report, it was necessary to use certain approximations and estimate epidemiological indicators: registered prevalence (the number of currently living patients diagnosed with CM) and registered incidence (the number of new cases). According to the Report, the registered prevalence was respectively: 286/100 thousand in 2016 (i.e., about 120 thousand patients), 2020 — 271/100 000 in 2020 (65% of the population — men). The registered incidence was 43.7/100 000 in 2016 and only 17.6/100 000 in 2020, which is most likely due to diagnostic limitations related to the COVID-19 pandemic [34, 35]. Importantly, according to data recorded by the National Health Fund, the average age of diagnosis of CM in Poland in 2016–2020 was 60 years.

The limitations of screening and diagnostic activities in Poland are evidenced by the results of analyses of patients registered with HCM. It is worth analyzing these data in relation to analogous literature data — the prevalence of HCM in Poland was as follows: 2016 — 35/100 000, in 2021 — 36/100 000, which is about 20% of the estimated number of patients in Poland (1:500) [35]. According to publications presenting data registered in other countries — in the United States, the prevalence of HCM was 52/100 000 in 2013, in 2019: 74/100 000 [36]; in the United Kingdom 48/100 000 [8]; in China: 76/100 000 population [37]. The cited results also differ from the prevalence data, but they are higher than in Poland.

AVAILABILITY OF SPECIALIST CARDIAC CARE — THE PATHWAY OF THE PATIENT WITH CARDIOMYOPATHY IN POLAND

Both the diagnosis of CM and its treatment require specialized cardiac care. The report on CM developed as part of the project "Maps of health needs in the field of cardiology" allows for an estimated analysis of the course of treatment of patients with this disease in Poland.

An analysis of the so-called "patient pathways" in the healthcare system in Poland in 2016–2021 (65 383 registered patients with ICD-10 for CM; 65.4% men) showed that the diagnosis/first registration in the National Health Fund system with the ICD-10 code for CM takes place in hospital conditions in 93.4% of patients. This is beyond doubt due to the diagnostic requirements of this group of diseases. However, the hospitalization mode is noteworthy – CM diagnosis is made in 68.2% of patients during acute hospitalization, and only in 25.1% during scheduled hospitalization [38]. Undoubtedly, diagnosis of CM at the exacerbation stage is too late.

The further fate of the above-mentioned group in the healthcare system is even more worrying. Between years 2016–2021, among patients hospitalized acutely 23% died and another 28% did not reappear in the health care system with a code corresponding to the diagnosis. Similar data for patients diagnosed during scheduled hospitalization were as follows: 18% of patients died, and as many as 44% of patients did not reappear in the healthcare system. After hospitalization and diagnosis, only 15.28% of patients were referred for specialized cardiac care [38].

Another limitation of care for CM patients in Poland is the lack of access to genetic testing, which is currently a very important component of CM diagnostics and screening of families of CM patients .

These data registered by the National Health Fund system have their numerous limitations, but they indicate the following fact — in the field of screening, diagnostic tests, and care for CM patients in our country, we have a number of limitations, which require effective actions to improve the quality of care. These data show how we deviate from the optimal model, which is the treatment of patients with CMs in specialist centers providing appropriate hospital and outpatient facilities closely cooperating with each other.

THERAPEUTIC MANAGEMENT

The management of CMs is strictly determined by the type of CM and its predominant clinical picture. In order

to make a diagnosis, the presented symptoms, the results of imaging and genetic tests, morphological, functional, and often histological assessment (biopsy of cardiac tissue) are important. Diagnosis of a specific CM, including differentiation of CM from phenotypic images occurring in metabolic or storage diseases, allows for personalized and dedicated clinical management.

The general principles of CM management include symptom control, ensuring life comfort for patients, inhibiting disease progression, and preventing possible complications, e.g., life-threatening arrhythmias. Patients require constant care and regular follow-up — clinical assessment and optimization of management is recommended every 1 or 2 years or after each episode changing the current clinical course of the disease [1].

