

Genetic testing in cardiomyopathy: recommendations for the European Union

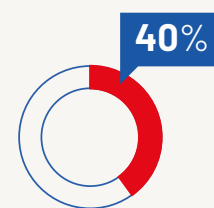
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Why is genetic testing for cardiomyopathy important?

Cardiomyopathy care is at the forefront of cardiovascular genomic innovation. Genetic testing is a recognised standard of care, yet large numbers of people and families across Europe are not able to access it. Cardiomyopathy is a complex heart muscle condition that is partly caused by genetic factors;^{1,2} up to 40% of people with cardiomyopathy have a known family history.³ The discovery of associated genes was a turning point for cardiomyopathy and broader cardiovascular care.^{4,5} Since then, cardiomyopathy has emerged as a pioneer in targeted treatment development in cardiovascular disease area.^{1,2,6,7}



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Genetic testing for cardiomyopathy remains inconsistently implemented, under-resourced and unevenly reimbursed between countries and care settings. This is both a missed clinical opportunity and a systemic weakness that perpetuates avoidable harm across generations. Genetic testing is now recognised as a core component of cardiomyopathy care. International professional societies recommend that all people with confirmed cardiomyopathy be offered genetic testing. This consensus marks a new era and requires health systems to ensure equitable and prompt access to genetic testing.

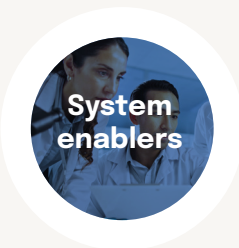
Genetic testing can transform care for people with cardiomyopathy and their families. Specific benefits include:

- **differentiating between cardiovascular conditions** that have similar clinical presentation yet require different treatments^{1,8}
- **identifying undiagnosed relatives** at higher risk of life-threatening events⁹⁻¹¹
- **enabling preventive therapies** in people diagnosed with cardiomyopathy at higher risk of life-threatening events^{1,8}
- **reducing uncertainty** about the cause and progression of cardiomyopathy in people who share the same clinical presentation¹
- **administering the correct dose of targeted treatment** for specific types of cardiomyopathy¹²
- **facilitating reproductive counselling** to support decision-making around family planning.^{1,8}

Beyond clinical benefits, genetic testing also enables health system resources to be allocated effectively and for innovative treatments to be developed. Costs have continued to decline for genetic testing,¹³⁻¹⁵ and studies have shown that it is cost-effective, as it removes the need for regular monitoring of relatives, while ensuring early treatment and reduced hospitalisations.¹⁶⁻¹⁹ Further scientific advances will likely expand the role of genetic testing to assessing increased risk caused by multiple gene variants and predicting how people will respond to treatment, as well as accelerating the discovery of targeted treatments.^{20,21}

Understanding the challenges facing European Union (EU) Member States in preparing health systems for genetic testing in cardiomyopathy

Key actions	Current realities and challenges		
<p>Health systems must finance genetic testing to ensure universal and equal access to innovation and personalised treatment for families with cardiomyopathy, regardless of their ability to pay.</p>	<p>➤ Only 37% of adults with cardiomyopathy receive genetic testing, and just 62% of their relatives do.²²</p>	<p>➤ Four of twelve countries surveyed did not fully reimburse genetic testing for first-degree relatives of people diagnosed with cardiomyopathy.</p>	<p>➤ Four of twelve surveyed countries did not reimburse post-mortem genetic testing, with families having to pay out of pocket or through private health insurance.</p>
<p>Data-sharing systems and infrastructure are needed to support innovation and the development of new targeted treatments.</p>	<p>➤ Experts report difficulties in accessing and sharing data that could support care delivery and research.²³</p>	<p>➤ While most of the surveyed countries had established guidelines for data management, these remain limited and inconsistent.</p>	<p>➤ Cross-border data-sharing is not structured or consistent in the EU, especially for post-mortem data.</p>
<p>Providing genetic education and training for the workforce will allow health systems to deliver quality genetic testing in the future.</p>	<p>➤ Four of twelve countries surveyed reported the availability of structured training and certification for cardiogenetic professionals.</p>	<p>➤ More than one in three experts surveyed did not have access to a dedicated cardiogenetic service or genetic lab.²⁴</p>	
<p>Equal access to high-quality genetic testing can be supported by initiatives such as formal reference centres, care pathways and expert centre networks.</p>	<p>➤ Eight of twelve countries surveyed reported having no national reference network for genetic testing for cardiomyopathy or having a network that needed further development.</p>	<p>➤ While nine of the surveyed countries had national reference centres, three countries reported having none or only having a centre under development.</p>	
<p>Shared decision-making ensures that people with cardiomyopathy understand the rationale for genetic testing and its implications.</p>	<p>➤ Experts report that some centres have not implemented shared decision-making tools and processes, and families are not actively involved.²⁵</p>		
<p>Psychosocial professionals are valuable members of the multidisciplinary team who support people with cardiomyopathy to understand their diagnosis and its impact on their lives.</p>	<p>➤ People with cardiomyopathy often face fears and anxieties after receiving the diagnosis or genetic test results.^{15,26,27}</p>	<p>➤ Experts report that many health systems do not currently provide access to or reimbursement of psychosocial support.^{26,27}</p>	



