Cardiomyopathy UK
the heart muscle charity

James Lind Alliance
Priority Setting Partnerships

Cardiomyopathy:
The 10 priority questions for research

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Foreword

Alison Fielding, Vice President (Patient Voice), Cardiomyopathy UK

This is an exciting time for cardiomyopathy research, and I am delighted to present to you, in this report, the future research priorities for cardiomyopathy.

When I first came across James Lind Alliance Priority Setting Partnerships, I could see the benefits and I was keen that the cardiomyopathy community should have the opportunity to participate in one. The partnership offers an equitable and scientific way to ensure that the interests of stakeholders from both the personal and professional sides of the cardiomyopathy world are represented.

The highest ranked research priority centres on the psychological effects of cardiomyopathy. It is there due to the strong voice of patients, who demonstrated, throughout the process, that cardiomyopathy is more than a physical diagnosis or group of symptoms.

As a patient organisation, it is important to Cardiomyopathy UK that we listen to our community. The Priority Setting Partnership has allowed us to improve our understanding of the research priorities, as decided by people affected by cardiomyopathy and healthcare professionals. We can now develop our research strategies for the next decade and beyond. My thanks go to everyone who participated and to those who made this possible.

Professor Perry Elliott, President, Cardiomyopathy UK

The Cardiomyopathy Priority Setting Partnership has given us a unique opportunity to bring together people with lived experience of cardiomyopathy and the healthcare professionals involved in their care. As clinicians, we are asked questions by our patients' and we do not always have the answers. This is the motivation to undertake research that answers the questions posed to us by those people in our care.

As researchers, however, we can have a focus on our own research projects and specialisms. The Cardiomyopathy PSP involved healthcare professionals and allied health professionals from different disciplines, including cardiologists, genetic counsellors, inherited cardiac nurses and physiotherapists. Each of them had their own perspective on cardiomyopathy research priorities.

Of course, people living with cardiomyopathy have other ways of viewing their condition and understanding the impact it can have on their everyday lives. Often this view can differ from those of healthcare professionals. This diversity in perspectives in the PSP process leads to a varied and broad list of priorities achieved through consensus.

I appreciate that Cardiomyopathy UK has led on this PSP with the James Lind Alliance and I look forward to seeing the outcomes of the research arising from these priorities.

The 10 priority questions for cardiomyopathy research

What is a priority setting partnership?

Priority Setting Partnerships (PSPs) are run in partnership with the James Lind Alliance (JLA). The James Lind Alliance is a non-profit making initiative bringing patients, carers and clinicians together in JLA priority setting partnerships. The purpose is to identify and prioritise unanswered questions for specific medial conditions or areas of health.

PSPs bring together doctors, nurses, and other healthcare professionals, patients, family members, friends and carers, whose views are all given an equal voice in the process. Through a process of consultation and a workshop, they all decide on the ten priority research questions.

