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Living with hypertrophic cardiomyopathy: patient perspectives from the UK and Ireland

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Authors:

Maryam Jetha,[1] Ciara O'Dwyer,[1] Mark Hughes,[1] Nuala Ryan,[2] Harshil Dhutia[3]

Affiliations:

[1]Medical Affairs, Cardiovascular, Bristol Myers Squibb, UK and Ireland
[2]Clinical Research Consultant and Patient Advocate, Nicolaides Baraitser Syndrome Worldwide Foundation, Dublin, Ireland
[3]Consultant Cardiologist and Clinical Lead for Inherited Cardiac Conditions Service, Glenfield Hospital, Leicester, UK

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Executive summary

Hypertrophic cardiomyopathy (**HCM**) is a common cardiac condition with a global prevalence rate of **1** in **200** to **1** in **500** people. Symptoms of **HCM** can be commonly mistaken for non-cardiovascular conditions, leading to misdiagnosis and delayed treatment.

To better understand their experiences of living with HCM and the perceived barriers to treatment, patients with HCM were invited to participate in two virtual roundtables to discuss their experiences with HCM diagnosis and management. From the perspectives of the patients who participated in the roundtables, the overarching theme that emerged was the variability in patient experience of HCM and access to treatment. Within this theme, **three key challenges arose**: **(1)** regional variance in access to healthcare and diagnosis pathways; **(2)** disparity between perspectives of patients and healthcare professionals regarding the importance of symptoms; and **(3)** gaps in mental health support and lack of digestible HCM information that is tailored for patients.

In this whitepaper, we suggest ways to address these challenges based on the insights provided by the roundtable participants. Firstly, educating general practitioners (GPs) about HCM signs and symptoms would help delineate the common overlap of HCM symptoms with other conditions, supporting patients in receiving a timely diagnosis and delaying the risk for disease progression. Secondly, additional or tailored support for patients during an HCM diagnosis would help them understand their condition, and developing post-diagnosis support resources would empower patients to take control of their health journey. Lastly, consistent access to specialists and multidisciplinary support would help reduce the disease burden for patients living with HCM.

☆ Introduction

Hypertrophic cardiomyopathy (HCM) is a common cardiac condition with a prevalence of 1 in 200 and 1 in 500 individuals worldwide.[1-4] It is often inherited and the child of an affected parent has a 50% chance of inheriting the condition.[3,4] In HCM, thickening of the heart muscle causes the walls in the heart to stiffen, thereby making it difficult for the heart to contract efficiently and pump blood around the body.[3]

It is estimated that **one-third of patients have a non-obstructive type of HCM**, where the thickened cardiac muscle does not impede blood flow from the heart.[4,5] The remaining **two-thirds of patients have obstructive HCM**.[5] In obstructive HCM, the heart muscle is so severely thickened that it causes an obstruction and reduces the blood flow from the heart to the body.

This may lead to symptoms such as **breathlessness, fatigue, chest pain, light-headedness, palpitations, and fainting** for some patients, while others may have no symptoms (i.e., they are asymptomatic).[3-6]

These symptoms are not specific to HCM and can be easily mistaken for other conditions that are not always associated with heart problems, leading to misdiagnosis and, therefore, delayed and sub-optimal treatment.[7,8]

To better understand patients' experiences with HCM diagnosis and management, **Bristol Myers Squibb (BMS) conducted two virtual roundtables with patients from the United Kingdom (UK) and Ireland**. We hereby present patients' perspectives on their HCM diagnosis and explore their experiences in the following areas: access to valid, patient-centric information; and treatment and care pathways.

Methods

This exploratory whitepaper was developed using a mixed-method approach in which qualitative and quantitative insights from patients living with HCM were combined with supplementary desk research. **BMS convened two virtual roundtables in which patients with HCM from the UK and Ireland participated**. Patients were invited to participate via clinician referral and through the patient networks and charities Cardiomyopathy UK, Croí, Irish Platform for Patients Organisations, Science & Industry, and Irish Heart Foundation. Five patients attended both roundtables and one new patient attended each roundtable, resulting in six patients per roundtable with a total of seven patients overall.