System enablers



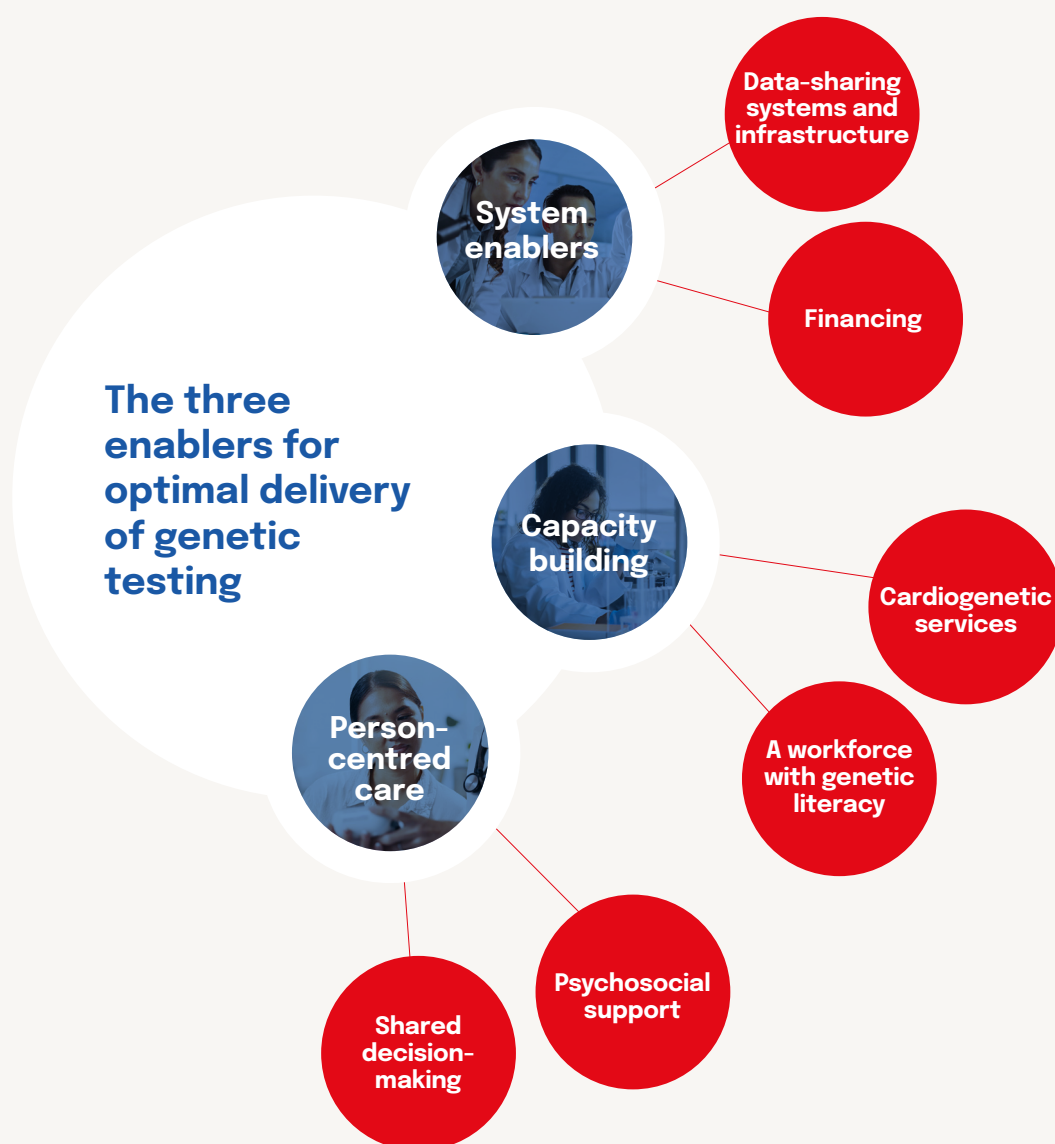
Capacity building



Person-centred care

How can the EU help close the gaps for optimal delivery of genetic testing in cardiomyopathy?

System enablers, capacity building and person-centred care are key to the optimal delivery of genetic testing for cardiomyopathy. These enablers align with the shared EU values of universality, access to good quality care, and equity and solidarity.²⁸ They also support the *European Charter of Patients' Rights* – in particular, the rights to prevention, access, innovation, personalised treatment, privacy and confidentiality, information, consent and free choice.²⁹





System enablers

What are the current EU efforts in improving access to genetic testing and data infrastructure in health systems?

