Cardiomyopathies Matter

A Policy Roadmap to improve cardiomyopathy detection and care in Europe

Cardiomyopathies Matter

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The following organisations have endorsed this report:

Content

Forewords

Dr Juozas Olekas, MEP

Member, MEP Heart Group

Europe must tackle cardiovascular diseases (CVDs) urgently. Together, CVDs kill more people than any other type of disease, and cost EU health systems and societies hundreds of billions of euros each year. Addressing this vast burden is vital to improve outcomes among individual patients and their families, as well as to strengthen health systems and societies.

Many CVDs are broadly known and well-understood – others are not. As a physician and member of the MEP Heart Group in the European Parliament, I believe the Cardiomyopathies Matter Roadmap is timely and much-needed to raise political attention about an under-recognised group of CVDs. Often inherited – hence seldom preventable – cardiomyopathies can increase the risk of early death, cause incapacitating symptoms, and impair the quality of life of patients and families. Costs to healthcare systems and society are often high, especially when patients may need to be hospitalised.

This Roadmap identifies pressing challenges and unmet needs in cardiomyopathy care that can and should be addressed by policy actions both at a national and EU level. Ensuring prompt and accurate diagnosis, promoting guidelines-led care, empowering patients and families, and fostering research and innovation are just a few of the priorities for action.

The European Union's current focus on CVD provides a timely opportunity to achieve this goal. CVDs are prioritised in the Healthier Together initiative and the EU4Health programme, for example. These and other initiatives could be further strengthened by including cardiomyopathies together with other less common and less preventable forms of CVD.

Ultimately however, it is Member States who must take measures to make sure their healthcare systems appropriately recognise this unmet medical need and care for patients with cardiomyopathies. The present Roadmap brings to light the most pressing challenges and recommendations in this context.

Fundamentally, this Roadmap echoes other stakeholders' calls for dedicated Cardiovascular Health Action Plans at both the EU and national level. I support this approach and the call in this Roadmap for specific attention to be given to cardiomyopathies within such action plans. Further, this Roadmap identifies a variety of ways in which the EU could effectively support national actions, in particular by establishing and communicating best practices that Member States could benefit from implementing.

COVID responses shone a light on the importance and effectiveness of EU-supported collaboration amongst Member states to address health needs and promote health system resilience. Now, we must continue this united effort to tackle Europe's number one killer – CVDs – including cardiomyopathies.

Prof. Maria da Graça Carvalho, MEP

Co-chair, MEP Heart Group

Research and innovation are critical to strengthen healthcare systems and consequently improve the standards of care in Europe. Despite affecting more than 60 million Europeans, and causing around a third of all deaths, CVDs attract much less research funding than other disease areas. As co-chair of the MEP Heart Group and member of the Committee on Industry, Research and Energy, I witness first-hand the gaps in this field. In particular, there are substantial research needs in inherited cardiac diseases, such as cardiomyopathies.

The EU can and should contribute to this effort. As an example, the European Innovation Council (EIC) Pathfinder Challenge on cardiogenomics, which aims to transform our knowledge and care of select CVDs including cardiomyopathies, is a step in the right direction but more needs to be done. The proposal for a regulation on the European Health Data Space (EHDS) is an unprecedented opportunity to bolster the use of secondary data for research in cardiomyopathies whilst making sure that patients are empowered and protected throughout the care pathway. The EHDS can reinforce existing cardiovascular registries to foster the sharing of knowledge and data for rarer CVDs. I strongly believe in the promise of the digitalisation of our European healthcare systems that will bring substantial value to the entire CVD community, including cardiomyopathy patients.

The Cardiomyopathies Matter Roadmap sets us on the right path, offering concrete solutions to the various challenges and unmet needs in cardiomyopathies. The EU and national-level recommendations, both building on existing frameworks whilst exploring opportunities in future legislation, account for a comprehensive blueprint of what the future of cardiomyopathy care and research should look like. We must now ensure that these recommendations go one step further and translate into policy changes that benefit patients.

Brando Benifei, MEP

Co-chair, MEP Heart Group

Premature deaths from CVDs have fallen remarkably in recent decades owing to substantial progress in disease prevention and treatment. However, CVDs still have a significant impact on patients and societies. Less common CVDs, such as cardiomyopathies, greatly contribute to this burden and yet are given insufficient attention in EU and national-level initiatives.

Actionable policy changes are urgently needed to alleviate this burden. Among the many challenges persisting in cardiomyopathies, misdiagnosis, late diagnosis and under-diagnosis are a key concern. Younger patients and athletes – who appear healthy – are particularly likely to go undiagnosed, and yet cardiomyopathies are a leading cause of sudden cardiac death in these groups. Improving the diagnosis of cardiomyopathy is crucial so that appropriate treatment and lifestyle advice can be given in a timely manner. This involves broader awareness and professional education in various healthcare settings, systems to ensure rapid referral to specialist cardiologists, and access to recommended diagnostics and assessments.

The present Cardiomyopathies Matter Roadmap is an essential tool to help EU and national policymakers drive positive changes for cardiomyopathy patients. It provides forward-looking recommendations not only with respect to early diagnosis, but also disease management, holistic support, patient empowerment and research and innovation.

As co-chair of the MEP Heart Group, I am proud to be part of this effort and am fully committed to promoting these recommendations in the European Parliament, together with my colleagues. I am convinced that future action to tackle cardiomyopathies will not only benefit patients but also healthcare systems and societies as a whole.

Glossary of abbreviations

ARVC	Arrhythmogenic right ventricular cardiomyopathy
CMR	Cardiac magnetic resonance
CVD	Cardiovascular diseases
DCM	Dilated cardiomyopathy
EACH	European Alliance on Cardiovascular Health
ECG	Electrocardiography
EHDS	European Health Data Space
EIC	European Innovation Council
ERN GUARD-Heart	The European Reference Network on Rare and Low Prevalence Complex Diseases of the Heart (GUARD: Gateway to Uncommon And Rare Diseases of the Heart)
ESC	European Society of Cardiology
EU	European Union
EU4Health	The EU's largest health programme
EUPATI	European Patients' Academy on Therapeutic Innovation
GDPR	General Data Protection Regulation
HTA	Health Technology Assessment
HCM	Hypertrophic cardiomyopathy
ICD	Implantable cardioverter defibrillator
MEP	Member of the European Parliament
NCD	Non-communicable disease
PROM	Patient-reported outcome measure
QoL	Quality of life

Executive summary

1. Introduction

Cardiomyopathies – a group of diseases affecting the heart muscle – have received little policy attention despite conferring substantial disease burdens that could be reduced through better awareness, detection and care.

The Cardiomyopathies Matter Roadmap aims to raise awareness about cardiomyopathies and to ensure they receive the policy attention they warrant from healthcare system decision-makers and influencers at the EU and national levels. To this end it:

- explains cardiomyopathies and their impact on patients, their families, healthcare systems and society
- identifies key challenges and unmet needs throughout the patient care pathway
- provides EU- and national-level policy recommendations to address these challenges and improve patient outcomes.

2. Understanding cardiomyopathies

Cardiomyopathies collectively affect ~1 in 330 people in Europe and can occur at any age. The prevalence of cardiomyopathy, and the associated disease burden, is rising and yet underdiagnosis, misdiagnosis and late diagnosis remain widespread. Some cardiomyopathies are often, or usually, caused by inherited pathogenic genetic mutations: a causative genetic mutation can be identified in around 20–60% of cases of the main four types of cardiomyopathy.

Effects on patient and families

Cardiomyopathies often cause heart failure, which can be incapacitating. They can also cause abnormal heart rhythms, which can be life-threatening.

Cardiomyopathies can significantly impair patients' health-related quality of life by limiting patients' activities in daily life and impairing psychological wellbeing. Their impact can also affect patients' employment, finances and education.

Families and informal caregivers can also be affected, e.g. by limiting their own ability to work owing to care activities, or because of financial pressures.

Healthcare and societal costs

Cardiomyopathies contribute to the enormous costs of cardiovascular diseases, which account for more healthcare spending than any other disease area. Cardiomyopathies confer significant healthcare costs per patient, primarily related to hospitalisations due to complications. The indirect socioeconomic costs to society through lost productivity must also be recognised.

3. Addressing challenges and unmet needs

Policy actions are necessary to address key challenges and unmet needs throughout the care pathway for cardiomyopathies – including diagnosis, disease management, and holistic support. Policies and investments are also needed to foster patient and carer empowerment, research and innovation in this field.

4. Conclusion and call to action

Health system decision-makers at all levels should recognise and address the substantial impact of cardiomyopathies on patients, families, health systems and societies. Unmet needs exist throughout the care pathway, and we call on decision-makers to act on the recommendations provided to tackle these – in collaboration with all relevant stakeholders.

Recommendations:

A dedicated Cardiovascular Health Action Plan should be developed and implemented at EU level and in each country – giving specific attention to cardiomyopathies. The recommendations in this Roadmap – summarised below – should be implemented within or in association with EU and national Cardiovascular Health Action Plans.