CM patients require specific management. A systematic approach to therapy goals takes into account several key tasks, which are listed below [1]:

A. *Lifestyle modification and rejection of habits closely correlated with an increased risk of life-threatening arrhythmias, development, or exacerbation of heart failure symptoms*

B. *Assessment of the SCD risk and indications for implantation of high-energy devices*

An important issue in all phenotypes of CM are indications for the implantation of high-energy devices in both primary and secondary prevention of SCD. The qualification process for implantation of an ICD should be preceded by informing the patient thoroughly about the implantation method, as well as benefits and consequences of having the device. Moreover, a clinical assessment of possible complications resulting from the possibility of e.g., inadequate discharges/interventions or therapy complications in long-term follow-up, should also be presented to the patient [1].

Primary prevention of SCD

In the primary prevention of SCD, the indications for implantation of high-energy devices are different for different CM phenotypes. SCD risk assessment is carried out using calculators dedicated to individual CMs. In the case of high risk, indications for implantation are considered. In the absence of indications for implantation, a reassessment of SCD risk is required every 1 or 2 years in each CM patient; accelerated assessment might be necessary depending on changes in clinical status.

Risk calculators take into account various parameters typical of individual CM.

In HCM, the HCM-RISK calculator based on age, unexplained syncope in history, LV outflow pressure gradient, maximum LV wall thickness, left atrial size, presence of NSVT or SCD in the family history, classifies patients into the following groups:

- very high risk of SCD over a 5-year period (estimated at >6%),
- intermediate risk (4%–5%),
- or low risk <4%.

In addition, the personalized decision to implant an ICD also takes into account the systolic activity of the LV and the extent of the scar in the heart muscle.

ICD implantation should be considered in HCM patients with estimated high risk and can be considered in patients with intermediate risk while taking into account possible complications and the impact of the device on various aspects of the patient's life. In the case of low risk, SCD implantation may also be considered in primary prevention, when ejection fraction (EF) is at least <50% or there is extensive late gadolinium enhancement on cardiac magnetic resonance (CMR) >15%. However, there are no data on the effect of quantitative scar assessment or EF on risk estimates obtained using the HCM Risk-SCD calculator. Patients with DCM and NDLVC are somewhat different; in them the decision to implant an ICD is also made on the basis of genetic risk factors for SCD (Table 3). In this population, eligibility for ICD therapy in primary prevention of SCD is determined by the standard value of LVEF (applies to patients with LVEF ≤35% despite at least 3 months of optimal pharmacotherapy) and/or specific genotype. Genotype, as well as the presence of late gadolinium enhancement on CMR imaging and the presence of ventricular arrhythmias, are of particular importance in the decision to implant an ICD in patients with LVEF >35%. Implantation of the device in DCM patients should also be

Table 3. Genes associated with a high risk of sudden cardiac death in patients with DCM and NDLVC

Gene	SCD frequency/year	Risk factors for SCD
LMNA	5%–10%	Estimation of the 5-year risk of life-threatening arrhythmias LMNA risk score (https://lmna-risk-vta.fr)
FLNC	5%–10%	LVEF <45%, LGE on CMR
TMEM43	5%–10%	Sex: Male Female + one of the following: LVEF<45%, presence of uncorrected ventricular tachycardia (nsVT), >200 extra excitations/day on Holter-ECG, LGE on CMR
PLN	3%–5%	Estimation of the 5-year risk of life-threatening arrhythmias PLN risk score b (https://plnriskcalculator. shinyapps.io/final_shiny) LVEF <45%, LGE on CMR, NSVT
DSP	3%–5%	LVEF <45%, LGE on CMR, NSVT
RBM20	3%–5%	LVEF <45%, LGE on CMR, NSVT