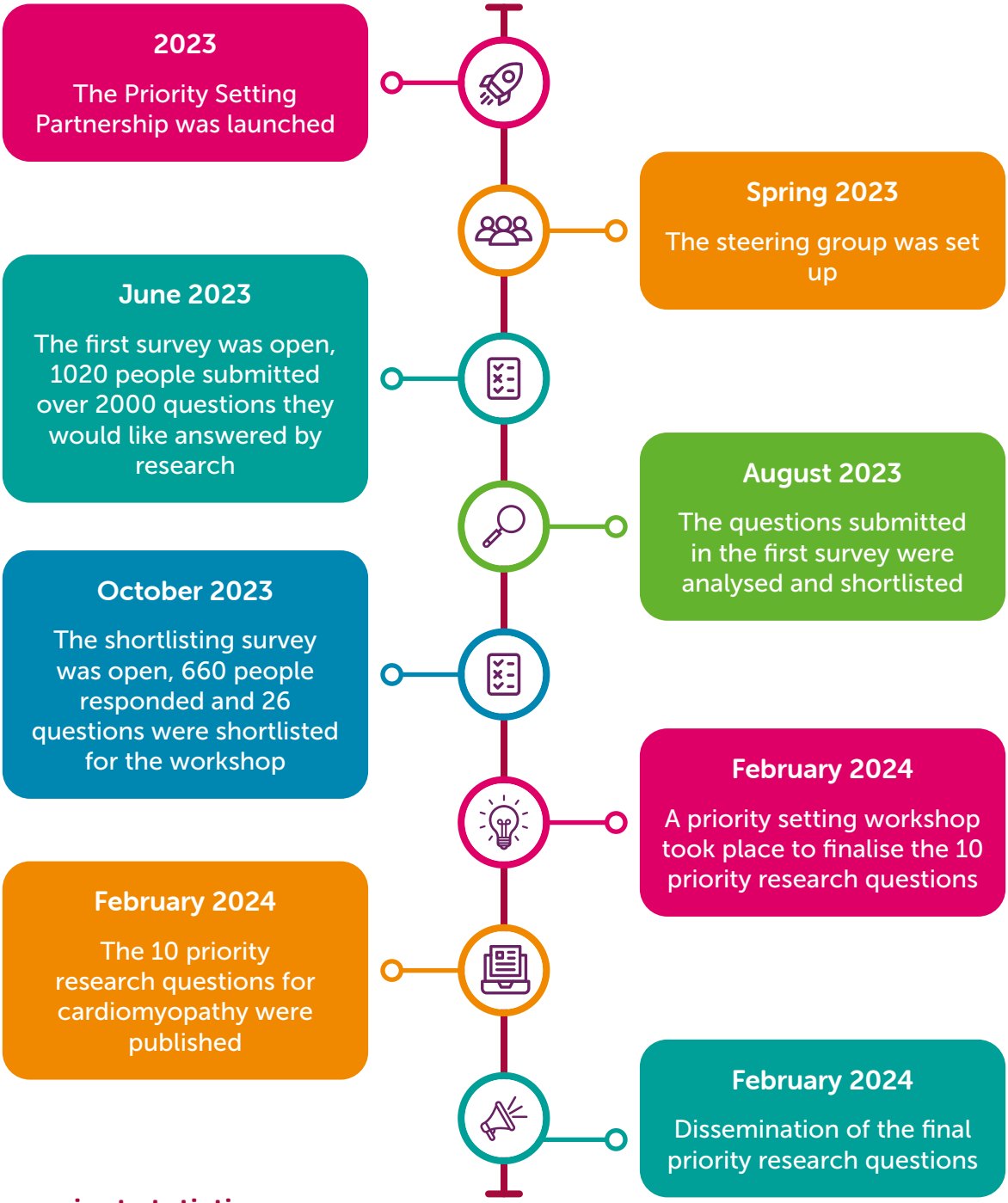
Why did Cardiomyopathy UK choose to run a priority setting partnership?

Cardiomyopathy UK recognised that while more research was increasingly being conducted into cardiomyopathy, research topics were decided on by researchers. These questions did not always correspond to the topics that people affected by cardiomyopathy were interested in. Cardiomyopathy UK wanted to address this and give people with lived experience the opportunity to shape research into their condition. The James Lind Alliance offers a robust and well-respected process for deciding on the research priorities, which was an important factor in deciding to run a cardiomyopathy priority setting partnership.

What is cardiomyopathy?

- Cardiomyopathy is a disease of the heart muscle: 'cardio' means heart, 'myo' means muscle and 'pathy' means disease.
- It isn't a single condition, but a group of conditions that affect the structure of the heart and reduce its ability to pump blood around the body.
- Around 1 in 250 people in the UK are affected.

The process



Key project statistics

- 7 partner organisations who supported with promoting the surveys.
- 1020 individual responses to the initial survey.
- Over 2000 questions submitted.
- Over 60 questions were listed in the shortlisting survey.
- 660 responses to the shortlisting survey.
- 26 questions taken to the final priority setting workshop.
- 31 workshop participants.

Priority 1:

What are the emotional and psychological impacts of living with cardiomyopathy? How are these best treated and managed?

"I found it very interesting, but not surprising that the top priority question agreed by attendees of the workshop was related to the emotional and psychological effects of living with cardiomyopathy. This was one of the top priorities in the three working groups, which shows that this is a real issue for those affected by cardiomyopathy. We know that mental wellbeing is so closely related to physical wellbeing and so if our research question can lead to better access, treatments & management of mental health, I'm sure it will have a positive impact holistically leading to improved quality of life for all."

- Ann Harrison-Power, relative

"Living with a condition like cardiomyopathy impacts almost every area of life, not least of which is the impact on wellbeing. However, uncertainties still remain around how best to manage and treat the emotional and psychological aspects of living with the condition. As a heritable disease, the impact across the wider family is a major consideration and my hope is that this question will stimulate research that will provide greater understanding and insight around how best to look after everyone affected by cardiomyopathy."