National decision-makers should implement:	EU should support via:
Early diagnosis	
• Professional education and decision-support tools (including primary care) • Regular cardiovascular health checks based on a life-course approach • Genetic testing after cardiac arrest or sudden cardiac death (patients aged <50 years) • Family screening for cardiomyopathy among patients' close family members (including genetic testing, counselling and support) • Measures to raise public awareness of heart failure and specific cardiomyopathy manifestations	• EU-funded projects (e.g. EU4Health) • Supporting best practices sharing (Healthier Together initiative and EU best practice portal) • Disseminating data on cardiomyopathy epidemiology and clinical outcomes • EU-funded awareness campaigns
Access to specialist cardiology care	
• Best practices sharing on efficient referral pathways, organisational models, workforce resourcing and digital infrastructures to ensure all patients with cardiomyopathies have prompt access to multidisciplinary care led by cardiologists with expertise in cardiomyopathies	• Supporting best practices sharing • Further leveraging the ERN GUARD-Heart, EHDS, and the Directive on patients' rights in cross-border healthcare to facilitate cross-border access to specialist cardiomyopathy care
Disease management	
• Measures to promote cardiomyopathy guidelines implementation and adherence (education, decision-support tool, audit and benchmarking) • Sufficient resourcing of relevant workforces and infrastructures, and reimbursement of recommended investigations (including cardiac magnetic resonance and genetic testing) and treatment approaches	• Joint action to identify key barriers to guidelines implementation and adherence • Extension of ERN GUARD-Heart actions • Establishing a European Cardiovascular Health Observatory giving specific attention to cardiomyopathies • Leveraging cardiomyopathy registries through the EHDS
Holistic support	
Measures to: • Ensure patients and carers have access to a holistic assessment of cardiomyopathy impact • Ensure access to relevant supportive care, including psychosocial support and reimbursement of psychological intervention • Promote the work of patient organisations	• EU-funded projects (e.g. EU4Health) • Supporting best practices sharing • Considering the emotional impact of cardiomyopathies in current and future EU initiatives on mental health
Empowering patients and carers	
Measures to promote: • Shared decision-making and self-care for patients and carers (via education, improved patient-physician communication, transition care programmes, and disease-specific digital health tools) • Patients' and carers' access to and control over their personal electronic health data • Patients' and carers' involvement in research decision-making (e.g. ethics committees) • Patients and carers' involvement in decision-making regarding relevant healthcare policies, services and technologies	• ERN GUARD-Heart • EU-funded projects (e.g. EU4Health) • Supporting best practices sharing • EHDS • Common EU HTA Framework – improving patient involvement • EUPATI
Research and innovation	
• Support for research and innovation in cardiomyopathies via research projects in defined priority areas	• EU-funded projects • A Cardiovascular Health mission to support the proposed EU Cardiovascular Health Action Plan
• Suitable research infrastructures (e.g. registries)	• Using EHDS to drive EU-wide data harmonisation and utilisation • Establishing a European Cardiovascular Health Data Knowledge Centre
• Suitable data-sharing policies that facilitate health research in collaboration with all relevant stakeholders	• Addressing the current fragmentation and divergence of national implementations of the EU GDPR

Please see p.8 for glossary of abbreviations.

1. Introduction

Cardiomyopathies are a group of diseases affecting the heart muscle.[1] Often inherited, they collectively affect ~1 in 330 people,[2] corresponding to around 1.5 million people in the European Union (EU), UK and Norway.

Cardiovascular disease (CVD) is the number one killer in Europe[3] and a rising focus of EU health policy attention. CVD is a priority strand within the 'Healthier Together' initiative, which aims to help Member States combat non-communicable disease (NCDs).[4] Other relevant EU initiatives include the EU4Health, Horizon Europe (including the European Innovation Council Pathfinder Challenge), and the European Reference Network on Rare and Low Prevalence Complex Diseases of the Heart (ERN GUARD-Heart). The multistakeholder European Alliance on Cardiovascular Health (EACH) has recently called for the EU to go further by creating a dedicated EU Cardiovascular Health Action Plan and Cardiovascular Health Mission.[5]

Generally, CVD policy initiatives tend to focus on heart attacks and stroke, owing to their public prevalence and health impact, and their preventability. Insufficient attention is given to CVDs that are less common and often not preventable (e.g. owing to their inheritance).

Objectives

The Cardiomyopathies Matter Roadmap aims to raise awareness about cardiomyopathies and to ensure they receive the policy attention they warrant from healthcare system decision-makers and influencers at the EU and national levels. To this end it:

- explains cardiomyopathies and their impact on patients, their families, healthcare systems and society
- identifies key challenges and unmet needs throughout the patient care pathway, especially with respect to early diagnosis, optimal disease management, holistic support, patient empowerment, research, and innovation
- provides EU- and national-level policy recommendations to address these challenges and improve patient outcomes.

These diseases nevertheless confer substantial disease burdens that could be reduced through better awareness, detection and care. Indeed, these non-preventable forms of CVD will become an increasing fraction of total CVD, if future CVD prevention efforts are successful. Cardiomyopathies are an important form of CVD that are usually not preventable, but where early diagnosis and risk-based treatment could improve patient outcomes.[6] Only recently, cardiomyopathies are referred to by the Healthier Together and EIC Pathfinder Challenge initiatives. Importantly, these initiatives point to relevant opportunities to address unmet needs through early detection, genetic testing among patients and their families, improving guidelines adherence, and harnessing innovation.[4,7]

> *"Living with cardiomyopathy is something that accompanies you for the rest of your life, and although there are times when it is difficult to move forward with it, there is no other option but to hold on."*
>
> *Patient with cardiomyopathy, Spain*

Improving CVD care is particularly important in the aftermath of the COVID-19 pandemic, which significantly disrupted healthcare services,[8,9] increased the societal burden of CVD, and underscored the need to make healthcare systems more resilient and people-centric. Equity is also a key consideration: the EACH initiative has warned of "tremendous inequalities" between and within EU countries in patients' access to appropriate CVD care,[5] and the European Society of Cardiology (ESC) ATLAS survey documented wide variations in access to cardiology services.[10]

Methodology

This Roadmap was developed based on 1) a review of relevant published and grey literature, and 2) input from expert contributors (see page 3) from the fields of cardiology, cardiac nursing, economics, and cardiomyopathy patient advocacy – gained during an online group workshop, individual consultation meetings, and reviews of successive drafts.

2. Understanding cardiomyopathies

2.1 How common are cardiomyopathies?

The four major forms of cardiomyopathies vary in their prevalence – some being relatively common and others being classed as rare diseases (Box 1).

BOX 1. Features of the main types of cardiomyopathy.

Cardiomyopathy	Characteristic[1]	Approx. prevalence in general population[11]	Approx. % of cases in which a genetic mutation is identified[12]
Hypertrophic	Heart muscle thickens	1:500	30–60%
Dilated	Dilated heart with impaired contractile function*	1:250	20–50%
Arrhythmogenic right/ left biventricular	Heart muscle replaced by scar tissue or fat	1:2000–5000	50–60%
Restrictive	Heart muscle stiffens	Extremely rare	Unclear (may be up to 60%)

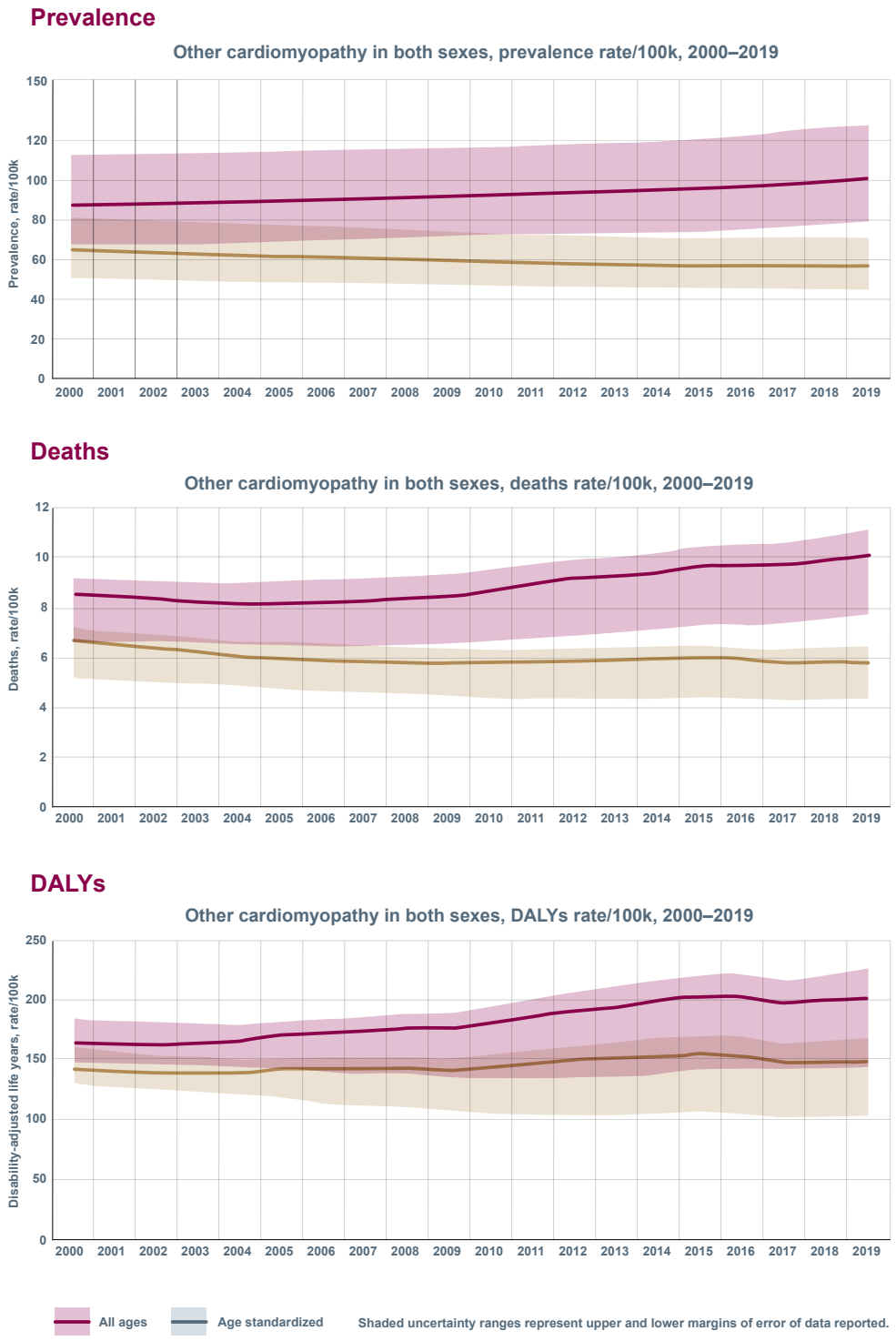
*Can occur towards the end of pregnancy or in the months following delivery (peripartum cardiomyopathy).[13]