Abbreviations: CMR, cardiac magnetic resonance; DSP, desmoplakin; FLNC, filamin C; LGE, late gadolinium enhancement; LMNA, lamina A/C; LVEF, left ventricular ejection fraction; NSVT, non-fixed ventricular tachycardia; PLN, phospholamban, RBM20, RNA-binding protein 20; TMEM43, transmembrane protein 43

considered in those with genetic risk factors and LVEF >35% and observed arrhythmias (VT, VEB, syncope). In selected cases, it can also be considered in patients with LVEF >35% without clinical signs of arrhythmia (nsVT, VEB, syncope), with the presence of genetic risk factors and, conversely, in patients without a risky genotype, but with episodes of dangerous ventricular arrhythmia [1].

The risk calculator in ARVC (arvcrisk.com) is characterized by high prognostic value in patients with pathogenic variants of genes encoding desmosomal proteins, especially plakophilin-2. In the case of association with pathogenic variants of other genes or in patients without pathogenic variants (gene-elusive), as well as in the case of concomitant significant LV damage, its value is lower. In such situations, individual risk stratification is recommended.

The most commonly used form of prevention is percutaneous implantation of a classic, transvenous ICD. In patients who do not require constant cardiac stimulation or cardiac resynchronization therapy, implantation of a subcutaneous ICD (sICD) is an attractive therapeutic option due to the reduction of the risk of complications of standard ICD therapy. At present, substernal ICDs (EV-ICDs) can be considered. EV-ICDs cannot be used in patients who have undergone sternotomy or have severe chest deformities. Wearable cardioverter-defibrillators, on the other hand, are indicated temporarily in patients waiting for a decision on ICD implantation (e.g., in the initial period of CM treatment) or waiting for ICD reimplantation (e.g., after removal of the system due to complications).

Secondary prevention of SCD

Indications for ICD implantation in the secondary prevention of SCD in CM patients are the same as in other cardiovascular diseases. In patients after sudden cardiac arrest caused by ventricular fibrillation or hemodynamically unstable ventricular tachycardia (or in patients in whom ventricular arrhythmia causes syncope and the origin of these events is not reversible), implantation of an ICD or a cardiac resynchronization therapy-defibrillator is recommended, regardless of the CM phenotype. In the case of ARVC, secondary prevention also includes the occurrence of long-term ventricular tachycardia without hemodynamic symptoms.

C. Treatment of cardiac arrhythmias

Treatment of ventricular arrhythmias

CM patients often require antiarrhythmic drugs or ablation of the arrhythmia substrate in the case of ventricular arrhythmias. The selection of pharmacotherapy should consider the type of CM and presence of cardiac systolic dysfunction.

Arrhythmia substrate ablation procedures, especially recurrent ventricular tachycardia, should be performed in experienced centers. This is due to the specific technical problems associated with such treatments: e.g., epicardial position of the arrhythmia substrate in ARVC, often intramuscular or epicardial arrhythmia substrate in DCM. In addition to the technical aspect of the procedure, patients with CMs undergoing ablation often require specialist cardiac treatment, which is available at tertiary centers. The need to consider performing a procedure using both endocardial and epicardial ablative access is increasingly emphasized. In addition to selected cases of DCM patients, good long-term results of VT ablation reported in the population of ARVC patients are noteworthy. They are significantly better than in the case of endocardial ablation only. Procedure planning based on algorithms analyzing the results of MRI and CT imaging can also increase the effectiveness of ablation.

Treatment of supraventricular arrhythmias and prevention of thromboembolic complications

The most common supraventricular arrhythmias are AF and atrial flutter. An aggressive rhythm control strategy based on transcatheter ablation improves overall survival of patients with HF and reduced LVEF, improves echocardiographic indicators, lowers biomarker concentrations, and improves patient quality of life [39, 40]. On the other hand, a special group of patients are patients with HF and impaired systolic function of the LV, where catheter ablation, due to its much higher effectiveness than antiarrhythmic pharmacotherapy, is recommended already in the first line of treatment. It should be remembered that performing AF catheter ablation does not exclude the use of optimal HF pharmacotherapy. It should be remembered that the diagnosis **of AF/atrial flutter will determine the decision to use anticoagulation**, which differs in different types of CM.