Of the seven patients who attended the roundtables, two were female. The patients were aged between **50 and 65 years** and came from the **Republic of Ireland, Northern Ireland, Scotland, and the South of England**.

Patient insights were captured between **November 2023** and **April 2024** via pre-meeting surveys and virtual roundtables. Before the first roundtable, patients were asked to complete a short survey about their experiences of diagnosis, where they find information related to their newly diagnosed disease, and their communication preferences (**Appendix**). Responses to this survey were used to guide discussions during the first roundtable, which was held in November 2023. The aim of this roundtable was to explore the patients' experiences from receiving their HCM diagnosis to the availability and accessibility of HCM-related information.

Before the second roundtable, patients were asked to complete another short survey about their experiences of living with HCM. This roundtable was held in April 2024 and explored the patients' experiences regarding their treatment and care pathways. The qualitative insights gathered from the patients were analysed with the support of an HCP who advised on potential opportunities to overcome patient-perceived barriers in their care pathway.

Through the methods outlined above, we identified three key challenges for patients living with HCM; these challenges form the basis for this whitepaper, which will now be presented.

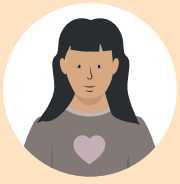
Challenges

An overarching theme that emerged from the roundtables was the variability in patients' experiences of their disease as well as access to care. Within this theme, three key areas of challenge arose: **(1)** regional variance in access to healthcare and diagnosis pathways; **(2)** disparity between the perspectives of patients and HCPs as to which symptoms are important; and **(3)** gaps in mental health support and easily understandable, HCM-related information for patients.

Regional variances

Discussions during the roundtables highlighted disparities in the pathway to diagnosis for patients with HCM. In response to the pre-meeting survey about their experiences of diagnosis, three patients reported receiving a diagnosis almost immediately in an emergency setting, one patient received a diagnosis within 3 months of symptoms, and one patient reported a diagnosis timeline spanning 1–5 years. The sixth patient did not respond to the question.

Differences in the approach to care between primary and secondary care settings can appear disjointed for patients with HCM. This is often frustrating for those patients who feel that their condition is not being properly managed by HCPs, highlighting the importance of a specialist service for patients with HCM. From the perspective of the patients in the roundtables, there was poor communication during the transfer between specialists, even in secondary care settings, which led to information gaps and uncertainty. Patients believed that this impacted the quality of care they received.



'Once you get past the general practitioner diagnosis, the treatment and care is fantastic.'

'To actually get to see a cardiologist is like pulling teeth, and I have to go through PALS each time.'*

*PALS is the Patient Advice and Liaison Service; a confidential advice and support service provided in every National Health Service (NHS) Trust in England and Wales.

In response to being asked how satisfied they were with their overall treatment and care, the satisfaction levels varied among patients.

Four patients were extremely satisfied, one expressed moderate satisfaction, and one patient showed a degree of dissatisfaction with their experience. In explaining their answers, one patient expressed immense gratitude for a diligent and attentive cardiologist who gave them a sense of security by being approachable, meaning that the patient was able to freely express their concerns. In contrast, another patient initially experienced a prolonged and impersonal diagnostic process but found exceptional care following a transfer to a new hospital. Another patient believed they were left to self-manage their condition without a clear care plan or routine checks beyond reliance on an implanted device, with dependency on this device leaving the patient feeling 'dehumanised'. Another patient was offered the opportunity to minimise hospital visits with use of a home monitor, attending check-ups every 6 months and annual ultrasound scans, ensuring consistent monitoring.

These disparities in experience were suggested to be due to possible regional differences in healthcare systems across the UK and Ireland.