Other cardiomyopathies include left ventricular non-compaction cardiomyopathy and Takotsubo cardiomyopathy (which is usually transient and associated with emotional or physical stress).

Overall, the disease burden from cardiomyopathies is increasing, as measured by the number of people diagnosed with cardiomyopathies, and the associated deaths and disability-adjusted life years (DALYs) caused (Figure 1).[14] Cardiomyopathies are widely underdiagnosed and hence these estimates are believed to be the 'tip of the iceberg'.

Cardiomyopathies can occur in patients of all ages. Overall, they are mainly first diagnosed in adults, but some forms – often the most severe – affect children.[12]

FIGURE 1. **Rising disease burden from cardiomyopathy.**



Data show prevalence, deaths and disability-adjusted life years (DALYs) from cardiomyopathy (excluding alcoholic cardiomyopathy) in the European region.[14] Source: Institute for Health Metrics and Evaluation (IHME). Used with permission

2.2 What causes cardiomyopathies?

Some cardiomyopathies are often, or usually, caused by inherited genetic mutations. A causative genetic mutation can be identified in around 20–60% of cases of the main four types of cardiomyopathy (Box 1). Indeed, cardiomyopathies are the most common inherited heart diseases – in the ESC Cardiomyopathy Registry, almost two fifths of cardiomyopathy patients had a documented family history of the disease.[15] Thousands of pathogenic genetic variations have been linked to cardiomyopathies, some of which are consistently associated with a poor prognosis while others have an unpredictable relationship with patient outcomes.[12] In many cases though, definitive pathogenic genetic variants cannot be identified.

Dilated cardiomyopathies can also be caused by certain viral infections, diseases of the endocrine and immune systems, cardiotoxic drugs (including some anti-cancer medicines), toxins, and alcohol, while hypertrophic cardiomyopathies can be caused by various rare diseases, such as amyloidosis and glycogen or lysosomal storage diseases.[1]

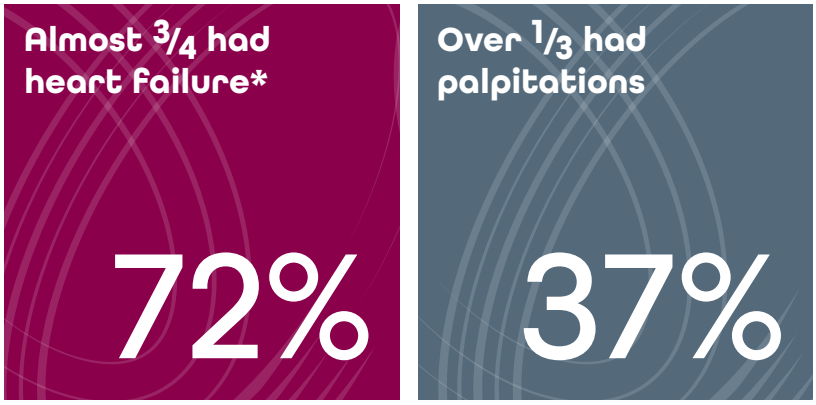
2.3 What are the symptoms and risks for patients?

Cardiomyopathies often reduce the ability of the heart to pump blood around the body, thereby causing other organs to fail. However, their severity and course vary. Most diagnosed patients have cardiac symptoms (such as heart failure and arrythmias) (Figure 2)[15] that may worsen over time. These can impose a significant burden on patients, increase the risk of early cardiovascular death, and necessitate life-long treatment, if available (Box 2). Some other patients may have few or no symptoms and their cardiomyopathy may never be diagnosed, unless it causes a fatal complication.The main symptoms of cardiomyopathies relate to heart failure and abnormal heart rhythms.[15,16]

- **Heart failure** symptoms include fatigue, chest pain, breathlessness and reduced exercise capacity – which can significantly incapacitate patients. Swelling in the ankles, congestion in the abdomen, and loss of appetite can also occur. Heart failure has a profound impact on quality of life, affecting professional, social and family life. Over time, heart failure worsens a patient's prognosis and increases the risk of death.[17,18]
- **Abnormal heart rhythms** (or 'arrythmias', such as ventricular tachycardia, ventricular fibrillation and atrial fibrillation) can cause palpitations, light-headedness, fainting and sudden cardiac death. Abnormal heart rhythms in patients with cardiomyopathy are the second leading cause of cardiac arrests that happen out of hospital – causing 40% of these often-fatal events.[10] Atrial fibrillation increases the risk of blood clots that can cause life-threatening complications, especially stroke.[6,17]

The risk of **sudden cardiac death** from cardiomyopathies is a key concern for patients, in addition to symptoms and functional impairment. It is also an important factor for assessing risk and guiding preventive treatment.[19–21] Cardiomyopathies can cause sudden cardiac death even in undiagnosed individuals (i.e. in whom a cardiac arrest may be the first manifestation of the disease) and in young, active people. Notably, cardiomyopathies are a leading cause of sudden cardiac death in young athletes[22,23] – with high-profile cases prompting public interest and recommendations for screening.[23,24]

FIGURE 2 . Heart failure and palpitations are common among patients with cardiomyopathy.[15]

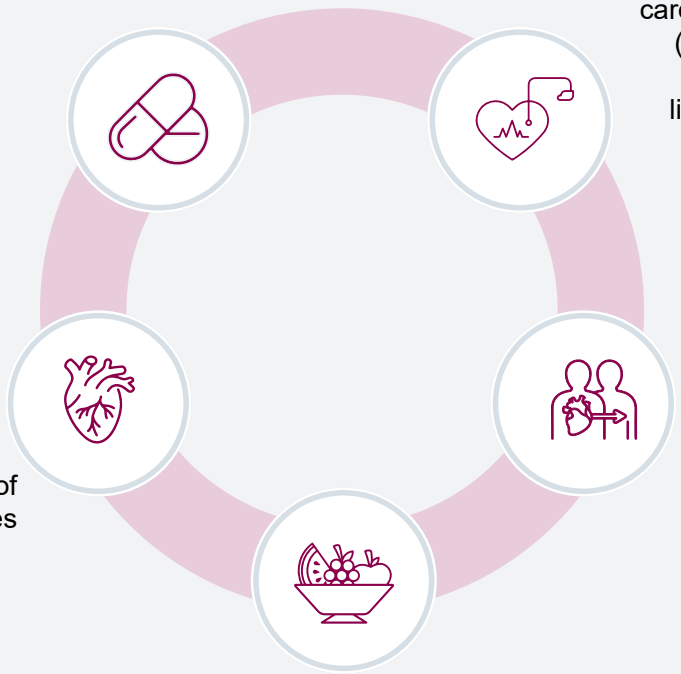


*New York Heart Association Class II–IV

BOX 2. How are cardiomyopathies managed?

Medications
to manage the symptoms of heart failure and abnormal heart rhythms

Surgically implanted devices
i.e. implantable cardioverter defibrillators (ICD) – which may be combined with cardiac resynchronisation devices (pacemakers) to improve heart function – to control life-threatening abnormal heart rhythms and prevent sudden cardiac death.



Surgery or interventional procedures
(e.g. catheter ablation) to relieve obstructive areas of heart muscle, treat arrhythmias or relieve regurgitation of blood through the heart valves

Heart transplantation
or ventricular assist devices are occasionally necessary – cardiomyopathies are the leading reason for heart transplantation[26,27]

Lifestyle measures
to ensure patients stay as healthy as possible (e.g. dietary measures, weight control, tobacco and alcohol limitation, avoidance of competitive sport), and adherence to disease-modifying drugs

'My quality of life did not change, but it is true that my life changed a lot, since from one day to the next there were things that I could no longer do, and it was a very radical change also because I was very young. I had to give up all kinds of sports and it was something that surprised me a lot, since I was very athletic. Luckily my mental state was fine, until the moment I had an ICD implanted, which was when I realized what I had...'