In the case of AF, in patients with HCM and amyloidosis, anticoagulant therapy **is obligatory**, regardless of the CHA_2DS_2-VA score. In RCM, its use **can also be considered** regardless of the CHA_2DS_2-VASc score. In other types of CM (DCM, NDLVC, or ARVC), anticoagulation should be used with CHA_2DS_2-VA ≥2 in men and ≥3 in women (can be considered **with a score of 1 for men and 2 for women**).

D. Comprehensive HF treatment

In the course of CM, symptoms of HF are common; they include HF with reduced EF, slightly reduced EF, or preserved EF. The HF type is associated with the type of CM and its severity, as well as comorbidities. The effects of "classic" HF pharmacotherapy depend on the CM type — they have a documented significance in DCM. Other types of CM require adequate modification.

It should be remembered that using diuretics, mineralocorticoid receptor antagonists, or ACEi/angiotensin receptor antagonists and neprilysin inhibitors, or LBAs may manifest as worse well-being of patients due to hypotension and dehydration. These medications require careful dosing, sometimes de-escalation, and often also discontinuation of treatment.

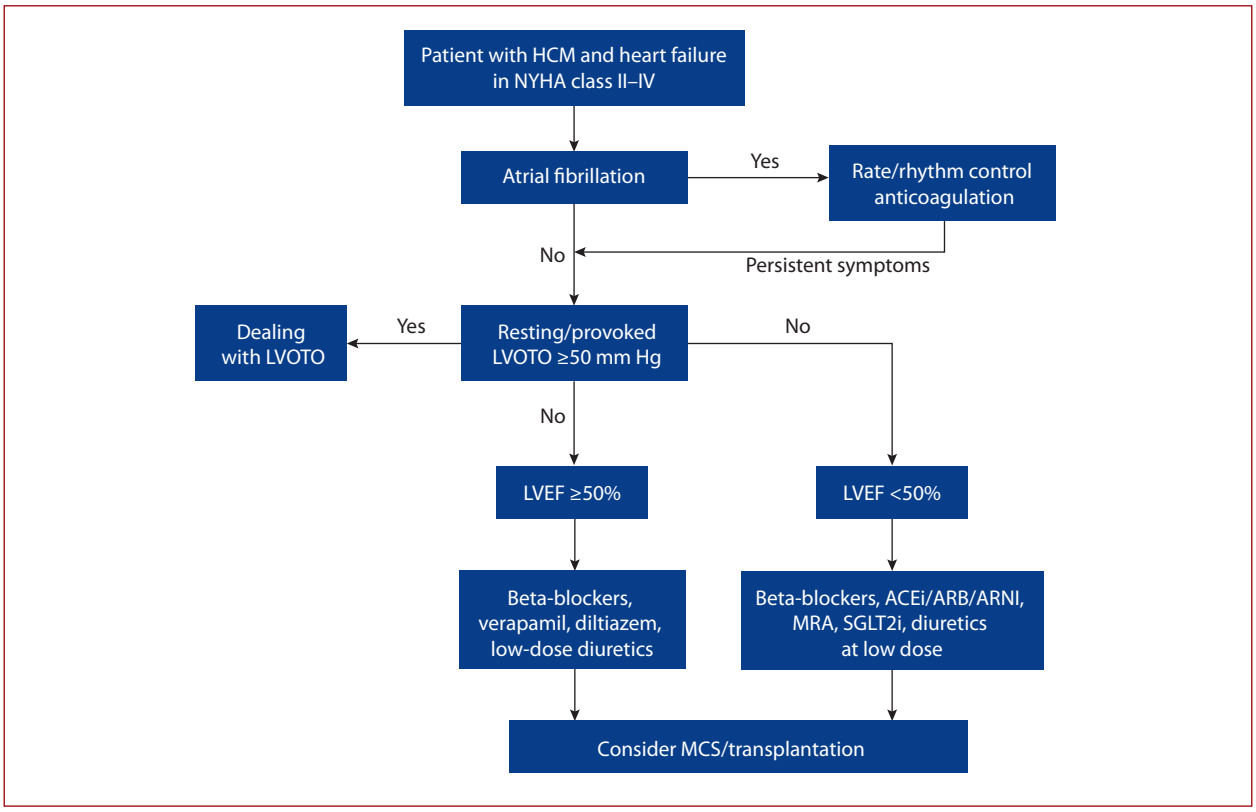


Figure 4. Treatment regimen for heart failure in patients with hypertrophic cardiomyopathy

Abbreviations: ACEi, angiotensin-converting enzyme inhibitor; ARB, angiotensin receptor antagonist, ARNI, angiotensin receptor antagonist and neprilysin inhibitor; LVEF, left ventricular ejection fraction; LVOTO, left ventricular outflow tract restriction; MCS, mechanical circulatory support; MRA, mineralocorticoid receptor antagonist; SGLT2i, sodium-glucose cotransporter type 2 inhibitor; other — see Figure 2

A treatment option for selected patients is implantation of cardiac resynchronization therapy or LV assist devices to support ventricular function. Heart transplantation is a therapeutic option for patients with CMs and advanced HF in NYHA class III–IV, or with severe, recurrent ventricular arrhythmias that do not lend non-responsive to available treatment.

