Cardiomyopathies Matter

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Family matters: tackling inherited cardiomyopathies in the next EU policy mandate

On World Heart Day 2023, the Cardiomyopathies Matter initiative welcomes the recent policy momentum on cardiovascular diseases at European Union level and in many European countries. These actions must address specific challenges faced by people affected by inherited cardiovascular diseases, including cardiomyopathies. Awareness about these conditions, and their familial nature, is often low – underlining the importance of the focus of World Heart Day on improving knowledge. Here we explain why family matters when it comes to improving diagnosis, care and support for these little-known but highly impactful conditions.

Cardiomyopathies – a group of diseases affecting the heart muscle – affect around 1.5 million people in the European Union (EU), UK and Norway. Cardiomyopathies can occur at any age and have a substantial impact on the lives of patients and their families and carers. They often lead to heart failure, which can be incapacitating, and can also cause abnormal heart rhythms, which can be life-threatening. Cardiomyopathies can significantly impair patients' and family members' quality of life (QoL) by limiting patients' activities in daily life and impairing psychological wellbeing. Their impact can also affect employment, finances and education. Costs to healthcare systems and society are often high, especially when patients may need to be hospitalised.[1]

Cardiovascular diseases are the leading cause of death across Europe[2] and are currently the focus of health policy attention at national and EU level. This action is critical to improving population health, achieving the United Nations Sustainability Development Goal Target 3.4 on non-communicable diseases, and improving health systems' resilience and sustainability. While disease prevention is rightly a major focus, non-preventable inherited cardiovascular diseases are often overlooked, despite their impact. Major forms of cardiomyopathy are indeed often, or usually, inherited. A causative genetic variant can be identified in around 20–60% of cases of the main four types of cardiomyopathy.[3]

New clinical guidelines published by the European Society of Cardiology (ESC) offer an unprecedented opportunity to improve the standards of care and support for all patients and families affected by cardiomyopathies across Europe.[4] While guidelines can support healthcare professionals and patients in their decision-making, broader changes to health systems and policies may be needed to ensure that patients have access to optimal care. Here we outline key EU-level policy recommendations necessary, in concert with national actions, to drive early diagnosis of cardiomyopathies, access to expert care, patient and family empowerment, and research and innovation – according to the Cardiomyopathies Matter Roadmap.[1] In the context of the 2024 European Parliament elections, and calls for action from the CVD advocacy community, these recommendations aim to serve as a blueprint for policy action in the 2024–2029 EU mandate.

The Cardiomyopathies Matter Roadmap provides comprehensive recommendations to EU and national policymakers and health system decision-makers to help improve outcomes among patients and families affected by cardiomyopathies.

Optimising patient care and early diagnosis – realising the value of genetics

All patients with cardiomyopathy should have prompt access to well-coordinated, individualised expert care based on the new ESC guidelines – with **'the family at its heart'**.[4] The ESC recommends a multidisciplinary approach where all relevant specialties collaborate as a team, together with patients and carers. The team composition depends on the patient's needs and local service availabilities, but may include general and sub-specialist cardiologists, general practitioners, imaging specialists, specialist nurses, psychologists, geneticists, and pathologists.

In general, patients with inherited cardiomyopathies need life-long monitoring. Genetic counselling and testing are now recommended in several important roles. Here we outline these roles before considering how the EU can support patient access.

> "Policymakers should create the conditions to favour the access to genetic testing and counselling to better understand the disease and predict outcomes."

Matteo Pinciroli, Global Heart Hub and AICARM, Italy

Optimising patient care: Genetic testing in patients with a clinical diagnosis of cardiomyopathy, or suspected cardiomyopathy, can often identify a causative genetic variant. Genetic testing is therefore recommended whenever it can help confirm the diagnosis, inform the prognosis, or guide decisions on treatment and family planning.[4] Genetic counselling by a suitably trained healthcare professional (before and after testing) is essential to help patients and families deal with the many potential implications of these tests. Indeed, genetic counselling is recommended for everyone with an inherited cardiomyopathy, regardless of whether they undergo testing. Looking to the future, genetics could offer a path toward precision medicine for some cardiomyopathies, with gene-editing techniques already under investigation.[5]

Family screening: Early, accurate diagnosis of cardiomyopathy is vital to allow patients access to recommended investigations, care and advice. Cardiomyopathies are often undiagnosed, diagnosed late, or misdiagnosed as something else – the symptoms can even be mistakenly thought to be psychological. Since cardiomyopathies often run in families, all first-degree relatives of patients should be offered screening using clinical tests for the condition; genetic testing should be offered if a disease-causing variant has been identified in the initial patient.[4]

Genetic testing and autopsy are also important after cardiac arrest or sudden cardiac death in young people, to detect any genetic heart disease affecting them and potentially other family members.[6,7]

Family planning: Prenatal or preimplantation genetic testing can also help in family planning, allowing patients to understand and manage the risks of passing their cardiomyopathy to their offspring.