Patient with hypertrophic cardiomyopathy, Spain

2.4 How do cardiomyopathies affect quality of life?

Cardiomyopathies can significantly impair patients' health-related QoL and psychological wellbeing (Figure 3).[28–41] In particular, cardiomyopathies can:

- limit patients' activities in daily life, including work, social and family activities, sports, or other strenuous activities
- lead to anxiety and depression. The psychological impact may relate to existing symptoms, disease progression, the risks of complications – including sudden cardiac death, and genetic inheritance of the disease. Hospitalisation is particularly impactful – especially if unplanned and due to acute complications.[42]

Invasive treatment with implantable devices can also have an important psychosocial impact on patients – despite the benefits.[35,43,44] For example, implanted cardioverter-defibrillators (ICD) can lead to anxiety and depression.[44]

Patients with cardiomyopathy can also experience:[19,47]

- problems in obtaining health or life insurance and mortgages
- out-of-pocket costs, e.g. travel to clinics and aspects of care that may not be covered by insurance (diagnostic tests, recommended supplements, and psychotherapy)
- reduced educational attainment among children and adolescents
- limitations to vocational training and employment, e.g. type of careers pursued, restricted functions at work, and time taken for treatment or rehabilitation, which may result in occupational disability and early retirement.

Families and informal caregivers can also be affected, e.g. by limiting their own ability to work owing to care activities, because of financial pressures,[47] or because of the profound impact of sudden cardiac death on families.

"Having a cardiomyopathy, having an implanted defibrillator, taking medication for arrhythmias, being a carrier of the gene, having limitations in physical activity, regular cardiology check-ups – there are a lot of changes that take some time getting used to, but being able to share the process with other patients helps you not to feel so alone."

Patient with cardiomyopathy, Spain

"Unfortunately, cardiomyopathy often makes its debut in families with the sudden death of a young child, just as it happened to us. It is very frightening and difficult to cope with discovering that this unnatural death is due to a rare disease that can also affect other members of the family, that we are talking about a chronic disease that requires medication and that can degenerate over the years."

Mother whose son died from sudden death due to arrhythmogenic cardiomyopathy, both parents having a genetic variant that may cause the disease, Spain

Did you know?

Heart failure – a key consequence of cardiomyopathy – can cause more severe physical impairment of quality of life than common serious chronic disorders such as chronic lung disease or arthritis.[45] Impairments in QoL independently correlate with mortality and hospital admission rates in patients with heart failure.[46]

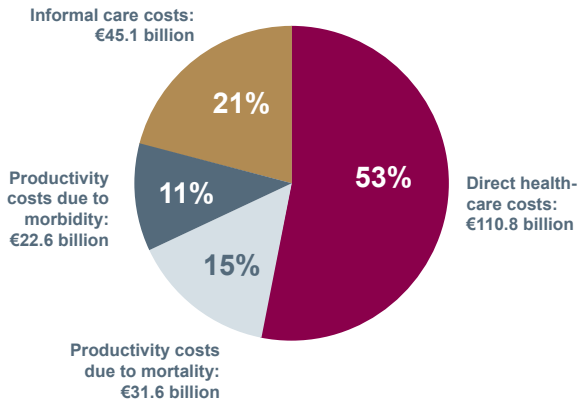
FIGURE 3. Patients with arrhythmogenic right ventricular cardiomyopathy (ARVC) in Germany express their own concerns, fears and experiences regarding the impact of their cardiomyopathy.[48]

2.5 What do cardiomyopathies cost?

Healthcare costs

CVD accounts for more healthcare spending than any other disease area[10] – around €110.8 billion/year across the EU (Figure 4).[49]

FIGURE 4. Cardiovascular diseases cost the EU €210 billion/year in 2015.[49]



Globally, cardiomyopathies make an important contribution to the enormous costs associated with heart failure. Heart failure accounts for 1–2% of healthcare budgets[50] and is the leading cause of hospitalisations in patients >65 years.[51] The contribution of cardiomyopathies to this burden is under-recognised, since these diseases often go undiagnosed in patients with heart failure.[18] Other costs include costs for intensive care and rehabilitation among survivors of cardiac arrest among patients with cardiomyopathies.

Several studies in Europe have demonstrated that cardiomyopathies confer significant healthcare costs, especially due to hospitalisations.

- Nationally in France, patients with cardiomyopathies are estimated to account for 11.3% of hospitalisations for heart failure, 33% of defibrillator implantations, 38% of mechanical circulatory supports, and 51% of all heart transplants.[26]
- In Germany, cardiomyopathies account for 65% of all heart transplants and 70% of all heart transplants in children under 15 years.[27]
- In Italy, around one in three patients with hypertrophic cardiomyopathy (HCM) treated at one cardiomyopathy centre required hospitalisation over a 6-year period – often this was unplanned and due to acute disease-related complications.[42]
- Recent data from Germany[52] and the UK[53] show that obstructive HCM confers a significant economic burden that rises sharply with increasing severity of heart failure, mainly because of inpatient hospital costs.

> *"Strategies aimed at preventing hospitalisations are an important target to reduce the burden of disease"*
>
> *Ciabatti et al.*[42]

Socioeconomic costs

Healthcare costs are only one part of the cost burden of any disease; the indirect socioeconomic costs to society must also be recognised. These include:

- productivity losses from death and illness among working age patients – bearing in mind that cardiomyopathies are often inherited and can affect patients at any age
- costs that carers forgo to provide informal, unpaid care to patients.

These indirect costs have not been quantified for cardiomyopathies in Europe but generally they account for almost half the total cost of CVDs to EU societies (Figure 4).[49] Evidence suggests that ICDs are cost-saving from a societal perspective – the costs of therapy being offset by gains in productivity through lives saved[54] – illustrating the importance of taking societal costs into account when assessing interventions for cardiomyopathies.

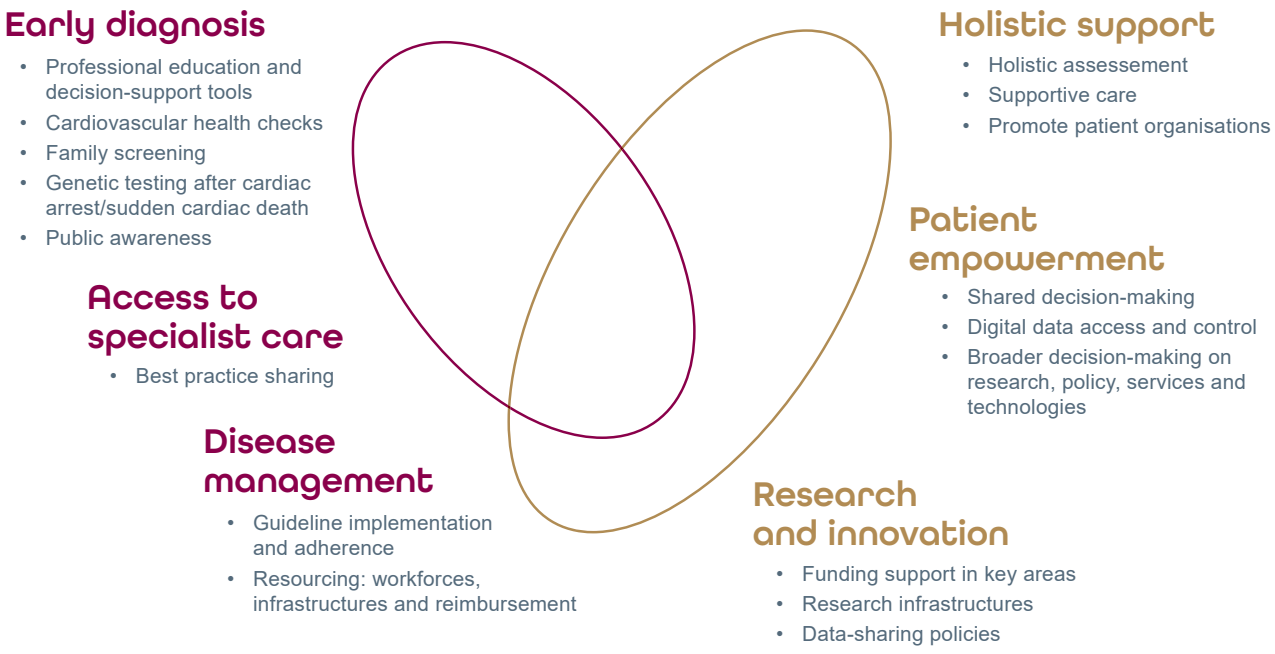
Given the scarcity of published data, it is a research priority to quantify the healthcare resource use and societal costs associated with cardiomyopathies so that decision-makers have access to a more accurate understanding of the true cost of cardiomyopathies when deciding on the introduction of innovative healthcare technologies (also see Section 3.6).

3. Addressing challenges and unmet needs

Policy actions are necessary to address key challenges and unmet needs throughout the care pathway for cardiomyopathies – including diagnosis, disease management, and holistic support. Policies and investments are also needed to foster patient and carer empowerment, research and innovation in this field.

This section offers policy recommendations at EU, national and local level (Figure 5) – and with specific reference to key EU vehicles defined in Box 3, supported by an explanation of the relevant unmet needs and rationale. Many of the recommendations address common issues shared with other chronic, rare, and genetic diseases, giving opportunities for aligned and synergistic policy action.

FIGURE 5. A Cardiovascular Health Action Plan is needed at EU and national levels – giving specific attention to cardiomyopathies based the priority areas shown here.



Overarching recommendation

A dedicated Cardiovascular Health Action Plan (as called for by EACH) should be developed and implemented at EU level and in each country – giving specific attention to cardiomyopathies.