Two patients complained about **having to travel further afield** because of the lack of a cardiologist at their local healthcare service. Notably, in a recent Cardiomyopathy UK survey involving patients in the UK, regional disparities in the quality of healthcare and treatment were also observed.[8] Although some respondents reported excellent care and access to cardiac rehabilitation, others reported difficulties in getting appointments and believed that they were 'left to manage alone'.[8]

The patients in these roundtables believed that they were inundated with information from a variety of sources, such as leaflets given by the specialist or treatment team, patient groups, charities, publicly available health information, or online search engines.

Patients found that using online search engines could be like going down a 'scary rabbit hole' due to the abundance of factual yet emotionally sterile information, and a lack of guidance beyond the clinical details. They also found that a lot of the information was too technical and difficult to understand, thereby indicating a need for visually appealing informational material to aid comprehension.

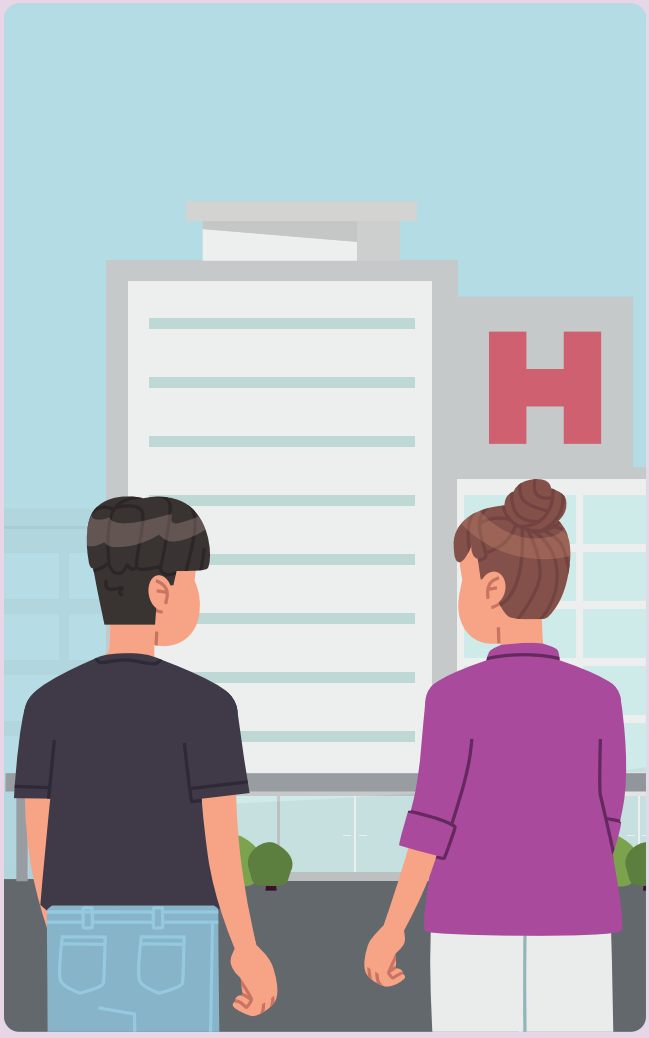
'I became obsessive, spending many nights lying in bed on my phone searching for information.'

Of the five patients who responded to the survey question: 'What do you wish you had known when you were diagnosed that no one told you?', one patient wished for earlier and more comprehensive information about the biology of their condition, potential effects on lifestyle, long-term prognosis, genetic implications, work advice, early counselling, and accessible support resources. Another expressed a longing for earlier clarity in diagnosis and better support. A third patient wished for a detailed care plan specifying hospital cardiology reviews and GP expectations, wanting structured guidance after diagnosis. These sentiments echo the recent Cardiomyopathy UK survey, which found that a lack of follow-up care left many patients feeling abandoned and without a clear treatment plan.[8]

Potential opportunities

To address the challenge of regional variances, we propose **consistent access to specialists and multidisciplinary support** to reduce the burden for patients living with HCM.