Individual CMs also require specific and flexible procedures modifying the general algorithm (presented earlier) [1]. Most of them concern HCM, as there is the most evidence in this area.

Management of hypertrophic cardiomyopathy

HCM management is determined by the presence and magnitude of the LV outflow tract obstruction (LVOTO) gradient and LVEF.

In patients without LVOTO, in whom LVEF reaches a value of < 50%, it is recommended to use pharmacotherapy dedicated to HF with reduced ejection fraction (LBAs, ACEi/angiotensin receptor antagonists and neprilysin inhibitors, mineralocorticoid receptor antagonists, sodium-glucose cotransporter type 2 inhibitors — flozins, and low doses of diuretic drugs). If LVEF ≥50%, LBA, verapamil or diltiazem, and low doses of diuretics are recommended. Therefore, the therapy is intended to improve clinical symptoms associated with systolic and diastolic dysfunction, which often coexist with disturbed inflow into the left ventricle, and to prevent the symptoms of angina. Symptoms suggestive of angina, after exclusion of significant stenosis in the coronary vessels and exclusion of LVOTO, can be alleviated by adding LBAs, calcium channel blockers, and nitrates (only in the group of patients WITHOUT LVOTO). Patients who do not respond to pharmacotherapy are potential candidates for heart transplantation (Figure 4).

In the presence of LVOTO (challenged or resting) >50 mm Hg, LBAs should be used, and if symptoms persist, calcium channel blockers (diltiazem or verapamil) should be used in gradually increased doses to maximum values. The ESC guidelines also include disopyramide in pharmacotherapy (available in Poland only as part of targeted import). The biggest novelty in the current guidelines is adding mavacamten (a cardiac myosin inhibitor) to therapy, which should be used alongside LBA or calcium channel blockers when symptoms persist. The drug can also be used as monotherapy in patients with contraindications to standard pharmacotherapy. Gradient-increasing drugs such as nitrates, digoxin, or phosphodiesterase 5 inhibitors should not be used in LVOTO patients (Figure 5).

There is still limited evidence for patients with LVOTO between 30 and 50 mm Hg.