- ▶ The EU has set the legal foundation for the protection of human rights in the implementation of best-practice genetic testing and the use of genetic data.³⁰⁻³²
- ▶ The European +1 Million Genomes initiative aims to improve secure access to genomics and clinical data across Europe, and has developed a maturity model and tools to support countries in assessing the readiness of their health systems.^{33,34}
- ▶ EU4Health has funded Joint Actions Towards the European Health Data Space (TEHDAS) to support Member States in the development of the guidelines needed to implement the European Health Data Space and the secondary use of health data between countries.³⁵

Why do genetic data require targeted action?

While the EU's General Data Protection Regulation covers genetic data, it does not provide sufficient guidance to ensure both privacy and appropriate use of genetic data for research and innovation.^{36,37}

Genetic data require a more complex approach and further efforts are needed to ensure alignment across Member States.

Recommendations for the EU

- ▶ **Address inequalities in access to genetic testing and counselling for rare diseases across Member States by:**
 - issuing a Council Recommendation on equitable access to genetic testing and counselling for rare diseases
 - directing resources via the European Regional Development Fund under the EU Cohesion Policy to support equitable access to genetic services and genetic counselling in countries that currently have limited capacity.
- ▶ **Establish a Joint Action on Genetic Data under EU4Health for the interoperability, integration and regulation of genetic data into health systems, supporting the implementation of the European Health Data Space.**



Capacity building

What are the current EU efforts in building capacity and strengthening expert networks?

- ▶ The European Reference Networks GUARD-Heart and VASCERN support sharing best practices for inherited cardiac conditions and vascular conditions, respectively. They also promote communication and education for healthcare professionals to facilitate access to expert services.^{38,39}
- ▶ The Safe Hearts Plan reinforces the EU's commitment to improving cardiovascular health and includes the development of an EU network on cardiovascular health centres. It also makes specific mention of cardiomyopathy as a concern for screening in high-risk populations and notes the importance of genetics for both personalised medicine and further research.⁴⁰

Where has this worked before?

The Europe's Beating Cancer Plan included the development of a network of comprehensive cancer centres in every Member State to ensure that 90% of people with cancer have access to high-quality care by 2030.⁴¹ The scope of this network includes genetic counselling and testing for inherited cancers.⁴²

Recommendations for the EU

- ▶ Include genetic testing and counselling expertise as part of the proposed EU network on cardiovascular health centres to improve access to early detection and personalised care in inherited cardiac conditions.
- ▶ Invest in the development of genetic literacy and genetic services through the Health System and Healthcare Workforce workstream in the EU4Health annual workplans.



Person-centred care

What are the current EU efforts in embedding person-centred care?

- ▶ The EU is committed to respecting patient rights and implementing person-centred care. The *European Charter of Patients' Rights* establishes key rights that must be respected.²⁹
- ▶ Through the EU, millions of euros have been allocated via Horizon Europe and EU4Health in support of research and piloting new models of care, across basic science, personalised medicine and person-centred approaches.^{43,44}
- ▶ Additionally, the EU has supported Member States through the Technical Support Instrument; in 2023, the flagship project 'Towards person-centred integrated care' focused on mapping organisation of care, developing a roadmap for integrated care and supporting digitalisation.⁴⁵
- ▶ The Safe Hearts Plan proposes a major drive in the field of personalised medicine, in the form of a new Council Recommendation in 2027 to improve the quality and consistency of personalised and integrated care in cardiovascular conditions.⁴⁰

Recommendations for the EU

- ▶ Invest in the development of shared decision-making, communication and digital health tools for genetic conditions in the EU4Health annual work programmes.
- ▶ Ensure the proposed 2027 Council Recommendations includes new guidance in genetic testing, and highlights cardiomyopathy as a pilot area for capacity building and new organisational models of person-centred care.

