# Living with cardiomyopathy: Patients' pathways and unmet needs

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***Related article***
by Mizia-Stec et al.

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Cardiomyopathies (CMs) are a heterogeneous group of pathologies characterized by structural and functional alterations of the heart [1]. Recent studies suggest that traditional classification methods fall short of meeting the precise needs of contemporary managing CMs [2]. These conditions are currently described from the perspective of morphology, function, and genomics in hypertrophic cardiomyopathy, dilated cardiomyopathy, restrictive cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy, left ventricular noncompaction, and partially acquired cardiomyopathy [3]. However, such broad categorization fails to capture the intricate variations and complexities inherent in CMs. In the last decade, advances in diagnostic techniques have enabled to move from a mechanistic to a phenotypic and then etiological approach in the work-up of CM patients. As a consequence, several new therapies that interact with the pathophysiological pathways of CMs, either by targeting the phenotype or by targeting the etiology, are now available [4]. For example, mavacamten can be used in sarcomeric hypertrophic CM, tafamidis in transthyretin cardiac amyloidosis, migalastat in Fabry disease, and myosin stimulators in dilated CM.

Although CMs are generally considered to be rare, their burden is substantial and occurs worldwide, across all ages, sexes, and ethnic groups. Data from the Global Burden of Disease databases suggest that the prevalence of CMs is increasing, driven by a combination of improved awareness, evolution of diagnostic pathways, and effective treatment options [5, 6]. However, there is growing evidence that the management of CMs is subject to regional variations and that diagnostic pathways and therapeutic advances have largely benefited patients in countries that are highly developed medically and scientifically.

This crucial issue has recently been addressed in an outstanding study published by Mizia-Stec et al in the Journal [7]. The authors aimed to investigate the management process of CMs in Poland in an attempt to identify the outcome for patients following the initial diagnosis of the condition. To this end, Mizia-Stec et al. [7] carried out a population-based cross-sectional study on data from the national healthcare provider sample acquired from 2016 to 2021 using ICD-10 codes to identify patients with CMs. Data from a total of 65 383 CM patients with CMs. Data from a total of 65 383 patients diagnosed as having CM between 2016 and 2021 were analyzed. Results showed that the diagnosis of CMs in Poland is made very late, mainly after referral to a tertiary hospital due to exacerbation of the disease and/or comorbidities. Furthermore, the mortality rate of CM patients was very high, particularly in patients who entered the health system only once (47% of all subjects) and in patients with the Charlson Comorbidity Index ≥5. In summary, the study underscores the unmet needs of CM management in Poland, by highlighting the suboptimal care of CM patients and the need for a broader availability of tertiary reference outpatient care facilities.

The authors of this study should be acknowledged for addressing a timely and relevant novel issue that was poorly investigated

in the past, as for the first time data have been provided on the treatment pathways defined as the sequence of visits to the public health system of thousands of CMs patients in Poland.

Mizia-Stec et al. [7] should be commended also for raising several important points that might interest all clinical cardiologists, regardless of their country of origin.

The first issue addressed by Mizia-Stec et al. [7] refers to the fact that CMs are still under-recognized in real-world clinical practice. As a matter of fact, many patients are still diagnosed with CM only after hospital referral for new symptoms and/or for comorbidity [8, 9]. This relevant problem has clearly emerged during the recent COVID-19 epidemic, as many CM patients have shown greater reluctance to seek medical advice similar to what was observed in other cardiac patients [10, 11].

The second point raised by the authors is the fact that CM patients have a delayed diagnosis and are correctly identified only when they are elderly. As a result, these patients also suffer from several comorbidities which, in turn, worsen their outcomes. Indeed, conditions, such as hypertension, obesity, diabetes mellitus, coronary artery disease, and peripheral artery disease often contribute significantly to ominous outcomes for CM patients even when they are adequately treated [12, 13]. Also, in the case of coexistence of aortic stenosis, transthyretin CM is associated with 2-fold higher mortality when compared with aortic stenosis alone [14].

Last, but not least, the study by Mizia-Stec et al. [7] emphasizes the fact that CM patients are often under general practitioners care rather than tertiary referral centers. This is at variance with currently recommended European guidelines [1], which state that CM patients should be followed by an ad hoc cardiomyopathy team, involving specialists in genetics, cardiac imaging, electrophysiology, cardiac surgery, and interventional cardiology (Figure 1). In recent years, a consensus has emerged that the best option for the management of CM patients should be a shared decision-making approach, involving discussion of risks and benefits of each treatment strategy, and matching clinical and procedural aspects with the needs and preferences of the individual patient. Therefore, the decisional process must be carried out by an experienced team working in a dedicated cardiomyopathy center of excellence. The concept of a "Heart Team", ideally including clinical and interventional cardiologists and cardiac surgeons, has been shown to improve discussion and decision-making between interventional and surgical approaches in both coronary artery and valvular heart disease. Following the same formula, in CM patients' management, a "Cardiomyopathy Team" should be that center of excellence analyzing and interpreting diagnostic tests contextualizing them against the clinical status of the patient, and critically appraising the need for therapies [15]. Implementing the strategical role of CM teams working in experienced centers is a critical step in the management of this patient population, as the results of both pharmacologic and non-pharmacologic treatments are largely dependent on the experience of the operators/institutions.

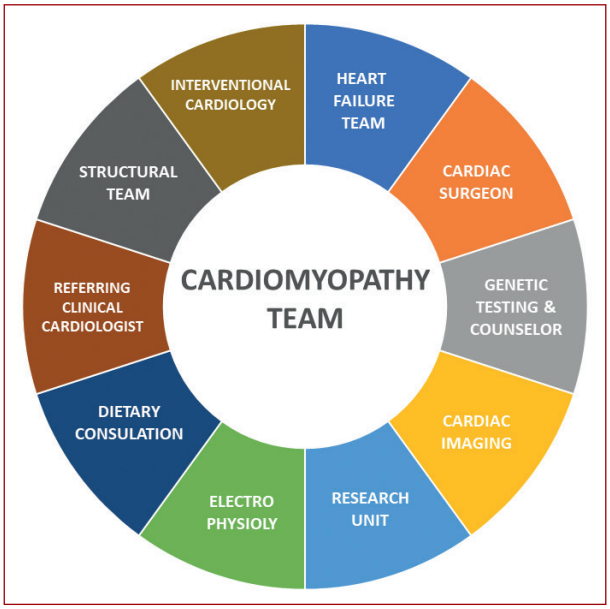


**Figure 1.** Multidisciplinary Cardiomyopathy Team

In conclusion, significant obstacles remain in instituting contemporary CM management in countries with fewer highly developed technical resources, physician expertise, or patient access to specialized care. Studies that analyze barriers to such pathways are crucial for introducing contemporary care for CM patients in the wider world.

## Article information

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