- Libby Jarman, relative

Priority 2:

How often should family members at risk of developing cardiomyopathy be screened and which are the best tests to use? When is it safe to stop screening?

> Assessment of risk in family members is something that most people with cardiomyopathy in the family will have experienced. For non-genetic cardiomyopathies, clinical screening of relatives provides a level of ongoing reassurance. However, in an already stretched healthcare system, it's important to know which tests provide the best predictive information of future disease as well as providing some level of certainty around when it is safe to stop testing. I hope this question will encourage ongoing research into this area and provide answers that allow for the most effective use of healthcare resources and importantly, provide reassurance for families affected.
>
> - Libby Jarman, relative

Priority 3:

Should treatment for cardiomyopathy be tailored to the individual, e.g. based on their specific gene variant, age or gender?

> The current "one size fits all" approach in healthcare is inefficient. To provide "patient centred care" treatments need to be tailored to an individual's personalised needs, considering their gene variant, age and gender along with their diet and lifestyle.
> - Ruth Martin-Harper, person living with cardiomyopathy

> I often hear these same questions from patients and families when we see them in the clinic and whilst we try our best to answer and care for them using the best evidence, sometimes that evidence is not strong enough to give definitive, tailored advice. Research into the Top Ten priorities can only help towards the realisation of personalised healthcare for cardiomyopathies.
> - Dr Tootie Bueser, Lead cardiac genetic nurse for the Inherited Cardiac Conditions (ICC) services at King's College Hospital and Guy's and St Thomas' Hospital

Priority 4:

What triggers the start of cardiomyopathy (e.g. age, stress, pregnancy, other health conditions)? How do these triggers work and can they be blocked?

> Cardiomyopathy is a very important research area that needs prioritising given the significant impact it has in the lives of our patients, and I'm delighted to hear this will now be addressed. Triggers and risk factors can be important and identifying them would be important to improve patient outcomes.
> - Professor Faizel Osman, Consultant Cardiologist

Priority 5:

Are there treatments which can prevent cardiomyopathy developing in people at risk? Are there treatments to stop it getting worse in people with symptoms?

> Most of the patients I see in the genetics clinic want to know – if they have the altered gene found in the family, how can they prevent the condition from occurring in them? Right now, all we can do is try to pick up any problems early. Preventing the condition from developing or finding ways to stop progression will be really key in helping people to decide if they want to have genetic testing for themselves and their children.
> - David Walker, Principal Genetic Counsellor

Priority 6:

What are the biological mechanisms that change heart muscle cells in cardiomyopathy? Could this understanding lead to new treatments?

> I am delighted that this topic has been chosen as a priority. To better understand the causes for any disease process is critical if we are to find solutions for preventing illness in the first place; cardiomyopathy is no exception to this dogma.
> - Dr William Moody, Consultant Cardiologist

Priority 7:

Why are people with the same genetic variant affected differently? Why do some people with a genetic variant never develop cardiomyopathy? Could this understanding lead to new treatments?

"I'm very thankful this is a research priority, my family has inherited ARVC affecting 7 family members, myself and 2 siblings inherited 2 DSG2 genes one from each parent. Despite having the same DSG2 genes it has affected us differently: My 95 year old mum has the gene but no disease, my sister died suddenly aged 46 and my brother died awaiting a heart transplant. My condition has progressed but I have a defibrillator and I'm on the latest medications so remain reasonable healthy. In one family the individual impact was different but devastating. We need to understand why we experienced cardiomyopathy differently in the one family, and research is vital to improving this understanding."

- Julie Taylor, person living with cardiomyopathy

"We currently tend to treat cardiomyopathy as one disorder when in fact there are many different types due different genes. When we understand the differences, we can work towards a more personalised approach to treatment and lifestyle management."

- Ruth Newbury-Ecob, Consultant Clinical Geneticist

Priority 8:

Do people with cardiomyopathy experience better outcomes if they are treated at a specialist clinic rather than a general clinic?

> A greater awareness of cardiomyopathies, primarily driven by the availability of new therapeutics, will inevitably lead to greater levels of diagnosis across the UK. Whilst this is undoubtedly positive news, it has the potential to highlight issues around how and where people receive the best and most appropriate care. This question will be key to an increased understanding around patient outcomes and will provide robust data to support service change if and where it is needed.
> - Libby Jarman, relative

Priority 9:

What does ongoing monitoring and long-term care for people with cardiomyopathy need to include?