HCM is a chronic disorder that can progress over time and patients may develop serious heart-related complications such as heart failure.[9] Thus, it is important to **detect and manage HCM and its symptoms promptly** to delay serious complications for as long as possible by **ensuring that patients have easy access to specialist centres and receive an early diagnosis and appropriate care**. Establishing criteria for HCM centres of excellence and including coordinated care at such centres may result in earlier diagnosis and treatment, thereby improving outcomes. Streamlining referral pathways and establishing regional networks – including hub-and-spoke models and providing genetic counselling and specialist nursing in a one-stop clinic – could help to shorten the diagnostic process, reduce logistical inconvenience for the patient and provide a better patient and family experience.



In addition, improving communication across health systems is crucial to reduce duplication and delays to diagnosis and optimal treatment. The use of digital health and artificial intelligence-based tools could also combat some of the barriers that patients face in receiving a timely diagnosis (e.g., digital health tools through telemedicine access can put patients in touch with appropriate specialists faster and result in earlier diagnosis).[10,11]

Importance of symptoms

Many patients with HCM may be asymptomatic, while others may experience symptoms such as shortness of breath, chest pain, light-headedness, and fainting.[3,4,6] For these patients, the first contact with the healthcare system to assess symptoms may be via their GPs, while some patients may have atrial fibrillation or cardiac arrests requiring emergency care.[3,4]

Based on the insights provided by the roundtable participants, the journey to HCM diagnosis for patients who first consulted their GPs after experiencing symptoms was often long and frustrating; misdiagnosis with other conditions (e.g., asthma, anxiety, thyroid issues) resulted in a delay in receiving the correct diagnosis and necessary testing needed to confirm the HCM diagnosis. Patients believed that the healthcare system was '**siloed**'; there was '**no joined-up thinking**' and '**a lack of understanding from GPs as to what HCM is**'.

'Nothing is joined up, nothing flows…the heart is the last thing they think about.'

'You're a whole person, and I was being looked at…very much tunnel vision as far as my asthma was concerned, and if they had done heart-related scanning, perhaps they might have picked up something then and I would not have had the obstructive element of HCM… I am left with what I've got.'

Patients believed that symptoms such as shortness of breath and tiredness may be overlooked and not considered as relating to a heart condition.

'A lot of people would have the shortness of breath and say it's asthma or it's their lungs or whatever, not think about their heart.'

'If I go back six years, you know, when I didn't think I had a problem, but I was experiencing symptoms, I was experiencing tiredness, shortness of breath.'

An additional frustration expressed by patients was the disparity between what they viewed as important versus what the HCP viewed as important. Patients believed that HCPs disregarded anecdotal accounts of how they really felt since the test results appeared fine on paper.

> *'I don't always feel well but, because I look well, you know, it's very difficult to get across that maybe something has changed in my health.'*

Potential opportunities

Symptoms of HCM can overlap with known conditions; therefore, education for GPs regarding HCM signs and symptoms is important to help delineate the common overlap with other conditions and ensure that patients receive the correct testing and a timely diagnosis.

Formalised HCM training programmes for cardiology registrars, improvements in genetics education across all medical training, and continuing professional development events in primary care will further help to raise awareness of HCM.

In addition to education for HCPs, improving the general health literacy among patients and the public through engagement with patient and public groups can help improve outcomes;[12] however, patients may need additional resources and support to understand their condition and its management. Ensuring consistency in the availability of accessible and alternative language communication templates, and providing clear information to patients about their care, will further help to support patients. Shared decision-making should also be considered with HCPs involving patients in treatment decisions to take into account their preferences and values.

Gaps in mental health support and information for patients

A diagnosis of HCM can be distressing and life-changing. Depending upon the disease severity, a HCM diagnosis may have a significant impact on the daily lives of patients.

'So, as far as my emotional mental state, I did have some dark days when going to bed thinking, you know, "is this it"?'

Thus, it becomes critical that such a diagnosis is given in a comprehensive and sensitive manner. It is imperative that patients are supported through their diagnosis journey; however, when receiving their diagnosis, some patients believed they were given an 'absolute worst-case

scenario'. One patient reported being copied into a letter between the cardiologist and GP, while another was told that they might die any day. Receiving a diagnosis in such a manner can impact patients' feelings and leave them extremely distressed.

