Friday 25 July 2025

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES
Share. Care. Cure.

ERN GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart

ERN GUARD-HEART BIMONTHLY NEWSLETTER YEAR 2025 NUMBER 3

1 September 2025 next board meeting ERN GUARD-Heart

Amsterdam, 18-07-2025

The next ERN GUARD-Heart Board Meeting will take place in Madrid on the 1st of September. Registration is now closed. A total of 43 participants will attend on-site, and 9 will join online. The agenda will be shared by email soon. ♥

ERDERA Survey on Criteria for Prioritising Rare Diseases for Advanced Therapy Development

Amsterdam, 18-07-2025

The ERDERA WP11 team has reached out to all ERNs with an important request: Help prioritize rare diseases for the development of Advanced Therapy Medicinal Products (ATMPs). Your expertise is essential to identify which factors are most important — from unmet medical needs to psychosocial impact and research readiness.

Complete the questionnaire here: http://tiny.cc/ATMP.

It will take approximately 15–20 minutes.

Deadline: 15th August.

Thank you for contributing to this crucial effort! ♥

10th Anniversary of the Heidelberg Institute for Cardiomyopathies

Heidelberg, 04-07-2025

This year marks the 10th anniversary of the Heidelberg Institute for Cardiomyopathies, a decade of remarkable progress, collaboration, and commitment to advancing patient care. Over the past ten years, the field of cardiomyopathies has seen transformative developments. Innovations in diagnostics, personalized therapies, and our understanding of disease mechanisms have reshaped clinical practice. These achievements have been made possible through strong national and international collaboration and the seamless integration of research and clinical care.

To celebrate this milestone, the organizing team warmly invites you to attend an international scientific symposium, bringing together leading experts from around the world.

For program details, please use the following link: https://www.kelcon.de/wp-content/uploads/2025/07/ICH.Heidelb Programm .pdf

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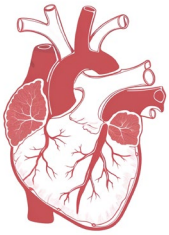


Introduction New ERN GUARD-Heart Network Manager Mari Murel

21 July 2025

I'm Mari Murel, and I'm excited to introduce myself as the new **Network Manager** for ERN Guard-Heart. Although I'll officially start in September, I've already started easing into the role since June by shadowing the Coordination team at **Amsterdam UMC.**

Over the past four years, I've been navigating the world of ERNs through my work as **Coordinating Project Manager for the ERICA project** (the Coordination and Support Action for ERN Research). Alongside ERICA, I've also been closely involved in other ERN-relevant initiatives such as **ERDERA** and **JARDIN**, which aim to strengthen research and data infrastructures across ERNs. I look forward to bringing these connections and opportunities into closer alignment with the goals of the ERN Guard-Heart network.

During my time with ERNs, I've had the privilege of working closely with the **Endo-ERN** network, and I've become increasingly enthusiastic about the unique role ERNs play in improving care for people living with rare diseases. It's been incredibly rewarding to witness the development of these networks—each one finding its rhythm, gaining structure, and building awareness. I've had the unique opportunity to collaborate with several networks—ERN Guard-Heart among them—and I'm now looking forward to focusing my energy and experience on supporting this network's further development. ERN Guard-Heart is already active and growing, and I'm excited to contribute to its journey toward becoming a fully established expert network. Before joining the ERN community, I worked as a **public health and environment researcher** at the **Dutch National Institute for Public Health and the Environment (RIVM)** and as **a policy and research officer** at the **European Partnership for Supervisory Organisations in Health Services and Social Care (EPSO)** in The Hague. These roles gave me valuable experience in managing international consortia, building collaborative partnerships, and organizing cross-border networking activities.

My academic background is in **Environmental Health and Epidemiology**, with a strong focus on sustainability. I'm Dutch, but I was born in Estonia to an Estonian mother and Dutch father, studied in Utrecht, and now live in The Hague with my Swedish partner, Klas. So I already have a very international heart. My Nordic roots have given me a deep love for winter sports—especially cross-country skiing. I'm a certified **NSKIV N3 cross-country ski trainer** and give lessons with **RW Rottemeren.** Since the Netherlands doesn't offer much snow, I train on roller-skis and take every opportunity to travel to Sweden or nearby countries when winter arrives. If you ever want to talk about skiing—or join a lesson—feel free to reach out! I'm always happy to connect over shared topics closed to my heart .