References

- Arbelo E, Protonotarios A, Gimeno JR, et al. 2023. *Eur Heart J* 44(37): 3503-626
- Wilde AAM, Semsarian C, Márquez MF, et al. 2022. *J Arrhythm* 38(4): 491-553
- Charron P, Elliott PM, Gimeno JR, et al. 2018. *Eur Heart J* 39(20): 1784-93
- Kääb S, Bondue A. 2025. *Eur Heart J* 46(19): 1784-86
- Geisterfer-Lowrance AA, Kass S, Tanigawa G, et al. 1990. *Cell* 62(5): 999-1006
- Maurer MS, Schwartz JH, Gundapaneni B, et al. 2018. *New Eng J Med* 379(11): 1007-16
- Olivotto I, Oreziak A, Barriales-Villa R, et al. 2020. *Lancet* 396(10253): 759-69
- Vogiatzi G, Lazaros G, Oikonomou E, et al. 2022. *World J Cardiol* 14(1): 29-39
- Brodt C, Siegfried JD, Hofmeyer M, et al. 2013. *J Card Fail* 19(4): 233-9
- Lopes LR, Syrris P, Guttman OP, et al. 2015. *Heart* 101(4): 294-301
- Li Q, Gruner C, Chan RH, et al. 2014. *Circ Cardiovasc Gen* 7(4): 416-22
- McGurk KA, Bilgehan N, Ware JS. 2024. *Circulation* 149(23): 1786-88
- Zeljkovic I. 2025. Interview [videoconference]. 22/05/25
- Sepp R. 2025. Interview [videoconference]. 15/06/25
- Biller R. 2023. Interview [teleconference]. 12/05/23
- Wordsworth S, Leal J, Blair E, et al. 2010. *Eur Heart J* 31(8): 926-35
- Sabater-Molina M, Garcia-Molina E, Tovar I, et al. 2013. *Cardiogenetics* 3(1): e5
- Catchpool M, Ramchand J, Martyn M, et al. 2019. *Genet Med* 21(12): 2815-22
- Alfares AA, Kelly MA, McDermott G, et al. 2015. *Genet Med* 17(11): 880-8
- Lairez O, Fournier P, Itier R, et al. 2024. *Presse Med* 53(1): 104223
- Aradhya S, Facio FM, Metz H, et al. 2023. *Am J Med Genet C Semin Med Genet* 193(3): e32057
- Heliö T, Elliott P, Koskenvuo JW, et al. 2020. *ESC Heart Fail* 7(5): 3013-21
- de Backer J. 2025. Interview [videoconference]. 30/05/25
- Zeljko I, Gauthy A, Manninger M, et al. 2024. *EP Europace* 26(9): 1-7
- Kaski JP. 2025. Interview [videoconference]. 17/06/25
- Balan C. 2025. Interview [videoconference]. 26/05/25
- Vasilakis A. 2025. Interview [videoconference]. 03/06/25
- Council Conclusions on Common values and principles in European Union Health Systems. 2006. European Union: 22/06/06
- Active Citizenship Network. 2002. *European Charter of Patients' Rights*
- Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes. 2008. European Union: 27/11/08
- Council of Europe. 2015. <https://rm.coe.int/inf-2015-6-e-dpi-dpn/1680a59c49>
- CM/Rec(2016)8 - Recommendation of the Committee of Ministers to the member States on the processing of personal health-related data for insurance purposes, including data resulting from genetic tests. 2016. European Union: 26/10/16
- European Commission. <https://digital-strategy.ec.europa.eu/en/policies/1-million-genomes?>
- The Beyond 1 Million Genomes Project. <https://b1mg-project.eu/resources/maturity-level-model>
- TEHDAS2. <https://tehdas.eu/project/>
- Rahnasto J. 2023. *J Law Biosci*: 10.1093/jlb/lsad029
- Kuru T. 2021. *Eur Data Protect Law Rev* 1: 45-58
- ERN GUARD-Heart. <https://guardheart.ern-net.eu/ern-guard-heart/missions/>
- VASCERN E. <https://vascern.eu/who-we-are/about-vascern/>
- European Commission. 2025. *The Safe Hearts Plan*.
- EU Network of Comprehensive Cancer Centres. <https://eunetccc2025.eu/>
- CraNE4Health. 2024. https://crane4health.eu/wp-content/uploads/2024/09/CraNE_WP6_D6.1._CCCN-definition-and-interfaces_fin_joint.pdf
- European Commission. https://health.ec.europa.eu/funding/eu4health-programme-2021-2027-vision-healthier-european-union_en
- European Commission. 2025. *Horizon Europe Work Programme 2026-2027*
- European Commission. https://reform-support.ec.europa.eu/towards-person-centred-integrated-care_en

About the International Cardiomyopathy Network

The International Cardiomyopathy Network (ICoN) is a newly established association of medical, scientific and lay stakeholders with the mission of improving the health of people affected by cardiomyopathy and related diseases. ICoN plans to undertake a rolling programme to transform the cardiomyopathy landscape in Europe through clinical education and patient engagement, so that more people with cardiomyopathy can access treatment and support when they need it.

About this document

This document has been developed based on desk research and workshops with key experts in cardiomyopathy. Experts included healthcare professionals with experience in cardiomyopathy and relevant fields, and people living with the condition and their relatives. It also includes findings from the accompanying policy paper *Genetic testing in cardiomyopathy: ensuring our future health systems leave no family behind*.

Coordination, research and drafting were led by Perry Elliott at ICoN and Karolay Lorenty, Ed Harding, Mo Forman and Kasia Trojanowska at The Health Policy Partnership, with design by Catarina Correia Marques. The Steering Committee, as co-authors, closely guided the development of recommendations for the EU.

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