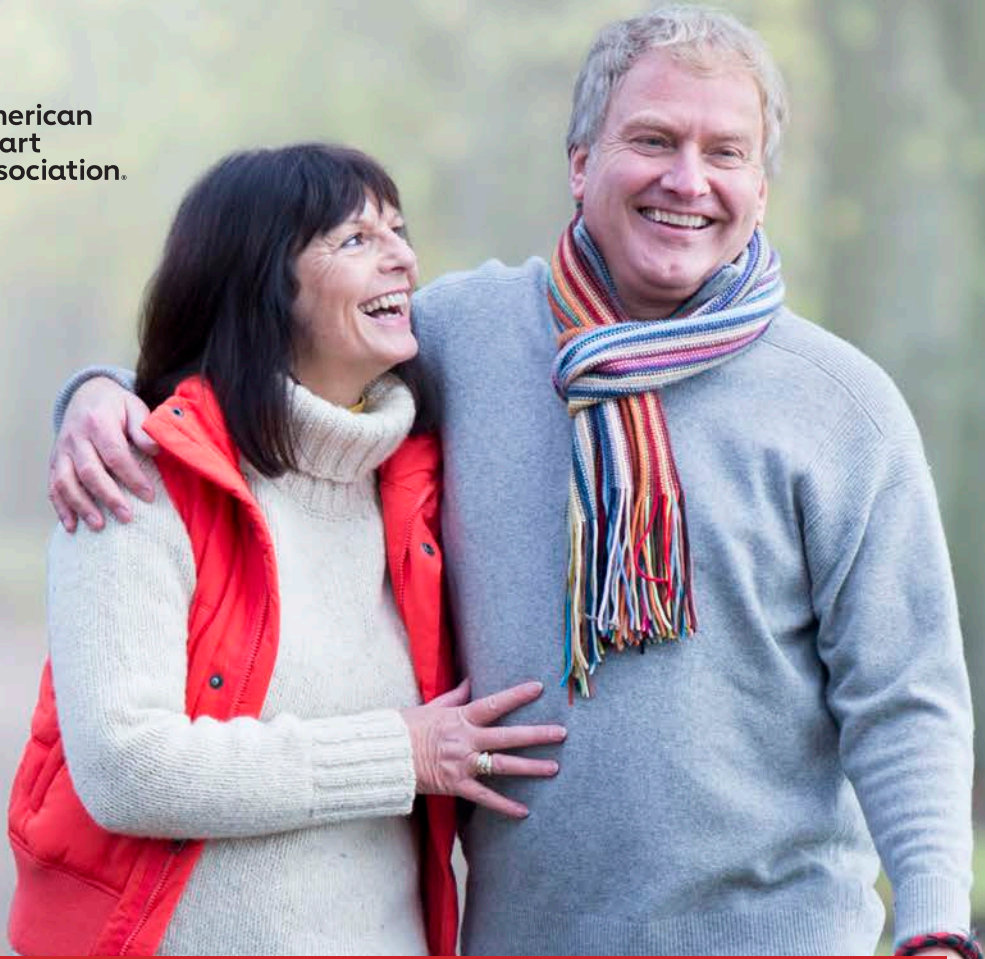




American
Heart
Association.



HYPERTROPHIC CARDIOMYOPATHY

ROUNDTABLE REPORT





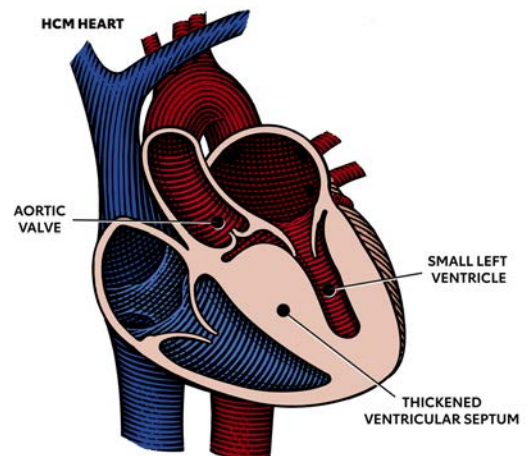
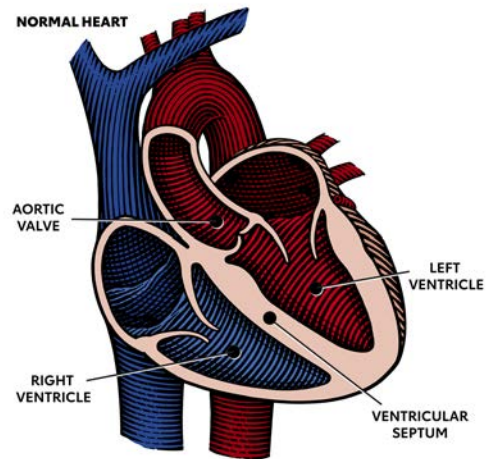
INTRODUCTION