> "Genetic testing is increasingly important in the care of patients and families with cardiomyopathies. It helps identify and confirm the specific cardiomyopathy, which will then help target treatments and advice that you can give to a patient. Genetic tests should be funded, so that there is no economic barrier for patients and their families."

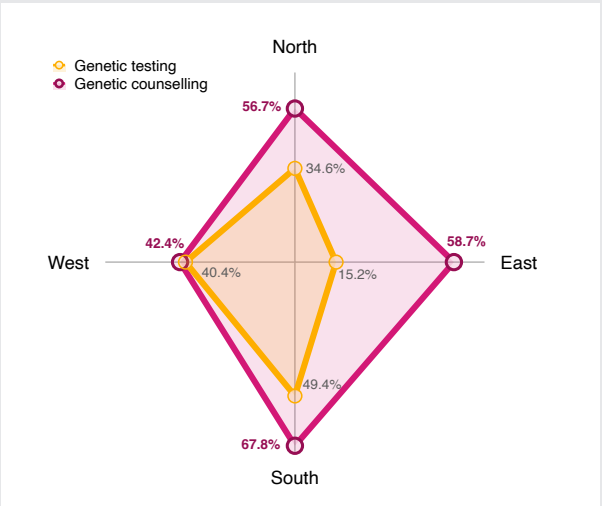
Dr Teofila (Tootie) Bueser, Guy's and St Thomas' Hospital NHS Foundation Trust and North Thames Genomic Medicine Service Alliance, Medicine Service Alliance, London, UK

How should the EU support genetic testing?

Despite guidelines, there are substantial variations between and within countries in the use of, and patients' access to, genetic counselling and testing. For example, in the ESC Cardiomyopathy Registry, genetic testing was performed in 37.3% of patients; this proportion differed between cardiomyopathy types and varied more than three-fold between different regions of Europe (Figure).[8] Genetic counselling was performed in 60.8% in of all patients. These rates could over-estimate typical levels of use, as expert centres are more likely to perform genetic testing than those with less expertise. Genetic testing procedures can also vary between centres within individual countries, further creating disparities in access. There are additional limitations and variations in the availability of prenatal or preimplantation genetic diagnosis.

In England, the governing authority, National Health Service England decided to centralise and fund all genetic testing. Seven regions were created and designated certified genetic laboratories were assigned according to disease expertise. This was followed by mainstream cardiogenetic testing and a whole programme of education in genomic medicine. The aspiration is to offer equal access to high-quality genomic medicine to all.[9]

FIGURE. Genetic counselling and testing are underused across Europe.[8]



Data show the percentage of patients (N=2963 index patients and relatives) within the ESC Cardiomyopathy Registry who underwent genetic testing and genetic counselling in each region of Europe.[8] Individuals were recruited between December 2012 and December 2016. Definitions: North Europe = Denmark, Finland, Great Britain, Lithuania, Sweden; East Europe = Belarus, Czech Republic, Hungary, Poland, Romania; South Europe = FYR Macedonia, Greece, Italy, Portugal, Serbia, Spain, Turkey; West Europe = Austria, France, Germany, Netherlands.

The Cardiomyopathies Matter Roadmap recommends various policy actions to improve diagnosis and care at national level – including the appropriate use of genetic testing and counselling. These include measures to support healthcare professional education and adherence to European clinical guidelines, reimbursement of recommended genetic counselling and tests, and resourcing for genetics laboratory and workforce capacity – since few countries reach workforce levels per capita.[10]

The EU can add value to these actions by:

- Supporting Member States in developing early disease detection through the life-course **European Cardiovascular Health Check** recommended by the multistakeholder European Alliance for Cardiovascular Health (EACH).[11] This umbrella activity includes targeted screening for metabolic and inherited risk factors, detection and precision diagnosis, intensive monitoring and appropriate personalised interventions. It offers a basis for national policy actions to ensure all patients and families have access to genetic counselling and testing, and personalised care, based on the new ESC cardiomyopathy guidelines.
- Further supporting the work of the GUARD Heart European Reference Network **(ERN),** which provides specialised treatment and knowledge exchange about rare and complex cardiovascular diseases – including inherited cardiomyopathies. Actions include driving more ERN integration into national systems and establishing a more sustainable source of funding for the networks.[12]
- Putting forward a **Joint Action** to identify key barriers to the implementation of and adherence to European guidelines for inherited cardiovascular diseases, including cardiomyopathies.
- Encouraging the sharing of national best practices among Member States for access to genetic counselling and testing both for diagnosis and in family screening, e.g. via the Healthier Together initiative and EU best practice portal. These should take account of the roles of specialist nurses and primary care physicians, as well as cardiologists and geneticists. Examples of good practice include national networks in France, Spain, Sweden, and the UK.[1,9]
- Sharing best practices and policies to ensure patients do not face stigma or discrimination following a positive genetic test result for any inherited cardiovascular disease (e.g. in employment, education, or financial services), in accordance with the European Pillar of Social Rights where this applies. Parallel to the development and implementation of any generalised genetic testing programme, the confidentiality of patients' data must be legally guaranteed to avoid stigmatisation. In particular, data must not be used by public or private insurance companies to discriminate against patients, as this could have repercussions, e.g. with respect to their ability to obtain mortgages.
- Further leveraging the **Directive on patients' rights in cross-border healthcare** to facilitate cross-border access to specialist cardiomyopathy care, for example by increasing awareness of patients' rights to cross-border healthcare (both among health professionals and patients and their families/carers), reducing the administrative burden of the procedures, and ensuring a more even implementation of the directive by EU countries.

> "Patients' associations should be promoted and integrated into the healthcare process for rare and very rare cardiac conditions."
>
> **Arbelo E,** et al. 2023 ESC Guidelines for the management of cardiomyopathies[4]

Supporting and empowering patients and families

Patients and families affected by cardiomyopathies play a key role in managing their conditions. They should be empowered as partners in all aspects of their own care and provided with the necessary advice and support.

Cardiomyopathies also can present many challenges for patients and families, including fears about sudden cardiac death and of discrimination following a diagnosis, implications for daily activities (such as sports), and family planning. Holistic psychosocial support is therefore an important part of multidisciplinary cardiomyopathy care.[4]

In addition to promoting genetic counselling, the EU should help here by funding targeted projects, e.g. via EU4Health and sharing best practices on:

- the promotion of self-care and shared decision-making for patients and carers, e.g. via education and health literacy initiatives, improved patient-physician communication, and digital health tools.

> Our 15-year-old son died of sudden cardiac death in his sleep. He had no symptoms and, one night, he went to sleep and never woke up. When I talk about this tragic moment for our family, I talk about a tsunami. Suddenly you hear about a disease that you had never heard of before – a rare disease.

Ester Costafreda, SAMS, Spain

- measures to provide patients' and carers' access to and control over their personal electronic health data, leveraging the **European Health Data Space** (EHDS).
- ensuring equitable access to holistic support services, including psychological and mental health services, family planning and pregnancy advice.
- promoting the role of patient organisations in supporting self-care and in decision-making regarding relevant healthcare policies, services, health technology assessment (HTA), and research priorities.

The ERN GUARD-Heart is already working to empower patients and families through the European Patient Advocacy Group, which is responsible for bringing the voice of patients and their families with rare heart diseases to the Network Board and for supporting the Network with its activities on patient engagement and external communication. Patient representatives can provide the patients´ perspective and thus contribute a lot to the aims and activities of the ERN. Their view adds value and can show clinicians the unmet needs within patient communities. The ERN Guard Heart also give funds for National Meetings to increase awareness about inherited heart diseases.

Despite some high-profile cases, the public may often be unaware of inherited cardiomyopathies and the fact that cardiomyopathies are a leading cause of sudden cardiac death in young people and a leading reason for heart transplantation. The EU should therefore:

- implement an EU-funded awareness campaign to raise public awareness of inherited cardiovascular diseases and associated symptoms and risks.
- promote public training in cardiopulmonary resuscitation (CPR) and the use of automated external defibrillators and adequate and equitable access to these devices (e.g. public buildings, subways, pharmacies, markets, sports centres, schools and universities).

Research and innovation

Further research is vital to address knowledge gaps regarding cardiomyopathies, including the role of genetics, and to drive progress toward precision medicine approaches.

