

Hypertrophic Cardiomyopathy in Non-specialized Centers: an Opportunity to Standardize Clinical Practice

Miocardopatía hipertrófica en centros no especializados: una oportunidad para ordenar la práctica real

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Hypertrophic cardiomyopathy (HCM) occupies a unique place among cardiovascular diseases. For decades, its definition was based on the presence of left ventricular hypertrophy not attributable to abnormal loading conditions. However, the current understanding of the disease is much broader: HCM constitutes a heterogeneous group of conditions with highly diverse genetic bases, pathophysiological mechanisms, clinical presentations and disease progression. This complexity explains why its management cannot be reduced to the mere echocardiographic detection of increased wall thickness, but rather requires a comprehensive clinical assessment, precise phenotypic characterization, risk stratification, family history investigation and, where appropriate, the integration of cardiovascular genetics. (1–8)

In this context, the Hypertrophic Cardiomyopathy Registry in non-specialized centers, presented by Cáceres et al. in this issue of the Argentine Journal of Cardiology represents a valuable contribution to understanding how this disease is diagnosed and managed in real-world settings. (9) The study included 160 adult patients from 8 provinces, with a mean age of 48 years and a predominance of male gender. Echocardiography was performed in 97.5% of cases, cardiac magnetic resonance imaging (CMRI) in 60%, and genetic testing in 40%. Among the patients evaluated via genetic testing, diagnostic yield was high, with a predominance of MYH7 and MYBPC3 variants; additionally, left ventricular outflow tract obstruction was observed in 34.3% of the cohort, and late gadolinium enhancement was common among those who underwent CMRI. (9)

The main strength of the study lies not only in its numbers, but in the scenario it describes. Most patients with HCM do not initially present at units

specialising in familial heart disease, but at general practices, outpatient centers or institutions without structured circuits for this condition. Therefore, understanding what happens in these settings is essential for identifying opportunities for improvement, guiding training strategies and building more effective care networks. In this regard, the registry has a unique special merit: it shifts the focus from the specialized center to the place where the patient's clinical history often begins.

The results show active cardiological practice, with high use of echocardiography and significant incorporation of CMRI and genetics. This point is worth highlighting, as it reflects that contemporary diagnostic tools are already part of the clinical practice of many cardiologists working outside specialized centers. In addition, the registry reveals that access to and the indication for these resources are not yet uniform. This observation should not be interpreted as a weakness of the study, but rather as one of its most important messages: the quality of diagnosis in HCM depends not only on individual medical knowledge, but also on the availability of tests, referral pathways and the organization of the healthcare system.

A particularly interesting aspect is the high proportion of delayed enhancement among patients assessed with CMRI and the high diagnostic yield of genetic testing in that subgroup. These findings likely reflect appropriate clinical selection of patients referred for more complex studies. They also open up an opportunity for future phases of the registry, in which it could prove highly valuable to characterize in greater detail the criteria of referral for CMRI and genetic testing, the differences between patients who underwent and those who did not undergo these tests, and the type of genetic panel used. This information would allow

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for a better understanding of how these tools are integrated into daily practice and would help to optimize their use in different clinical settings.

As with any real-world registry, the observational and retrospective design should be valued precisely for its ability to reflect standard care conditions. The inclusion of patients with a confirmed or highly probable diagnosis reflects a common situation in medical practice: not all patients have initial access to CMRI, cardiovascular genetics or assessment by specialized teams. In a condition with multiple differential diagnoses and possible phenocopies, this reality reinforces the need to continue moving towards shared diagnostic criteria, minimum assessment protocols and progressively more standardized referral circuits.

The Argentine Society of Cardiology's 2025 Argentine Consensus on the Diagnosis and Treatment of Hypertrophic Cardiomyopathy provides a particularly timely framework for interpreting the results of this registry. (7) The document proposes a comprehensive approach that includes clinical assessment, family history, electrocardiogram, echocardiography, CMRI, genetics and systematic risk stratification, always adapted to the patient's context and available resources. Furthermore, it emphasizes the need to consider differential diagnoses, phenocopies, family screening and longitudinal follow-up. (7) In this regard, the registry by Cáceres et al. shows that many of these recommendations are already in practice, albeit in a heterogeneous manner, and that the current challenge lies in transforming the available knowledge into reproducible care pathways.

Hypertrophic cardiomyopathy requires not only precision medicine but also organized medicine. It is not enough to have CMRI, genetics or advanced therapies if the patient is not identified, characterized and referred in a timely manner. Similarly, the absence of every resource in a center should not prevent an adequate initial assessment. Doppler echocardiography, family history, electrocardiography, 24-hour Holter monitoring, stress testing or exercise echocardiography, when available, remain invaluable tools for guiding diagnosis, quantifying obstruction, assessing symptoms, detecting arrhythmias and estimating risk. (5–7)

The registry also invites reflection on the very concept of a 'non-specialized center'. In Argentina, as in many other countries, specialization depends not only on patient volume, but also on the existence of multidisciplinary teams, access to advanced imaging, cardiovascular genetics, septal reduction surgery, septal ablation, electrophysiology, advanced heart failure care and family counselling. From this perspective, the solution should not be to draw a rigid line between specialized and non-specialized centers, but rather to create functional networks between the two. The clinical cardiologist occupies a central role in this network: he suspects the condition, initiates the investigation, supports the patient and decides when referral is necessary.

Recent therapeutic developments make this model of care even more important. In addition to conventional strategies, such as beta-blockers, calcium channel blockers, disopyramide, septal myectomy, septal ablation and implantable cardioverter-defibrillators, a new era marked by cardiac myosin inhibitors has emerged. Studies with mavacamten demonstrated a reduction in the obstructive gradient, symptomatic improvement and a reduced need for septal reduction therapy in selected patients with symptomatic obstructive HCM. (10–13) More recently, aficamten has expanded the field of research with significant results compared to conventional treatment. (14) This therapeutic evolution reinforces the need to accurately characterize the obstruction, systolic function, symptoms, and echocardiographic progression. The arrival of more specific therapies does not replace the need for rigorous clinical assessment; on the contrary, it makes it even more necessary.

For all these reasons, the work by Cáceres et al. should be read as more than just a descriptive registry. It is an invitation to organize care for HCM in Argentina. Its data show progress, heterogeneity and concrete opportunities for improvement. The registry provides national, multicenter, real-world information on a complex disease in a setting underrepresented in the literature, and its greatest value lies in transforming that information into a tool for action. Future stages may delve deeper into the characterization of patients studied using CMRI and genetics, into the criteria for indicating advanced investigations, into family screening and into referral pathways. Far from diminishing the value of the work, these questions point to its natural course of development.

Finally, this registry contributes to the field of hereditary genetic cardiomyopathies by transforming clinical perception into concrete information: HCM requires specialized knowledge, but also healthcare organization. Its findings, together with the 2025 Argentine Consensus, should stimulate more integrated, equitable and reproducible care for Argentine patients with HCM. Continuing medical education, the implementation of simple protocols, the prioritization of accessible tools, the early referral of complex cases and the establishment of regional networks are realistic steps towards reducing inequalities and improving the quality of care.

Conflicts of interest

None declared

(See authors conflicts of interest forms on the website).

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