Hypertrophic cardiomyopathy is a progressive disease in which the walls of the heart thicken and stiffen, making it difficult for the heart's chambers to adequately fill up or pump out blood.

There are two types of HCM. In an estimated two-thirds of people with HCM, the wall that separates the two bottom chambers of the heart becomes enlarged and restricts blood flow out of the heart (obstructive hypertrophic cardiomyopathy).¹

About one-third of people with HCM do not have significant blocking of blood flow (nonobstructive hypertrophic cardiomyopathy).¹ The heart's main pumping chamber is still thickened and may become increasingly stiff, reducing the amount of blood taken in and then pumped out to the body with each heartbeat.

HCM is the most common form of inherited heart disease, and it can affect people of any age. It is estimated that 1 in every 500 adults living in the U.S. has HCM, although HCM is underdiagnosed.² People with HCM face a risk of death more than three times greater than the average person of the same age without HCM.



1. Raj MA, Ranka S, Goyal A. Hypertrophic Obstructive Cardiomyopathy. [Updated 2022 Oct 31]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK430820/>

2. Hypertrophic cardiomyopathy (HCM). American Heart Association. Updated May 13, 2022. Accessed December 16, 2022. <https://www.heart.org/en/health-topics/cardiomyopathy/what-is-cardiomyopathy-in-adults/hypertrophic-cardiomyopathy>

People with HCM have a higher risk for experiencing atrial fibrillation, heart failure, stroke and even sudden cardiac arrest. HCM has been identified as the most common reason for cardiovascular sudden death in young people.

Yet HCM is not widely known or understood by the public, and health care professionals can struggle to provide a timely diagnosis or guideline-directed treatments. These gaps illustrate a need for education, since early detection and management may help reduce some of the consequences of HCM – including serious complications and comorbidities – and may improve patient outcomes.



The American Heart Association, a global leader in the fields of cardiovascular disease and stroke, is dedicated to discovering, disseminating and applying scientific knowledge. Recognizing the importance of confronting HCM and the broad range of individuals working on various aspects of the disease, the AHA hosted two previous roundtable discussions on HCM.

The first virtual roundtable, held in 2020, included patients and multi-disciplinary health care professionals. It covered topics including diagnosis challenges and education resource gaps. The second roundtable was held in-person at Scientific Sessions in 2021. It included members of the AHA's HCM Scientific Advisory Group and highlighted updates to HCM scientific guidelines.

A third roundtable was held August 22, 2022. The objective was to build on previous discussion and to identify steps needed to achieve an "ideal state" for patients with HCM through a guided discussion with patients and multi-disciplinary health care professionals. The virtual roundtable was facilitated using a semi-structured interview guide with HCM patients and multi-disciplinary health care professionals. The discussion recording was transcribed, and inductive and deductive thematic analyses identified key themes that are outlined in this report.



BARRIERS TO DIAGNOSIS

A timely diagnosis is an important part of an HCM patient’s care journey to ensure access to effective treatments that are key for extending length and quality of life.

During the 2020 roundtable, several barriers to timely HCM diagnosis were cited. These included access to health care due to preexisting conditions, regional differences in care, access to affordable care, physician gaps in understanding treatment guidelines and incomplete patient information to reach diagnosis. The conversation continued in this roundtable where health care professionals and patients agreed that patients face a variety of barriers in receiving an early diagnosis.

“...but even before they have access to a knowledgeable clinician, **somebody** has to make the right diagnosis.”



Patients expressed challenges in finding the right HCP to provide an HCM diagnosis, including being directed to the proper specialist by their primary care physician or cardiologist. Patients also faced challenges accessing facilities providing top HCM care, such as an HCM Center of Excellence. The high cost of HCM care and getting health insurance to cover specialized care were also noted as barriers.