Cardiomyopathies Matter calls for the EU to dedicate targeted research funding via Horizon Europe, building on the current European Innovation Council (EIC) support for a Pathfinder Challenge on cardiogenomics that aims to transform our knowledge of and care for cardiomyopathies. The GEREMY consortium, a project aiming at developing new effective therapies for inherited arrhythmogenic cardiomyopathy and which was recently granted €8 million in funding by Horizon Europe, is a good example of EU-funded research that could tangibly benefit patients.[13]

To provide the foundation for future research, the EU must

- support cardiomyopathy patient registries and ensure the EHDS enables EU-wide harmonisation and utilisation of the real-world data they generate to benefit research and clinical care.
- promote suitable data-sharing policies that facilitate health research – addressing the current fragmentation and divergence of national implementations of the **EU General Data Protection Regulation** (GDPR).[14]

> "At a policy level, it is very important to take the patient's vision into account in any documentation or strategy. Combining the vision of the physician – the cardiologist, who is the expert in treating these types of diseases – and that of the patient, who is the expert in living with these diseases, will give a more complete and holistic vision."

Ester Costafreda, SAMS, Spain

More broadly, the Cardiomyopathies Matter Roadmap also supports recommendations by EACH for a Cardiovascular Health Mission (modelled on the Cancer mission) and a European Cardiovascular Health Data Knowledge Centre integrating existing registries, electronic health record platforms, patient and citizen-generated data, etc.[1,11]

Conclusion

Cardiovascular diseases must be centre-stage in EU-level health policies in the coming years. To achieve this, Cardiomyopathies Matter supports the call by EACH[11] for a dedicated Cardiovascular Health Action Plan at EU level, as well as in each country. This should give specific attention to cardiomyopathies and other inheritable forms of cardiovascular diseases – taking account of the Cardiomyopathies Matter Roadmap.[1]

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Contributors

This publication was developed by the Cardiomyopathies Matter Secretariat (Dentons Global Advisors, Brussels, Belgium) with review and input from the following expert contributors:

Dr Teofila (Tootie) Bueser RN PhD
Director for Nursing & Midwifery, South East Genomic Medicine Service Alliance, Guy's & St Thomas' NHS Foundation Trust; Chief Nurse, North Thames Genomic Medicine Service Alliance, Hosted by Great Ormond Street Hospital NHS Foundation Trust, London, UK

Ester Costafreda
Chair and Board Member, SAMS (Asociación para Pacientes y Familiares con Síndromes Arrítmicos relacionados con la Muerte Súbita), Spain

Tomás Fajardo
President, Cardioalanza, Spain

Dr Pablo Garcia-Pavia
Director, Inherited Cardiac Diseases and Heart Failure Unit, Dept of Cardiology of Hospital Universitario Puerta de Hierro, Madrid, Spain; Member, ERN GUARD-Heart*

Prof. Stefan Janssens
Chairman, Department of Cardiovascular Diseases, University Hospitals Leuven, Leuven, Belgium

Prof. Hugo Katus
Senior Professor, Department of Internal Medicine III (Cardiology, Angiology, and Pulmonology), Heidelberg University Hospital, Heidelberg, Germany; Chair, European Society of Cardiology Innovation Think TankProf. Iacopo Olivotto

Dr Stellan Mörner
Senior Consultant Cardiologist and Associate Professor, Dept of Public Health and Clinical Medicine, Umeå University, Umeå, Sweden; Member, ERN GUARD-Heart

Prof. Iacopo Olivotto
Head of Cardiomyopathy Unit, Department of Experimental and Clinical Medicine, University of Florence; Head of Pediatric and Transitional Cardiology, Meyer University Children Hospital, Florence, Italy

Matteo Pinciroli
Chair, Cardiomyopathy Patient Council, Global Heart Hub; Patient Advocate and Co-Founder, AICARM (Italian Association for the study and research of Cardiomyopathies); Associate Partner, European Patient Advocacy Group, ERN GUARD-Heart*

Sean Taylor
Director of Science and Public Health, World Heart Federation, Geneva, Switzerland

Prof. Maria T. Tome Esteban
Consultant Cardiologist and Professor of Practice Cardiology, Cardiovascular Clinical Academic Group, Inherited Cardiac Condition Service, St George's Hospital NHS Foundation Trust and St George's University of London, London, UK

Patricia Tiramani
Development Executive, Global Heart Hub

Prof. Jean-Noël Trochu
Head, Thorax and Nervous System Institute and Heart Failure and Cardiomyopathies Unit, University Hospital, Nantes, France

Dr Rogier Veltrop
LMNA lab Mechanobiochemistry and Cardiac Cell Signalling, Cardiovascular Research Institute Maastricht, Maastricht University, Maastricht, The Netherlands; Co-Founder LMNAcardiac Foundation

*European Reference Network on Rare and Low Prevalence Complex Diseases of the Heart

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