I'll also be participating in the upcoming General Assembly in Madrid, and I'm very much looking forward to meeting many of you there in person! ♥

Who are our ePAG's? Stefan Bassant & Dr. Rogier Veltrop – LMNA Cardiac Diseases Network

Maastricht, 18-07-2025

We are Stefan Bassant and Dr. Rogier Veltrop, co-founders of LMNAcardiac.org—a patient-driven network dedicated to advancing research, connecting the LMNA cardiac community, and advocating for better treatments. Our mission is deeply personal, as we both live with LMNA-related cardiac disease.

Stefan, an entrepreneur and investor from the Netherlands, recognized the urgent need to unite patients with leading researchers and clinicians. His journey began with commissioning research in early 2021 to identify key experts in the field. This led to his connection with Rogier, a molecular cell biologist and LMNA researcher at Maastricht University.

Rogier, also affected by LMNA-related cardiac disease, has undergone a heart transplant and dedicated his scientific career to studying LMNA mechanobiochemistry in 3D cardiac systems at the Cardiovascular research institute MAASTRICHT (CARIM). His work provides crucial insights into how the LMNA mutation affects heart cells, paving the way for potential treatments.

Together, we envisioned LMNAcardiac.org as more than just a website—it's a movement. A place where patients, families, researchers, and clinicians can collaborate, share knowledge, and drive progress. Today, our global network includes experts, clinicians and almost 1.000 LMNA patients worldwide.

We believe that collaboration is the key to accelerating research and developing life-saving therapies. By bridging the gap between patients and scientists, we aim to transform the future of LMNA-related cardiac disease.

If you or a loved one are affected, you are not alone.

Read more and join us at www.lmnacardiac.org and be part of a growing community dedicated to improving lives and finding a cure. ♥

LMNA Cardiac Diseases Network

Dr. Rogier Veltrop (left) and Stefan Bassant (right), cofounders of LMNAcardiac.org

Congratulation to ERN GUARD-Heart ePAG Lorraine McGlinchey

Dublin, 18-07-2025

We are proud to share that Lorraine McGlinchey, ePAG representative, graduated with her Doctorate from Ulster University this June. Her PhD thesis focused on: "Healthcare professionals' knowledge and experience of Inherited Cardiac Arrhythmias (ICA), and their views towards and confidence in caring for individuals with ICA in a surgical environment." A fantastic achievement and a valuable contribution to the field. Congratulations, Lorraine! ♥

Jardin - Introducing the ERN Service Directory

Luxembourgh, 7-2025

As part of JARDIN's commitment to improving the visibility and accessibility of the European Reference Networks (ERNs), we're pleased to highlight a valuable resource developed by the European Commission: the ERN Service Directory.
The European Reference Networks (ERNs) Service Directory is a searchable online platform that provides patients and healthcare professionals across Europe with access to information about the 24 ERNs. This online directory includes details on the different ERNs, their member hospitals, clinical and research centres, and healthcare providers by country, as well as the specific diseases they cover. It helps patients, healthcare professionals, and researchers locate specialised expertise on rare diseases and complex conditions across Europe.

Explore the ERN Service Directory:
https://webgate.ec.europa.eu/ernsd/screen/public?npage=ern_portal.html ♥

New Toolkit & Informative Webinar to Strengthen ERN–Industry Collaboration

Luxembourgh, 07-2025

A new resource has been launched to help bridge the gap between European Reference Networks (ERNs) and life sciences companies, aiming to advance research and care for people living with rare diseases: the Together For Rare Diseases (T4RD) Toolkit. Presented during an informative webinar co-hosted by Together4RD and ERDERA on 23 June 2025 , the Toolkit offers practical guidance for building and managing public-private partnerships in this complex field.
Linked to ERDERA's Work Package 25.4(ERN strategic alignment), the Toolkit supports the development of an "ERN Living Lab" and contributes to ERDERA's broader mission to align research strategies, foster innovation, and accelerate the translation of research into better care. ♥

→ Access the Toolkit **here**
→ **Watch the webinar**

ERDERA's Networking Support Scheme

Luxembourgh, 7-2025

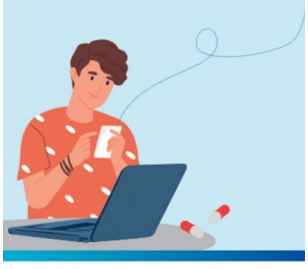
The European Commission's ERDERA Networking Support Scheme is a funding initiative designed to foster collaboration and knowledge exchange on rare diseases and rare cancers across Europe.
This continuously open call invites proposals from researchers, clinicians, patient advocates, and research managers. Eligible events must involve participants from at least three ERDERA countries and focus on rare disease or rare cancer research. Meetings can be held in person or in hybrid formats to maximise accessibility.
Applicants may request up to €30,000 per event to cover costs such as travel, meeting logistics, and digital platforms. The first assessment round will follow the 7 October 2025 deadline, with new rounds every six months until April 2029 or until the budget is fully allocated.
Full details, including guidance notes, evaluation criteria, and the list of eligible countries, are available on the **ERDERA website**. ♥

ERN Youth Panel - Call for Young Voices in Rare Disease Advocacy

Luxembourgh, 07-2025

The European Reference Network Youth Panel is looking for motivated young people (18–25) across Europe who are living with a rare or complex disease, or are siblings of someone who is. This initiative will work across the ERNs, focused on improving care transitions into adulthood and youth engagement in health policy and advocacy!