> *'I was left at 42, super-fit guy in those days, was stunned, in tears, apologising to my wife thinking this was the end of my life. Every night I thought I was going to die. I was in a terrible mental state.'*

Patients voiced their displeasure at the lack of mental health support for them and their families. A diagnosis of HCM often involves changes to the activities and exercises that a patient can undertake safely.[13,14] In addition, the genetic element of HCM can impact family members; consequently, the impact of a HCM diagnosis is not only felt by the patients themselves, but also by their families and the friends who support them.[8,15]

> *'Part of cardiomyopathy, it's not just the patient—it's the people who support them as well.'*

It was common for patients to continuously have to advocate for their own care rather than the healthcare team proactively taking responsibility; however, for patients with no prior exposure to the healthcare system, there can be long periods of silence before they hear back after a test or appointment, making them anxious and stressed.

'I was able to get to my diagnosis quite quickly, which is really, really good, but after that point, that's where the, sort of, wheels fell off the wagon and the same cardiologist that I saw became very distant.'

Patients also believed they were burdened with having to lead the communication. In many instances, the healthcare team did not understand or have complete information about a patient's background, leaving further gaps in communication; the responsibility of communicating all the correct information was thus left with the patient. '**I had to keep phoning up and saying, "When am I going to see a cardiologist?**" And they kept saying, "**Well, you're on the waiting list"**. I said, "**But I'm supposed to be seen every year**".'

Even if the communication was timely and efficient, it was not always patient-friendly, which left patients with even more questions.

'You don't get spoken to directly, you get a letter done to your GP with you in cc, which is very sort of dehumanising.'

Some patients described charities as being important sources of support and information.

'My saviour was the Irish Heart Foundation, and a cardiomyopathy support group, which I became very involved with for years when I could. Also, the British Heart Foundation, they had a cardiomyopathy support group. I got an awful lot of my support and information from there over the years. They've been super to me. Meeting other people with the condition was just amazing because we all learn from each other.'

Patients believed that some gaps in support were being filled by charities.

'There's a nurse you can call. There are weekly chat groups you can check into.'

Potential opportunities

It is important for patients to have comprehensive care packages. Supporting patients during a diagnosis of HCM means more than just offering mental health services: it involves delivering the diagnosis with clarity and compassion, ensuring the condition is easily understood, and providing clear guidance on accessing further information and support.

The development of comprehensive post-diagnosis support resources could include provision not only of factual details but also emotional guidance that can help empower patients to navigate their health journey with confidence and knowledge.

Providing comprehensive information packs outlining the condition specifics, tailored management pathways and treatment options would encourage informed decisions throughout the patient journey. Patients also highlighted a need for clearer communication.

Conclusions

From the perspectives of the patients who contributed to these roundtables, the overarching theme that emerged was the **variability in the patients' experiences of living with HCM and access to specialist care**. **Three key challenges arose within this theme:** regional variance in access to healthcare and diagnosis pathways; disparity between the perspectives of patients and HCPs regarding the importance of symptoms; and unmet needs in mental health support and HCM-related information for patients. There are potential opportunities to address these challenges that may be explored in future research: **(1)** education for GPs regarding HCM signs and symptoms to help delineate the common overlap of HCM symptoms with other conditions; **(2)** support for patients during a diagnosis of HCM to ensure that they understand their condition, and the development of post-diagnosis support resources to empower patients to take control of their health journey; and **(3)** consistent access to specialists and multidisciplinary support to reduce the burden for patients living with HCM.

From diagnosis onwards, every patient deserves consistent access to specialists and comprehensive multidisciplinary support, including mental health services, integrated with their treatment and care. By equipping patients with the necessary tools and resources to manage their condition, we can significantly reduce the burden of living with HCM and empower patients to take control of their health journey.

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