Although current EU policy vehicles can support many of the actions recommended in this Policy Roadmap, a dedicated Cardiovascular Health Action Plan and Cardiovascular Health Mission are needed to tackle Europe's number one killer.[5]

The following recommendations in this Roadmap should be implemented within or in association with EU and national Cardiovascular Health Action Plans.

BOX 3. Key EU policy vehicles

This Roadmap refers to the following EU initiatives relevant to improving outcomes for patients with cardiomyopathies in Europe:

Healthier Together Initiative
This instrument is intended to support EU member states to identify and implement effective policies and actions to reduce the burden of non-communicable diseases. CVD is one of its five main strands, offering opportunities to benefit patients with cardiomyopathies.

The Healthier Together initiative highlights forward-looking actions, policies, guidance and best practices that countries can implement with EU funding.[4] It is supported by the **EU best practice portal**, through which governmental and non-governmental actors can submit proposals for best practices.

Horizon Europe
The largest EU funding programme for research and innovation, Horizon Europe is a key EU instrument to help boost research on cardiomyopathies. Within Horizon Europe, the **European Innovation Council** (EIC) currently supports a Pathfinder Challenge on cardiogenomics to transform our knowledge of and care for cardiomyopathies.

EU4Health
A principal EU funding programme for health, this offers many opportunities to improve cardiomyopathy detection and care.

ERN GUARD-Heart
The European Reference Networks are virtual networks that aim to tackle complex and rare conditions requiring highly specialised treatment and knowledge. ERN GUARD-Heart, the European Reference Network dedicated to rare and low-prevalence complex diseases of the heart, is an important platform for supporting improvements in care for rare cardiomyopathies. The network brings together 44 expert healthcare providers from 16 different member states.

European Health Data Space (EHDS)
The EHDS, together with the General Pharmaceutical Legislation and the Health Emergency Preparedness and Response Authority, is a fundamental element of the European Health Union. It should allow health data to contribute to research and the development of innovative therapies.

EU Directive on patients' rights in cross-border healthcare
This directive is foundational to ERN GUARD-Heart, the EHDS and other initiatives, and hence could be leveraged further to improve cross-border access to specialist cardiomyopathy care.

3.1 Early diagnosis

Recommendations

National decision-makers should implement:	EU should support via:
• Professional education and decision-support tools to promote early clinical suspicion of cardiomyopathies (including in primary care) and facilitate referral to a cardiologist • Regular cardiovascular health checks based on a life-course approach • Genetic testing for cardiomyopathies and other inherited heart diseases after cardiac arrest or sudden cardiac death (patients aged <50 years) • Family screening for cardiomyopathies among patients' close family members according to guidelines – including appropriate resourcing and reimbursement to improve access to genetic testing, counselling and adequate psychosocial support • Measures to raise public awareness of heart failure and specific cardiomyopathy manifestations (including sudden death in athletes)	• EU-funded projects (e.g. EU4Health programme) • Supporting best practices sharing through the Healthier Together initiative and EU best practice portal • Disseminating data on cardiomyopathies epidemiology and clinical outcomes in Member States (e.g. via annual updates) • EU-funded awareness campaigns

Early diagnosis of cardiomyopathies is very important so that appropriate treatment and lifestyle advice can be given.[19,20]

However, in practice, cardiomyopathies are widely undiagnosed[55] or diagnosed late, when significant damage or complications may already be present. Younger patients and athletes are particularly likely to go undiagnosed or misdiagnosed. Because cardiomyopathy patients are often young and appear to be healthy, their symptoms may not be taken seriously. Genetic testing and autopsy are essential after cardiac arrest or sudden cardiac death in young patients, to detect or rule out an underlying cardiomyopathy or other genetic heart disease.[21,25]

Supporting early detection by physicians

Cardiomyopathies may be encountered for the first time in various healthcare settings and specialties. Therefore knowledge and awareness are required broadly among healthcare professionals (e.g. physicians, nurses, pharmacists and psychologists) in order that the disease is recognised or suspected at the earliest opportunity.

> *"I was sent to a psychiatrist to get my (arrythmia) problems under control! I was told that I have a mental illness. Possible differential diagnoses (including a cardio-myopathy) were not taken into account."*
>
> *Patients with ARVC, Germany*[48]

In particular, primary care physicians (general practitioners) and paediatricians are typically the first doctors to see patients who may have an underlying and unrecognised cardiomyopathy. However, most primary care physicians have limited experience in diagnosing and managing cardiomyopathies. Indeed, evidence from Sweden suggests that knowledge gaps may exist among physicians and nurses even in internal medicine and cardiology clinics.[56] Moreover, the symptoms of cardiomyopathies are often non-specific and can be mistaken for other conditions. Now, 'long COVID' may complicate this situation.[57] As a result, cardiomyopathies can be misdiagnosed and patients' referral to cardiologist delayed.

Professional education and decision-support tools to promote early clinical suspicion of cardiomyopathies, including by primary care physicians, are therefore important to facilitate referral to a cardiologist.

A yearly health check in primary care (including a thorough family history) could help improve early cardiomyopathy identification in specific groups at risk, and this has been outlined in the Healthier Together Initiative.[4] EACH also calls for a life-course approach for screening of metabolic and inherited risk-factors, detection, and precision diagnosis at birth (which should include newborn screening for inherited metabolic diseases), throughout childhood, and at specific junctures in adulthood under a European Cardiovascular Health Check.[5] This should include families affected by inherited cardiomyopathies.

Family screening

When patients are diagnosed with a cardiomyopathy, the construction of a pedigree to assess familial disease and screening of close family members is recommended to allow early detection of additional cases. Screening may combine clinical and genetic tests – the latter are discussed further in Section 3.3.[12,25,58] However, despite its importance, the use of familial screening varies across Europe.[15,59,60]

Where screening is available, measures are needed to optimise uptake rates among relatives – such as the provision of appropriate information by healthcare providers.[58] Primary care physicians have important role in encouraging screening, because of their close connection to the families. Genetic testing also requires expert interpretation, genetic counselling and psychosocial support. Systems should be established to ensure that individuals found to have pathogenic genetic variants causing cardiomyopathies are followed up, according to guidelines.[19,20,25]

The Healthier Together initiative heads in this direction by recommending that Member States implement screening or regular medical follow-up of genetically burdened families. The initiative also recommends improving the early detection of structural heart diseases (which include cardiomyopathies).[4]

"My doctor had never seen an ARVC patient before!"

Patients with ARVC, Germany[48]

"Genetic counselling and testing were performed in a substantial proportion of patients but less often than recommended by European guidelines."

Heliö et al. 2020[60]

3.2 Access to specialist cardiology care

Recommendations

National decision-makers should implement:	EU should support via:
• Best practices sharing within and between Member States on efficient referral pathways, organisational models, workforce resourcing and supportive digital infrastructures to ensure all patients with cardiomyopathies have prompt access to multidisciplinary care led by cardiologists with expertise in cardiomyopathies	• Supporting best practices sharing through the Healthier Together initiative & EU best practice portal • Further leveraging the ERN GUARD-Heart, European Health Data Space (EHDS), and the Directive on patients' rights in cross-border healthcare to facilitate cross-border access to specialist cardiomyopathy care

Optimal cardiomyopathy care is led by cardiologists with expertise in cardiomyopathy, but also requires other specialists and services, and hence a multidisciplinary team approach is often recommended.[15,19, 21,25,61,62]

Organisational models for cardiomyopathy care vary.[19] There is some evidence that certain aspects of care (e.g. genetic testing) may be best provided in specialised, multidisciplinary expert centres.[15,25,63–66] However, expert centres might not have capacity to care for all patients with cardiomyopathies and a less centralised approach might be more practical in some healthcare systems.[19] Regardless of the organisational model used:

- all patients and families should be managed according to **internationally agreed guidelines**[19] (see Section 3.3)
- **patients and carers must be empowered** in treatment decision-making based on appropriate information and dialogue with the healthcare team (see Section 3.5).
- care must be **well-coordinated** at the different levels between the various healthcare professionals and providers. For example, patients hospitalised because of cardiomyopathies require coordinated follow-up care after discharge from hospital to reduce the risk of rehospitalisation.

Two solutions to improving care coordination and empowering patients and carers are:

- **Specialist cardiomyopathy or heart failure nurses,** who play an important role both in multidisciplinary care coordination and supporting patients and families.
- Harnessing **digital health tools** to improve the efficiency, accessibility and quality of care.[67,68] For example, **electronic health records** can allow for improved information sharing and care coordination. **Telemedicine services** and smart wearable technologies can support many functions, including home monitoring, triaging and rehabilitation and patient self-care,[69,70] although they are not a default substitute for in-person consultations. Unlocking the potential of digital health tools requires investment in infrastructures (e.g. data science centres) and training, and suitable reimbursement frameworks.

The EU can play a role in supporting patient access to specialist care by

- supporting best practices sharing
- facilitating access to cross-border care via the ERN GUARD-Heart and the EHDS – the latter allowing data-enabled continuity of care and cross-border reimbursement for patients who access care in another. The EU Directive on patients' rights in cross-border healthcare was foundational to these and other initiatives, and hence could be leveraged further to improve cross-border access to specialist cardiomyopathy care.[71]

BOX 4. Expert centre networks: best practice examples in France and Spain

France

Cardiogen national referral network

In 2014, the French Ministry of Health established the Cardiogen national referral network for hereditary or rare heart diseases, including cardiomyopathies (www.filiere-cardiogen.fr). Funded mainly by the Ministry, the network provides funding (e.g. for specialist nurses, genetic testing and counselling) to 64 accredited centres across France, including 21 centres entirely dedicated to cardiomyopathies.