If pharmacological management is ineffective, when LVOTO >50 mm Hg and clinical symptoms indicate NYHA

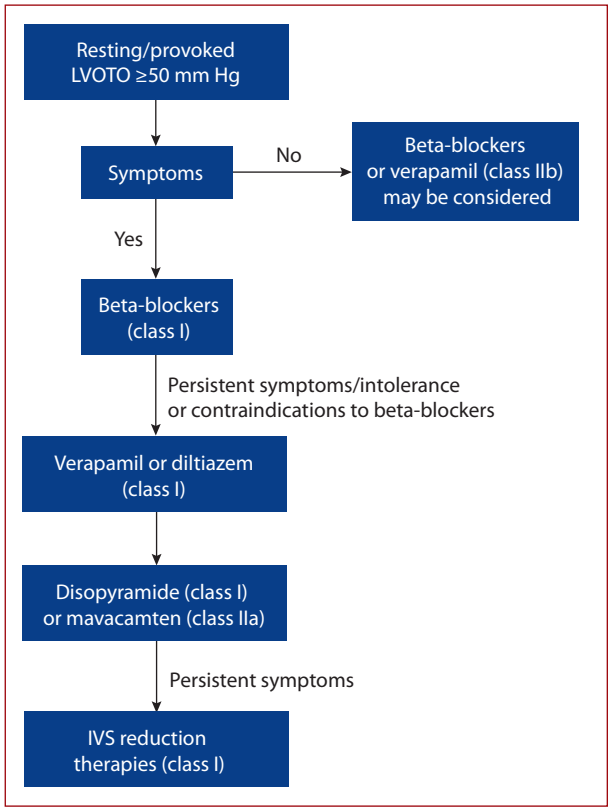


Figure 5. Management of hemodynamically significant LVOTO narrowing in patients with hypertrophic cardiomyopathy

Abbreviations: IVS, intraventricular septum, LVOTO, left ventricular outflow tract

class III or IV, there is still the option of using the septal thickness reduction procedure, i.e. alcohol ablation or surgical ventricular septal myectomy, which gives slightly better long-term effects. Such treatment may also be considered in people with NYHA II and additionally moderate/severe mitral regurgitation, AF, or left atrial dilation.

In patients with LVOTO > 50 mm Hg qualified for septal thickness reduction procedure, who have moderate/severe mitral regurgitation, surgical repair of the subvalvular apparatus or valve replacement should be considered, as well as valve repair when it becomes regurgitated as a result of myectomy. In such cases, when the symptoms are accompanied by AF, surgical ablation and closure of the left atrial appendage may be considered.

The qualification scheme for ICD implantation in HCM patients does not differ from the general scheme presented earlier, but the qualification in primary prevention is based on the stratification of the risk of SCD with the use of an HCM-specific calculator. The current ESC guidelines provide data on the possibility of estimating the risk of SCD in children and adolescents (HCM-RISK).

In conclusion, it should be emphasized that in the clinical management scheme in CMs, the primary goal is to ensure the life comfort of patients, control their clinical symptoms, inhibit further progression of heart disease, and prevent life-threatening arrhythmias. Clinical evaluation of patients is recommended every 1 or 2 years or after each episode changing the current clinical course of the disease. In addition, it should be borne in mind that the documented modulators of the clinical course in HCM are hypertension, diabetes, and obesity; in DCM, the modulators include viral infections, hypertension, toxic factors, and pregnancy; in ARVC, they are sports and viral background.

PATIENT CARE — THE 2023 ESC RECOMMENDATIONS AND EXAMPLES OF SYSTEMIC SOLUTIONS FROM OTHER EUROPEAN COUNTRIES

In order to optimize the diagnosis and treatment of CM patients, individualized and at the same time expert, systematic, coordinated, often multidisciplinary patient care is necessary. It should be remembered that it is necessary to take care not only of the patient but also of his/her family because this information is very important for all of them. Hence, multidisciplinary teams for CM should be created. It is worth noting that CMs manifest in various clinical situations. In some patients, they are the first and irreversible manifestation of the disease (sudden death), sometimes they are detected by accident, or their symptoms appear gradually as the disease progresses. CM manifestations include not only cardiovascular but also a whole range of non-cardiac symptoms (e.g. neurological, neuromuscular, ophthalmological, nephrological). CMs, in a large proportion of cases, are genetic diseases and can manifest at different ages, so appropriate pediatric and adult care is needed. To sum up, it is necessary to provide multidisciplinary care, and the team's composition will depend on the patient's clinical profile [1].