> I am pleased that this is a research priority to understand what long-term care should include. From my experience as a cardiomyopathy patient it is critical to support the patient and their carer (whoever that may be) with a written, tailored plan to support that patient when they are discharged covering all aspects of their care, who will deliver it, when and who to contact, and how, in case of questions and emergency.
> - Tony Croft, person living with cardiomyopathy

Priority 10:

What are the best approaches to cardiac rehabilitation for people with cardiomyopathy?

> I'm really pleased that cardiac rehabilitation research is being prioritised by the charity. I believe cardiac rehab is a key step in our journey with cardiomyopathy. Cardiac rehab is brilliant for building confidence in your body again following a sudden change in health such as a new diagnosis, a cardiac event or cardiac surgery. It's important that we, as patients, know how to exercise safely but also how to manage our physical and psychological health generally when living with cardiomyopathy. I believe a holistic approach to cardiac rehabilitation would best meet our needs. I know a lot of cardiac rehabilitation services now offer support and advice around physical health (exercise, diet etc.) and psychological health (adjusting to change, coping strategies. building confidence etc). I think the aim of cardiac rehabilitation should be to empower cardiomyopathy patients to confidently manage their own health day-to-day in collaboration with their medical team. I look forward to seeing future developments in cardiac rehabilitation for cardiomyopathy patients..
>
> - Jenny Taylor, person living with cardiomyopathy

Questions 11-26

11. Are there tests which can predict the risk of getting worse, heart failure and/or sudden death?

12. Can gene therapy be used to prevent cardiomyopathy developing in people at risk or to treat people with symptoms?

13. What is a safe and beneficial level of exercise for adults and children with a genetic risk of cardiomyopathy?

14. Which people with cardiomyopathy benefit most from an ICD (implantable cardioverter defibrillator)?

15. What is a safe and beneficial level of physical activity or exercise for people with cardiomyopathy?

16. What genetic and environmental factors influence the risk of getting worse, heart failure and/ or sudden death?

17. What are the best ways for people to monitor their symptoms at home and to know when to seek medical help?

18. What are the long-term side-effects of treatment? Can drugs with fewer side-effects be developed?

19. What happens to people with cardiomyopathy as they get older? How does the condition change over time?

20. Can drug treatment reverse changes to the heart to a point where people can safely stop their medication?

21. Can stem cells be used to repair or restore damaged heart muscle in people with cardiomyopathy?

22. How is cardiomyopathy linked to atrial fibrillation (an irregular and fast heartbeat)?

23. What are the best ways to treat heart failure in people with cardiomyopathy?

24. What causes fatigue in people with cardiomyopathy and how is it best treated and managed?

25. What lifestyle changes help avoid getting worse, and/or reduce their risk of heart failure and sudden death?

26. What are the best ways to treat and manage breathlessness in people with cardiomyopathy?

Next steps

Cardiomyopathy UK is working with partners on the PSP and members of the Steering Group to disseminate the priorities for future research. **This includes:**

- Communicating the research priorities in newsletters, email communications and on social media.
- Submitting abstracts to relevant conferences to have posters or presentations.
- Submitting abstracts to relevant journals.
- An opinion piece in a journal.
- Holding events with researchers.
- Identifying research finding opportunities to secure funding to conduct research.

It is most important that the research takes place and people affected by cardiomyopathy get the answers to the questions they have. As part of these plans, Cardiomyopathy UK is in the process of becoming a research funding charity.

Who was involved?

Cardiomyopathy UK led the cardiomyopathy future research priorities partnership with the James Lind Alliance.

PSP Lead: Laura Cook (with Charlotte Gallagher as the initial lead and Katharine McIntosh in the interim)

JLA Adviser: Louise Dunford

PSP Coordinator: Beckie Gray

Information Specialist: Kristina Staley

We would like to thank the members of the Steering Group:
Alison Fielding, Andy Smith, Brian Halliday, Caroline Coats, Ella Field, Helen Alexander, Jayne Partridge, Jenn Zhang, Jenny Moon, Libby Jarman, LaRisha Porter, Marcia Malcolm, Pauline Aiston, Robbie Jones, Ruth Newbury-Ecob, Sandra Miller, Tootie Bueser

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