Cardiogen's mission is to 1) provide recommendations and educational material, 2) develop clinical research and create an effective database, 3) foster communications on rare or hereditary heart diseases.

The project is regarded as having improved the overall standard of care for patients with cardiomyopathies in France and has provided patients and healthcare professionals free access to:

- Education targeted both to patients and healthcare professionals
- A comprehensive database to facilitate referred medical care, allowing patient data from referral and competence centres to be aggregated in a unique patient medical file
- Contact details of relevant healthcare professionals, institutions and patient organisations
- A psychology resource centre to facilitate patient access to psychological support
- A free monthly online consultation with patients and families to share feedback on living with rare or hereditary heart diseases.

Spain

CSUR: Spanish National Health Service's Centres, Services and Units of Reference

The Spanish Ministry of Health grants the accreditation of Centres, Services and Units of Reference (CSUR) to centres complying with a series of standards, such as consideration of patients' rights, the implementation of a quality assurance program, or the preparation of an annual audit plan.[72,73]

There are currently seven CSURs in inherited cardiac diseases. These CSURs must demonstrate extensive knowledge and experience in the management of these cardiac diseases and prove that they have the necessary equipment and personnel to provide patients with quality care. Moreover, they must:

- Cover the entire national territory and attend to all patients under equal conditions regardless of their place of residence
- Provide multidisciplinary team care: health care, support for diagnostic confirmation, define therapeutic and follow-up strategies and act as a consultant for the clinical units that routinely look after these patients
- Ensure continuity of care between each stage of the patient's life and between care levels
- Evaluate the results
- Provide training for other professionals.

3.3 Disease management

Recommendations

National decision-makers should implement:	EU should support via:
• Measures to promote cardiomyopathy guidelines implementation and adherence via ongoing education and decision-support tools, and implement practice audit and benchmarking processes according to harmonised standards • Sufficient resourcing of relevant workforces and infrastructures and reimbursement of recommended investigations (including cardiac magnetic resonance and genetic testing) and treatment approaches (medical, interventional and surgical)	• A joint action among Member States (funded by EU4Health programme) to identify key barriers to CVD guidelines implementation and adherence, including cardiomyopathies • Extension of ERN GUARD-Heart actions to improve delivery of high-quality, accessible and cost-effective healthcare – e.g. via benchmarking, best practice sharing, and data sharing • Establishing a European Cardiovascular Health Observatory giving specific attention to cardiomyopathies to help share good practices and support their implementation via national cardiovascular health action plans as recommended by EACH[5] • Leveraging cardiomyopathy registries through the European Health Data Space to share comparable data to aid benchmarking and research

Investigations

Various investigations are recommended to confirm the diagnosis of cardiomyopathies, assess risks, and personalise treatment.[12,19,20,74] However, recent research by the ESC has revealed some gaps in the investigations done in practice.[6,15,60,75]

Highly technological options are available today. However, clinical suspicion is essential before patients are referred to sophisticated tests. Vague indications for tests will only be followed by vague (and perhaps unnecessary) healthcare responses, and this may waste resources. Thus, the gatekeepers of these diagnostic pathways should be clinicians with specific experience in cardiomyopathies.

> *"The results … highlight the need for implemented guidelines on the recommended tests for the evaluation and follow-up of patients with cardiomyopathies."*
>
> *Gimeno et al.*[6]

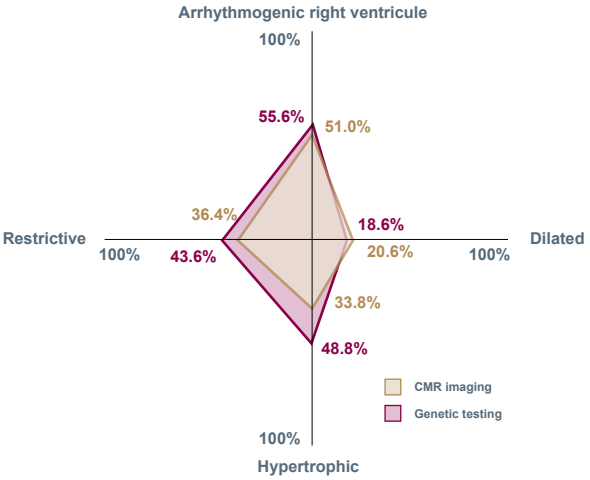
> *"An improvement regarding access, training, and reimbursement is necessary to provide wider application of CMR in diagnosis."*
>
> *Mizia-Stec et al.*[75]

Imaging

Cardiac magnetic resonance (CMR) plays an important role in cardiomyopathy care,[19,20] yet only 29.4% of patients in the multinational ESC Cardiomyopathy Registry underwent CMR at baseline or within 1 year.[75] CMR was underused in all cardiomyopathies (Figure 6). Its use also varied considerably accross Europe (Figure 7), even though these were mostly tertiary centres and teaching hospitals.[75] The underuse of CMR likely relates to local restraints, i.e. relatively high costs, incomplete reimbursement, limited access to dedicated cardiac scanners, and a lack of skilled staff.

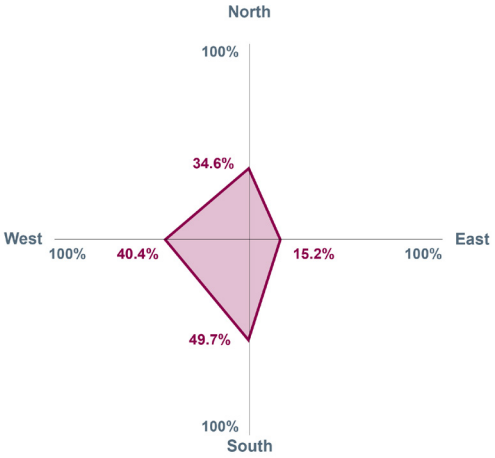
The use of other clinical tests such as Holter electrocardiography (ECG) and exercise tests also varied.[6,15]

FIGURE 6. Cardiac magnetic resonance (CMR) imaging and genetic testing are underused – to varying degrees – in all major cardiomyopathy types.



Data show the percentage of patients within the ESC Cardiomyopathy Registry who underwent baseline CMR (N=3208)[75] or genetic testing (N=2963 index patients),[60] according to the type of cardiomyopathy.

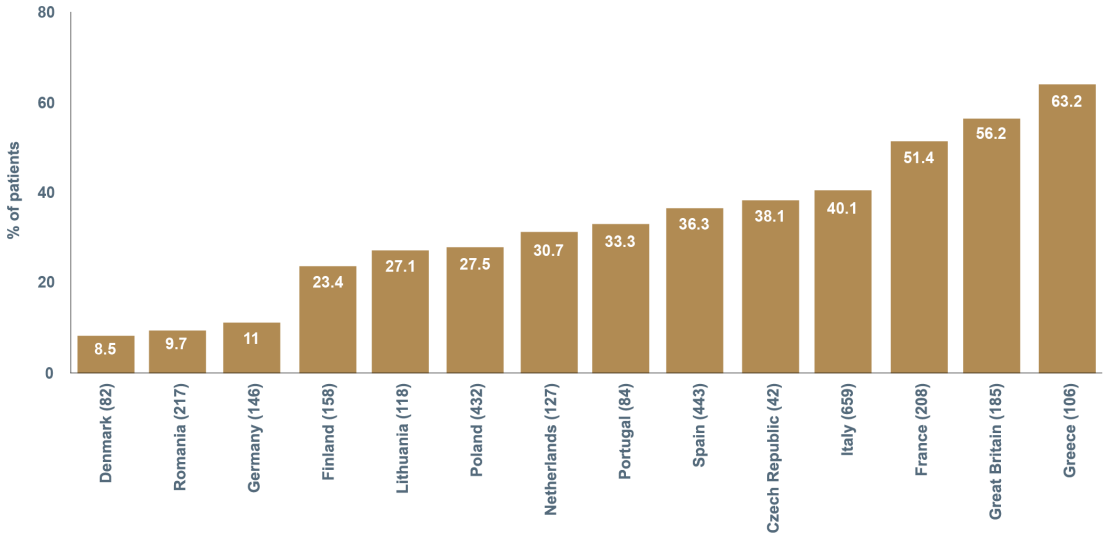
FIGURE 8. The use of genetic testing varies across Europe.



Data show the percentage of patients (N=2963 index patients and relatives) within the ESC Cardiomyopathy Registry who underwent genetic testing in each region of Europe.[60]*

*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.

FIGURE 7. Use of cardiac magnetic resonance imaging varies across Europe.



Data show the percentage of patients within the ESC Cardiomyopathy Registry who underwent CMR at baseline or within 12 months.[75] Bracketed values = total number of patients studied.