Therefore, the optimal multidisciplinary team should include (Figure 6) [1]:

- adult and/or pediatric cardiologists/cardiac surgeons, especially those who specialize in cardiogenic conditions;
- clinical geneticists to analyze and interpret the results of genetic tests [41];
- cardiac imaging radiologists, including CMR experts;
- specialist teams focused on multidirectional analysis of endomyocardial biopsy;
- teams of nurses/psychologists and/or genetic counselors.

Telemedicine care solutions should be an integral component of CM patient care. Patient associations also play an important role in patient care. This is complemented by national/international networks, such as the European Reference Network for Rare and Low-Frequency Complex Heart Diseases (ERN GUARD-Heart) (https://guardheart.ern-net.eu), which enable the exchange of information relevant to CM patients. The "cardiomyopathies matter" project has made a significant contribution to the dissemination of CM findings at the European level, providing

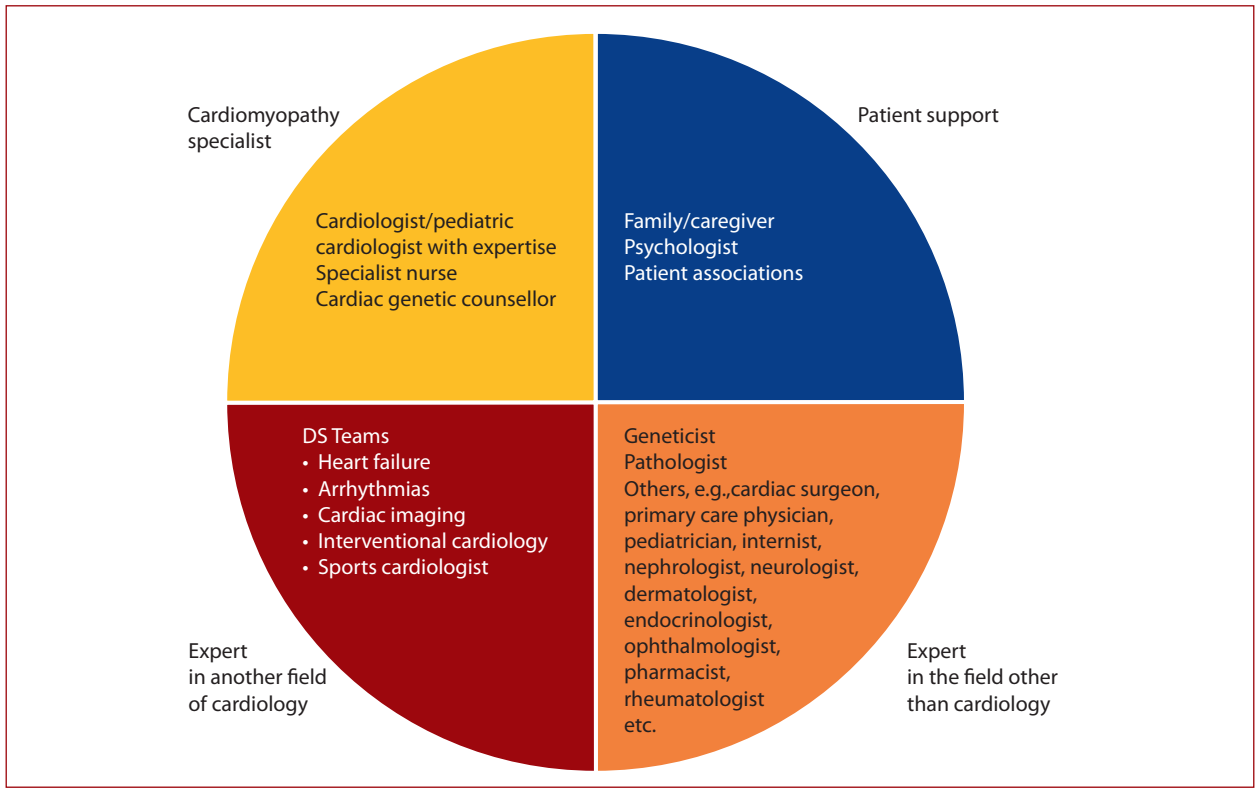


Figure 6. A multidisciplinary team providing comprehensive care for a patient with cardiomyopathy

a valuable source of data on CM and other CM-related topics that are constantly updated (https://cardiomyopathiesmatter.org/).

Example of systemic solutions for CM patient care in Europe: France

In 2014, the French Ministry of Health established the national reference center Cardiogen and a national reference network for rare and hereditary heart diseases, including CM (www.filiere-cardiogen.fr) [42].

The network including 64 accredited centres throughout France is financed by the Ministry of Health and has 21 centers dedicated exclusively to CM patients. Moreover, Cardiogen

- presents current recommendations and provides educational materials;
- develops clinical trials and creates useful databases;
- facilitates communication between centers/doctors/medical staff/patients about rare and hereditary heart diseases;
- provides funding (e.g., for specialist nurses, genetic testing, and genetic counseling).

The implementation of this project has improved the level of care and standard treatment of CM patients, while providing all interested parties (patients/healthcare professionals) with free access to:

- educational materials for both patients and healthcare professionals;
- a database that improves the referral system, enabling the collection of data from reference and specialist centers in dedicated patient medical records;
- contact details of relevant healthcare professionals, facilities, and patient organizations;