Similarly, HCPs identified obtaining appropriate referrals and connecting patients to the right level of care as barriers to timely HCM diagnosis, especially with the scarcity of care for HCM patients in the southern United States. Low health literacy and low patient resources were also identified as barriers to diagnosing HCM, which can also pose challenges for obtaining accurate family medical history. In addition, the nuances and complexity of HCM, along with HCM’s overlap with other disease processes, continue to be barriers to timely diagnosis.

POTENTIAL SOLUTIONS TO OVERCOMING BARRIERS TO DIAGNOSIS

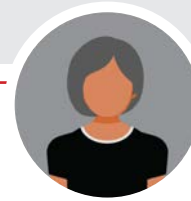
Several potential ideas to overcoming barriers to a timely HCM diagnosis emerged from the roundtable.

Patients suggested that there should be more public education about HCM to raise awareness about HCM and its signs and symptoms. This would also require improving general health literacy among patients and the public. Patients also thought that athletes could be regularly screened for HCM and that in general, there should be better access to mental health services for patients living with HCM.

HCPs also identified the need for more education on HCM among professionals, including increased training on HCM for cardiologists and surgeons. Digital health and artificial intelligence-based tools could also combat some of the barriers that patients face for a timely diagnosis. For example, use of digital health tools through increased telemedicine access could put patients in touch with the proper specialists faster. HCPs also suggested that providing a carrier screening during pregnancy could lead to better and earlier diagnoses. At the systems level, HCPs agreed that they need to continue to build up the model for the coordination of care, perhaps by using a champion-based model.



*I'm from the south, and I felt like there was a scarcity of care in lots of areas...and so I think that we need to train more physicians and train more physicians that would be willing to go to places [where] there are not centers. **Not everyone can fly to Mayo Clinic, and it's not just training cardiologists, it's training surgeons.... we [need] to have a full team.***





IMPROVING COORDINATION OF CARE

It can be challenging for to HCPs to treat and help their patients manage HCM.

HCPs discussed the challenges they face when collaborating with members of their health care team and identified ways that HCPs could work together as a team to provide optimal patient care.

To build up the model for the coordination of care, HCPs should include patients at the center of their care, while involving patients' families in HCM education and cascade genetic testing. Integrating telemedicine could increase access to specialists. Improving communication across electronic medical records and health systems is also crucial to optimal treatment. HCPs discussed the inequities that exist within medicine from primary care to specialty care and noted the importance of allocating resources to address them. Health care systems and payors should also allocate more financial resources to facilitate coordination across departments and health systems.



Assuming you have a team that thinks it's important, I think you start to figure out ways to work together and communicate together. Different EMRs can cause a lot of miscommunications... patients often have providers at different hospitals, different systems, different places. And so, trying to keep everyone on the same page is challenging.



KEY COLLABORATORS AND CONTRIBUTORS

Roundtable participants identified the key collaborators and contributors for delivering optimal care and the role each should play in improving the diagnosis and treatment of patients living with HCM.

Patients suggested that schools be included to improve health education. They also expressed a desire to have increased access to allied health professionals to learn more about exercise and nutrition to help manage HCM symptoms. Patient advocacy organizations like the Hypertrophic Cardiomyopathy Association were also identified as key resources in connecting patients to optimal care.

HCPs again recognized that patients, alongside their families, should be at the center of their care. In addition to HCM specialists, key collaborators and contributors that should be a part of the coordination of care include primary care physicians, imaging specialists, genetic counselors and genetic labs, and allied health professionals such as exercise physiologists and registered dietitians.

HCPs noted that the CDC’s genomics division supports guideline documents that show an evidence base for genetic testing, making them vital to elevating HCM to a “Tier 1 Disorder” and backing genetic testing for HCM. HCPs also discussed the importance of including business managers, health care systems leadership, and insurers/payors as key collaborators to create a value-based model for HCM to overcome silos and barriers to optimal patient care. Lastly, HCPs acknowledged that the “Centers of Excellence” model requires more scientific rigor and oversight.



I think that there are ways to work with your health system and your leadership to show them that the comprehensive care is actually meaningful, most importantly, but also can be profitable to the health system.





RESOURCE GAPS

Resource gaps can have a negative impact on the timely diagnosis and provision of guideline-directed treatment of HCM. So there was discussion about current patient and HCP resource gaps and steps that can be taken to address them.