Genetic testing

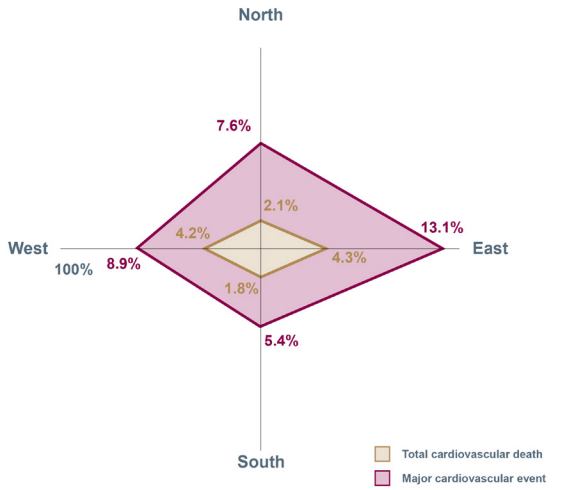
Genetic testing in patients is often useful and recommended to confirm the cause of cardiomyopathy, clarify the prognosis and risk of complications, and guide management.[12,19,20,25,58,74] It can also assist family planning decisions, and support research efforts.

Genetic testing is also important in screening for inherited cardiomyopathies among the close family members of patients (Section 3.1). Genetic testing of family members can offer long-term cost-effectiveness benefits by excluding the presence of mutations causing cardiomyopathies – thereby avoiding the need for serial clinical follow-up in these family members.[58,76,77]

Genetic testing requires qualified laboratory services and expert interpretation, together with counselling to help patients and families deal with the psychological, social, professional, ethical, and legal implications.[12,25,58] Whenever genetic variants classified as being of 'uncertain significance' are identified in patients these should be re-evaluated periodically because our knowledge about their clinical significance might have changed.[12,25]

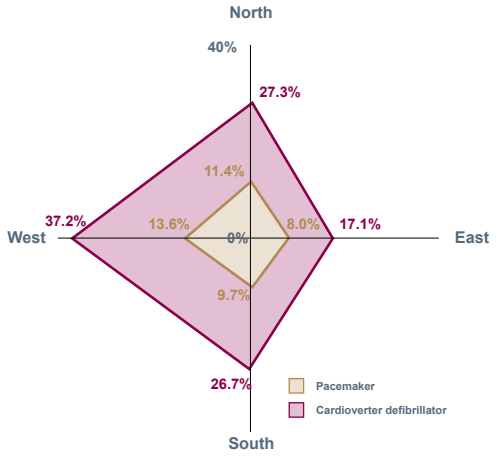
Despite its important role, the use of genetic testing is suboptimal and variable across Europe. For example, in the ESC Registry, genetic testing was performed in 37.3% of all patients, varying between cardiomyopathy types (Figure 6) and European regions (Figure 8).[60] Genetic counselling was performed in 60.8% of all patients, but this varied between different forms of cardiomyopathies (39.2–75.4%).

FIGURE 9. Patients with cardiomyopathy remain at risk for major cardiovascular events and death.



Data show rates of major cardiovascular events (cardiovascular death or urgent hospital admission for cardiac reasons) and total cardiovascular death at 1-year follow-up among patients with cardiomyopathy in the different European regions represented in the ESC Cardiomyopathy Registry.[6]

FIGURE 10. Practice variations exist across Europe in cardiomyopathy therapy.



Data show the percentage of patients in whom a cardioverter defibrillator or pacemaker was implanted at baseline within the ESC Cardiomyopathy Registry in each region of Europe.[15*]

Treatment

European and national guidelines exist for the treatment of cardiomyopathies and related heart failure and arrythmias[19,20,25,58,74] but these require updating and important challenges remain in cardiomyopathy treatment.

1/ Outcomes remain suboptimal and variable – many patients continue to experience heart failure and arrhythmias and are at increased risk of death.[6,17,78] In the ESC Registry, major cardiovascular events occurred in 7.9% of patients within 1 year of follow-up; this rate varied from 5.4% in Southern Europe to 13.1% in Eastern Europe (Figure 9).[6] An adverse outcome often results from a combination of late diagnosis and severe disease course.

2/ Practice variations exist in therapy – e.g. in the use of ICDs, cardiac resynchronization devices, pacemakers, catheter ablation and treatment to prevent blood clots (Figure 10).[6,10,15] Gaps between professional society recommendations and clinical practice, both for investigations and treatment, need to be better understood and addressed,[75] taking account of economical or structural/organizational factors.[15] Knowledge gaps may compromise the provision of care based on up-to-date evidence-based guidelines.[56]

The ERN GUARD-Heart should be supported to extend its actions to improve delivery of high-quality, accessible and cost-effective healthcare – e.g. via benchmarking, best practice sharing, and data sharing. EACH has also recommended a European Cardiovascular Health Observatory to help share good practices and support their implementation via national Cardiovascular Health Action Plans.[5]

Further specific action is also necessary to address disparities that exist across Europe in heart transplantation rates for end-stage cardiomyopathies[6] – along with other organ transplants – as highlighted by a multistakeholder, cross-disease Roadmap published in 2021.[79]

3/ Evidence base is often weak: Important gaps exist in the evidence-base supporting current cardiomyopathy guidelines, including a lack of randomised controlled trials (see Section 3.6).[19] Research is hampered by the many different causes of genetic cardiomyopathies and the lack of well-controlled registries. Generally, the rarer the disease, the more limited is the evidence base.

Some cardiomyopathies (e.g. hypertrophic forms) are characterized by a symptomatic burden but low rates of clinical outcomes such as cardiovascular death, which means that very long observation periods are needed to evaluate outcome and assess the value of any therapeutic intervention. Thus, research is hampered by a lack of 'surrogate' measures that can be used in research to predict long-term clinical outcomes.[80]

> *"To progress towards a better prognosis of patients with cardiomyopathies may therefore require more detailed aetiology work-up, refined risk stratification including recent data on MRI and genetics and may suggest more pro-active use of available therapeutics. Another way is probably to promote the development of 'Cardiomyopathies multidisciplinary teams' ... in order to manage the various aspects of these diseases including aetiology-oriented management."*
>
> *Gimeno et al.*[6]

4/ Patient-reported outcome measures (PROM) are under-used: PROM data provide important information about patients' symptoms, functioning and QoL.[39,81] At present, PROM data are not used widely enough to inform decision-making in clinical care, or in the assessment and reimbursement of treatments and services. PROM need to be developed and validated in collaboration with patient organisations and need to be non-burdensome to ensure adherence and support among patients.

5/ Comorbidities need integrated management: Patients with cardiomyopathy often have CVD risk factors and other co-morbidities.[16] Indeed, CVD risk factors (such as hypertension, diabetes mellitus and obesity) are associated with severe forms of cardiomyopathy.[82] Integrated assessment and management of these risk factors and comorbidities should therefore be strongly promoted.

6/ Limited personalisation of treatment: The heterogeneity of cardiomyopathies mandates personalised strategies and better targeted therapies. Therefore, much more research is needed to enhance precision in cardiomyopathy medicine (Section 3.6)

The Healthier Together Initiative recommends that Member States prioritise the management of structural heart diseases (which include cardiomyopathies).[5]

3.4 Holistic support

Recommendations

National decision-makers should implement measures to:	EU should support via:
• Ensure patients and carers have access to a holistic assessment of the impact of cardiomyopathies • Ensure access to relevant supportive care, including psychosocial support and reimbursement of psychological intervention • Promote the work of patient organisations (via signposting, collaboration, social media, funding etc)	• EU-funded projects (e.g. EU4Health programme) • Supporting best practices sharing (via Healthier Together initiative and EU best practice portal) • Considering the emotional impact of cardiomyopathies in current and future EU initiatives on mental health

"Fortunately, there are patient associations that put you in contact with expert patients who help you by sharing their own experiences. They also offer psychological support to help you cope with all these fears and anxieties, sleep disturbances, loss of energy, isolation, and the urge to cry."

Patients with cardiomyopathy, Spain

Cardiomyopathies can have diverse and profound implications on patients' health and QoL. This can create complex needs that can change over time.[41,83]

The needs of patients and carers should be assessed holistically, and addressed by providing access to supportive care services, information and tools. This may include the following aspects:

Using PROMs to assess QoL and wellbeing among patients and carers in clinical practice.[39]

Disease-specific rehabilitation to help ensure patients can function as normally as possible.

Psychological and **mental health** services to patients and carers, and including grief support in cases of sudden cardiac death[21].

Family planning and pregnancy advice. In some cases, preimplantation genetic testing can be used in conjunction with in vitro fertilisation to avoid passing a genetic mutation to offspring. However, this is not yet widely available for all types of cardiomyopathy and patients' access varies between EU countries.[84,85]

Advice and support services to help patients and families manage the impact of cardiomyopathies, for example on their physical, recreational, and daily activities, together with their wellbeing, finances, education, driving and employment.

Strengthening patient organisations, who play an important role in providing information and resources to patients and families, e.g. to support self-care, offer peer support, and to help patients navigate available services.[41]

Palliative care – often neglected, this should be an integral aspect of care according to guidelines, including symptom control and family and carer support.[86]

"It is particularly striking that only 15% of patients were offered psychological support, although 68% would have liked it."