- a psychological resource center facilitating patients' access to psychological support;
- a free monthly online consultation that allows patients and families to provide feedback about living with rare and inherited heart diseases.

An example of systemic solutions for the care of patients with cardiomyopathies in Europe: Spain

The Ministry of Health of Spain, after a thorough assessment, granted accreditation to centers, service providers, and reference units (CSUR Spanish — Reference Centers, Units, and Services) [43]. Accredited centers maintain appropriate standards, including those relating to patients' rights, and implement quality assurance programs or annual audit plans. CSUR centers have extensive knowledge and experience in managing a specific group of heart diseases. At the same time, they have appropriate equipment and staff necessary to provide patients with high-quality care. Full details available on the website: https://www.sanidad.gob.es/profesionales/CentrosDeReferencia/CentrosCSUR.htm [43].

There are currently seven CSUR centers in Spain in the field of hereditary heart diseases.

As part of its duties, CSUR:

- provides care and services for the entire territory of the country on equal terms, regardless of patients' place of residence;
- offers support from a multidisciplinary team such as healthcare and clinical observation, confirming diagnosis, and defining treatment strategies;
- ensures continuity of care at different stages of the patient's life and levels of healthcare;
- assesses the effects of treatment;
- offers consultations for departments normally dealing with this group of patients;
- provides training for other health professionals.

SUMMARY

Cardiomyopathies are a variety of myocardial diseases that require specialized diagnosis and care. Both genetic testing, modern stratification of the SCD risk, pharmacotherapy, and invasive treatment create new opportunities to optimize the management of CM patients. We do not provide all of these options to patients in our country. It seems necessary to take action to develop a diagnostic and therapeutic model that will allow for earlier detection of CMs in Poland and their effective treatment. There is a need to create a strategy to promote active, coordinated care for these patients and screening for their family members. The implementation of these preventive methods will slowly improve the prognosis in this population.

Article information

Conflict of interest: None declared.

Funding: None.

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