Patients noted that an initial diagnosis can be overwhelming, and there is often a lot of information provided in a single visit. There is a need to improve how information is provided, which could be supported by developing better ways for patients to record or document visits with HCPs. Patients cited a desire to connect with others who have an HCM diagnosis and a need for creating better pathways to peer-support resources. They also want more guidance on managing day-to-day living after diagnosis, particularly on diet and exercise.

HCPs highlighted significant gaps in exposure to HCM and relevant topics, such as genetics, during all levels of medical training. They also stated a need for more geneticists and genetic counselors, and a desire for standardized tools and methods to discuss genetic risk for HCM with families. Finally, several HCPs expressed a desire for tools and resources in multiple languages that they can review with patients, especially at the time of diagnosis. Having something to focus on and review would let patients take notes within the document and use it as a takeaway resource.

“ ...[HCM] can be managed, like you manage diabetes. I wish that I had had someone...put it in perspective. Not that it's not serious and not that I can be cavalier about it, of course not. But I mean, I still needed to live my life, my day to day. **”**



“ Something that the HCP can go through with the patient in the office...a document that they look at together during the visit and patients can jot down their questions or write notes, instead of saying it all and...making sure it's available for multiple languages, not just English. **”**



TECHNOLOGY

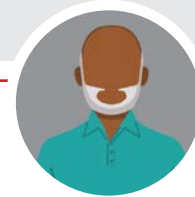
Although technology and digital tools have the potential to support the HCM patient journey, there are also limitations.

Patients highlighted the use of apps to learn and practice relaxation techniques and noted the benefits of online resources to track general health behaviors (e.g., exercise). They also said having an easily accessible and user-friendly way to track HCM-specific health metrics would be beneficial.

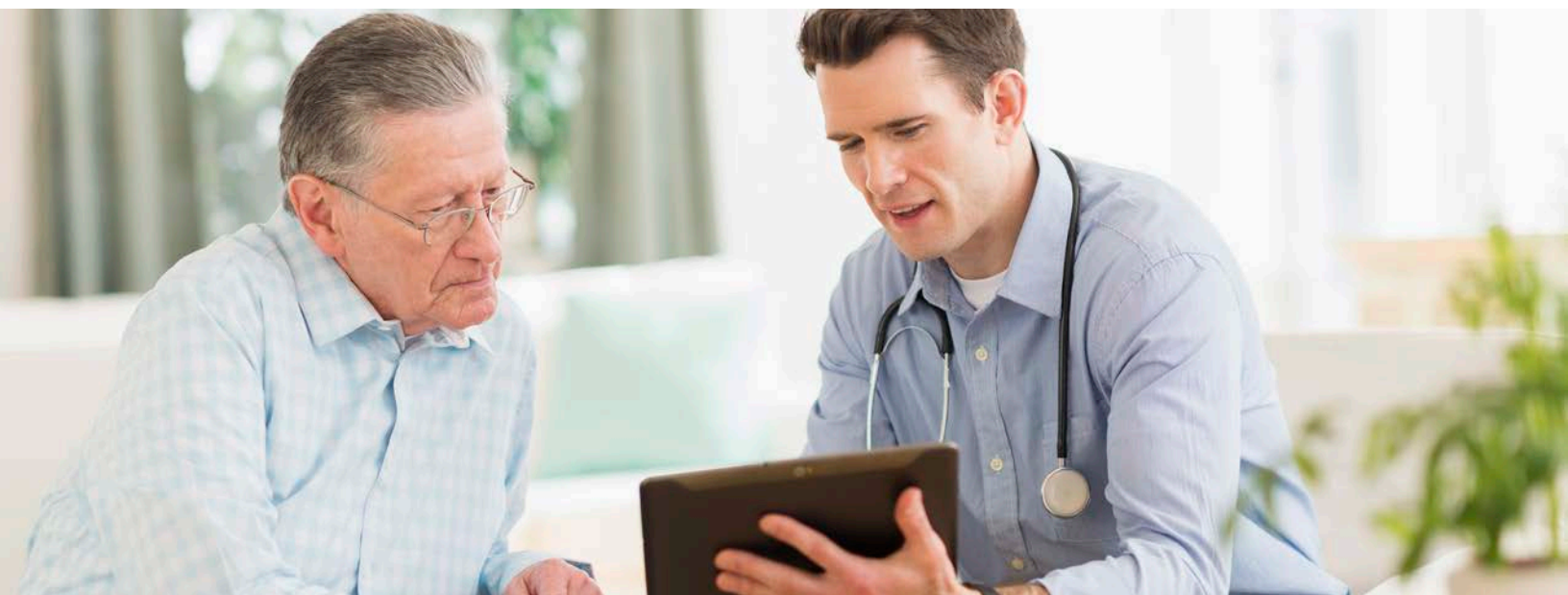
HCPs made note of technology tools and regulations around their use that might not be widely known or used, such as the information blocking rule which allows patients better access to their EMRs. They advocated for increased awareness and use of these resources. One person noted that not every disease or topic needs its own specific app, and we should make better use of existing tools and resources instead of using limited financial resources to create new ones. The potential for AI to assist with diagnosis was mentioned at several points. For example, one HCP described how AI can assist with echo imaging by flagging potential diagnoses for echos that meet specific criteria.