Ruth Biller (Chair), ARVC-Selbsthilfe e.V., Germany

FIGURE 11. Patients with arrhythmogenic right ventricular cardiomyopathy (ARVC) in Germany express their worries and experiences about coping with their cardiomyopathy.[48]

3.5 Empowering patients and carers

Recommendations

National decision-makers should implement measures to:	EU should support via:
• Promote shared decision-making and self-care for patients with cardiomyopathies and carers throughout their care pathway via: educational initiatives, improved patient-physician communication within clinics, transition care programmes, and disease-specific digital health tools • Improve patients' and carers' access to, and control over, their personal electronic health data • Strengthen patients' and carers' involvement in research decision-making (e.g. priorities and ethics committees) • Patients' and carers' involvement in broader decision-making regarding relevant healthcare policies, services and technologies	• Vehicles that promote patient education, ERN GUARD-Heart, European Patients' Academy on Therapeutic Innovation • Specific EU-funded projects (e.g. EU4Health programme) • Supporting best practices sharing via the Healthier Together initiative and EU best practice portal • Leveraging the European Health Data Space to improve data access • Using the new system set up by the EU Health Technology Assessment (HTA) Regulation to help improve patient involvement in HTA

In general patients want to live as normal a life as possible, and to be treated as respected partners in their care. Patient empowerment can be defined as a "process that helps people gain control over their own lives and increases their capacity to act on issues that they themselves define as important".[87] Promoting shared decision-making, self-care and health literacy are interrelated approaches to help achieve these.

Shared decision-making means that clinicians and patients jointly make health decisions after discussing options, potential benefits and harms, and considering the patient's values and preferences[88] – such as in the decision about the implantation of an ICD.[25]

Self-care includes patient and carers' medication adherence, diet and lifestyle measures, symptom monitoring and responses to any deterioration.[89]

Promoting shared decision-making and self-care throughout cardiomyopathy care are important to patient-centred care and improving patient outcomes. For decision-makers this entails promoting:

Health literacy via educational initiatives to ensure patients with cardiomyopathies and carers have access to understandable information (e.g. including via patient-friendly versions of clinical guidelines). The work of the ERN GUARD-Heart could be leveraged further at national level, since this network already provides information for patients in various EU languages and organises educational meetings for patients, families and associations. Social media should be employed where it adds value in educating patients and families.

Improved patient-physician communication between healthcare professionals and patients and carers, including adequate time and opportunity in consultations.[83]

Transition care programmes: Adolescent patients with cardiomyopathies need particular support during the transition between paediatric and adult care, as they take on a greater role in self-care. European and international guidelines on transition care exist,[90] though not specifically for cardiomyopathy patients. In families affected by cardiomyopathies, adults and children or adolescents with the disease should ideally be followed by the same physician.

Digital health tools have the potential to improve self-care[91] and should be leveraged specifically in cardiomyopathies. The Healthier Together initiative showcases inspiring best practices for using digital tools to empower patients and carers – facilitating shared decision-making, promoting self-management and providing access to up-to-date evidence-based guidelines.[5] The EHDS should also be leveraged to empower individuals through increased digital access to and control of their electronic personal health data.

Patient organisations, in their roles in providing information and support and advocating toward healthcare systems improvements, including patient-centred care.

Empowering patients and carers affected by cardiomyopathies also means strengthening their role in decision-making on research (e.g. regarding research priorities and in clinical study ethics committees) and their involvement in broader decision-making regarding relevant healthcare policies, services and technologies. Notably, the European Patients' Academy on Therapeutic Innovation (EUPATI) provides education that empowers patient representatives to engage and partner with stakeholders in these roles. The common EU framework on health technology assessment (HTA; building on the EUnetHTA initiative) offers an opportunity help strengthen patients' involvement specifically in HTA across Europe.

> *"The associations also help to spread the word by organizing conferences for patients and doctors specializing in family cardiopathies and by organizing training courses for a cardioprotected population."*
>
> *Patients with cardiomyopathy, Spain*

3.6 Research and innovation

Recommendations

National decision-makers should implement:	EU should support via:
• Support for research and innovation in cardiomyopathies via research projects in defined priority areas (Box 5)	• EU-funded projects, e.g. Horizon Europe programme, including Innovative Health Initiative (IHI), and EIC Pathfinder Challenges • A Cardiovascular Health mission, modelled on the Cancer mission, to support the proposed EU Cardiovascular Health Action Plan and stimulate further research on cardiomyopathies
• Suitable research infrastructures (e.g. registries)	• Using the European Health Data Space to drive EU-wide harmonisation and utilisation of data on cardiomyopathy care • Establishing a European Cardiovascular Health Data Knowledge Centre integrating existing registries, electronic health record platforms, patient and citizen-generated data and related CVD initiatives[5]
• Suitable data-sharing policies that facilitate health research in collaboration with all relevant stakeholders	• Addressing the current fragmentation and divergence of national implementations of the EU General Data Protection Regulation (GDPR)

Research Funding

CVD attracts much less research funding than cancer, even though CVD represents the greatest health challenge in Europe.[10] For example, CVD was not prioritised in Horizon 2020, the previous EU-research programme. Overall, the number of EU-funded projects is far fewer than those in other clinical areas[92] and no programme comparable to the European Beating Cancer Plan exists for CVD. Mobilising research and innovation should be a key part of EU- and national-level actions to improve cardiovascular health in general,[5] specifically to address challenges and unmet needs in cardiomyopathies (Box 4). Targeted funding for cardiomyopathies should be provided through Horizon Europe, including the Innovative Health Initiative and EIC Pathfinder Challenges. Patient organisations should inform decision-making on research priorities.

> *"If there is currently no cure, what about research?"*
>
> *Patient with ARVC, Germany*[48]

BOX 5. Priority areas for cardiomyopathy research and innovation

Disease

- Epidemiology (disease phenotypes and prevalence)
- Mechanisms (genetic, epigenetic and acquired)
- Course (heart failure, arrhythmias, chronic, advanced)

Burden

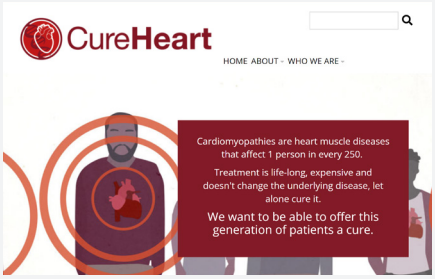
- Patients, carers and families: especially quality of life
- Healthcare systems: healthcare resource use and costs
- Society: indirect socioeconomic costs

Management

- Randomised controlled trials of therapeutic options
- Observational (real-world) registry studies of effectiveness and cost-effectiveness of therapeutic options
- Development and use of patient-reported outcome measures
- Refinement of risk scores (including cardiac magnetic resonance imaging, genetics and digital solutions)
- Innovative diagnostics, prognostics and therapeutics – especially personalised treatment strategies (including DNA and RNA-therapies)
- Innovative care delivery models (e.g. digital health tools and telemedicine)

BOX 6. Case studies in cardiomyopathy research and innovation.

CureHeart: international multistakeholder collaboration in action



The CureHeart project (https://cureheart.web.ox.ac.uk) brings together researchers from the UK, USA and Asia, together with commercial and patient advocacy partners, to develop effective genetic therapies for cardiomyopathies. This project is part of the Big Beat challenge, a global research award to solve the most urgent problems for heart and circulatory diseases patients. CureHeart researchers aim at repairing faulty genes in the heart with a simple injection in the arm. The project is funded by the British Heart Foundation to the tune of £30 million.

Germany: public funding of cardiomyopathy trials

Within the German Research Center for Cardiovascular Disease (DZHK) several interdisciplinary programs are pursued on the genetic causes, epigenetic modulation, and nucleic acid therapies for specific cardiomyopathies.
A unique nationwide registry for patients with cardiomyopathies (TORCH) now includes more than 2500 well-characterized patients.

Infrastructures and policies

National and EU-level investment is critical to implement digital data systems for the collection, analysis and sharing of good-quality, harmonised data – allowing continuous improvement and benchmarking of care practices and patient outcomes.[93]

Registries are particularly important to provide continuous real-world data on the disease burden of cardiomyopathies, treatment patterns and the associated outcomes and costs – extending and expanding the work of the multinational ESC Cardiomyopathy Registry[15] and national cardiomyopathy registries. The ESC EuroHeart initiative is also an important example of a collaborative platform for comparable registry-based real-word data on CVD.

At EU level:

The EHDS must be fully leveraged to support EU-wide harmonisation and utilisation of these data for the benefit of patients with cardiomyopathies.

EACH has recommended a 'European Cardiovascular Health Data Knowledge Centre' integrating existing registries, electronic health record platforms, patient and citizen-generated data and related initiatives in CVD in one large-scale action[5] – which should also encompass cardiomyopathies.

EACH has also recommended a Cardiovascular Health mission, modelled on the Cancer mission, to support the EU Cardiovascular Health Action Plan and stimulate further research on cardiomyopathies. EACH recommend that such a mission 'should place major emphasis on the translation of research outcomes and evidence into cardiovascular health policy and implementation at EU level'.

In addition to infrastructures, suitable data-sharing policies that facilitate health research are vital. The current fragmentation and divergence of national implementations of the EU General Data Protection Regulation (GDPR)[94] must be addressed.[93]

4. Conclusion and call to action

Health system decision-makers at all levels should recognise and address the substantial impact of cardiomyopathies on patients, families, health systems and societies and the unmet needs that exist throughout the care pathway. We call on decision-makers to act on the recommendations provided in this Roadmap – in collaboration with all relevant stakeholders – and we stand ready to engage in dialogue to support these actions.

These recommendations are aligned with, and contributory to, the urgent actions necessary to reduce the burden of CVD and to achieve the UN Sustainability Development Goal Target 3.4 on non-communicable diseases. They also support efforts to improve health systems' resilience and sustainability overall – through a focus on early disease detection, equitable people-centred care, and harnessing innovation and digital health systems toward improved outcomes for patients.

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