Interested or know someone that could be? Contact ern.youthpanel@uzgent.be as soon as possible! ♥



See the Flyer

Latest ERN GUARD-Heart Publications

1. Monda E, Biagini E, Blom N, Drago F, Krapels I, Krebsová A, Koubsky K, Khraiche D, Martins E, Merlo M, Michels M, Mizia-Stec K, Mörner S, Peña Peña ML, Planinc I, Robyns T, Rydberg A, Saenen J, Rodríguez Palomares JF, Rutger H, Sarquella Brugada G, Scheirlynck E, Schulze-Bahr E, Tfelt-Hansen J, Wolf CM, Hofman N, Amin AS, Wilde A, Charron P, Limongelli G. Current Management of Transition and Multidisciplinary Care of Patients with Inherited and Rare Cardiomyopathies in Europe: Results of the European Reference Network for Rare and Low Prevalence Complex Diseases of the Heart (ERN GUARD-HEART). Eur Heart J Qual Care Clin Outcomes. 2025 Jul 11:qcaf055. doi: 10.1093/ehjqcco/qcaf055. Epub ahead of print. PMID: 40643001.

2. Norrish G, Hall K, Field E, Cervi E, Boleti O, Ziółkowska L, Olivotto I, Passantino S, Khraiche D, Limongelli G, Weintraub RG, Anastasakis A, Biagini E, Ragni L, Sarquella-Brugada G, Cesar S, Prendiville T, McLeod K, Ilina M, Baban A, Ojala TH, Spentzou G, Bhole V, Gran F, Brown E, Delle Donne G, Khodaghalian B, Fernandez A, Daubeney PEF, Linter K, Kubus P, Uzun O, Bökenkamp R, Raimondi F, Marrone C, Medrano C, Gonzalez-Lopez E, Siles A, Luczak-Wozniak K, Bharucha T, Adwani S, Klaassen S, Castro FJ, Guereta L, Yamazawa H, Sinagra G, Popoiu A, Perin F, Chana B, De Wilde H, Rasmussen TB, Mogensen J, Mathur S, Centeno F, Reinhardt Z, Barriales-Villa R, Kubo T, Felice T, Radulescu C, Schouvey S, Chaker M, Kaski JP. Sex Differences in Children and Adolescents With Hypertrophic Cardiomyopathy. JACC Adv. 2025 Jul 4;4(8):101907. doi: 10.1016/j.jacadv.2025.101907. Epub ahead of print. PMID: 40618618; PMCID: PMC12272437.

3. Nicastro M, Vermeer AMC, Postema PG, Tadros R, Bowling FZ, Aegisdottir HM, Tragante V, Mach L, Postma AV, Lodder EM, van Duijvenboden K, Zwart R, Beekman L, Wu L, Jurgens SJ, van der Zwaag PA, Alders M, Allouba M, Aguib Y, Santome JL, de Una D, Monserrat L, Miranda AMA, Kanemaru K, Cranley J, van Zeggeren IE, Aronica EMA, Ripolone M, Zanotti S, Sveinbjornsson G, Ivarsdottir EV, Hólm H, Guðbjartsson DF, Skúladóttir ÁT, Stefánsson K, Nadauld L, Knowlton KU, Ostrowski SR, Sørensen E, Vesterager Pedersen OB, Ghouse J, Rand SA, Bundgaard H, Ullum H, Erikstrup C, Aagaard B, Bruun MT, Christiansen M, Jensen HK, Carere DA, Cummings CT, Fishler K, Tørring PM, Brusgaard K, Juul TM, Saaby L, Winkel BG, Mogensen J, Fortunato F, Comi GP, Ronchi D, van Tintelen JP, Noseda M, Airola MV, Christiaans I, Wilde AAM, Wilders R, Clur SA, Verkerk AO, Bezzina CR, Lahrouchi N. Bi-allelic variants in POPDC2 cause an autosomal recessive syndrome presenting with cardiac conduction defects and hypertrophic cardiomyopathy. Am J Hum Genet. 2025 Jul 3;112(7):1681-1698. doi: 10.1016/j.ajhg.2025.04.016. Epub 2025 May 22. PMID: 40409267; PMCID: PMC12256823.

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