...to have that [nutrition, exercise, and HCM stats] information in an app or some form like that might be beneficial for the patient, because then their immediate personal information is right there in hand.



Part of it is, can you get companies to care about this, and can it be financially beneficial for companies to build you tools that you want?





The use of telemedicine has increased, particularly during COVID when there was better institutional and financial support to increase patient access to this type of care. Telemedicine can play a critical role in connecting patients with specialists and genetic counselors, particularly patients who lack local access to those resources. There is also potential to improve telemedicine platforms and increase their usability for clinicians, such as software packages that would allow clinicians to more easily review echos remotely.

Finally, HCPs cited the need to improve EMRs. One HCP said “the struggle is real” when using EMRs, describing how scanned PDFs of patient results can be a barrier for sharing patient information among HCPs or health systems, and a limitation for using the information for research purposes. They also described how the cost of improving EMR platforms is a barrier, and that there is a need to make improving these tools financially beneficial for companies.



*As I last looked, there are about 4,500 to 5,000 genetic counselors in this country and maybe about a thousand geneticists... **Obviously, not nearly enough out there to be able to see patients where they are, so telemedicine is going to be a really important part of that.***



IDEAL STATE

Patients described an “ideal state” that included increased awareness and management of HCM among patients and health care professionals, a greater sense of empowerment and trust in health care systems; and improved connections with allied health, mental health and peer support services. They wanted to emphasize that HCM is not a “sudden killer” among diagnosed patients, and there should be better access to resources to help them learn to live with the disease. Patients also wanted to see faster referrals to specialist care when needed.

HCPs described an “ideal state” as increased awareness of and training in HCM and genetics at all levels of medical education, equitable and timely access to care and diagnosis, improved models of care, and scientific advancements to improve treatment and diagnosis. HCPs cited financial barriers to achieving this state, particularly the need to improve health care financing to facilitate HCM care coordination across departments and health systems. The role of technology and digital tools was also identified as key to achieving the “ideal state,” with HCPs noting both promise and limitations in their ability to improve HCM diagnosis and treatment.

“*...the future would be having these multidisciplinary heart team approach clinics all over the country and not just in pockets. It’s hard to get there... but that is what my goal is, by the end of my career, that we won’t have people dying because they can’t get to a place where they can get good care.*”



“*...at the end of the day, the most important thing is the patient. So, even if that patient needs to be taken care of by someone else, whether it’s a specialist, a different center, a different office, that those phone calls are not put on the back burner. Because if it wasn’t for advocating for myself, I clearly wouldn’t be here.*”



Key steps highlighted by HCP include:

- » Formalize and expand HCM training to cardiology fellows
- » Improve genetics education across all medical training
- » Increase access to genetic testing, mainstreaming genetic testing
- » Remove current inequities and ensure all individuals are diagnosed and involved in research
- » Establish criteria for HCM Centers of Excellence
- » Adopt an interdisciplinary “Heart Team” approach in geographically diverse areas
- » Adopt group care model for patients with HCM
- » Connect patients with mental and emotional health resources
- » Continue scientific advancements including funding for basic science research to understand HCM, finding a reliable HCM biomarker, and developing more effective medications
- » Increase buy-in from hospital administrators

CONCLUSIONS

Reaching an “ideal state” for diagnosing and treating HCM patients requires systematic changes at the patient, health care professional and health care system levels. Addressing the steps outlined during this discussion could vastly improve treatment, outcomes and quality of life for patients with HCM as well as support health care professionals as they provide timely, guideline-directed diagnosis and treatment.



ACKNOWLEDGMENTS

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