Wednesday 11 February 2026

ERN GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart

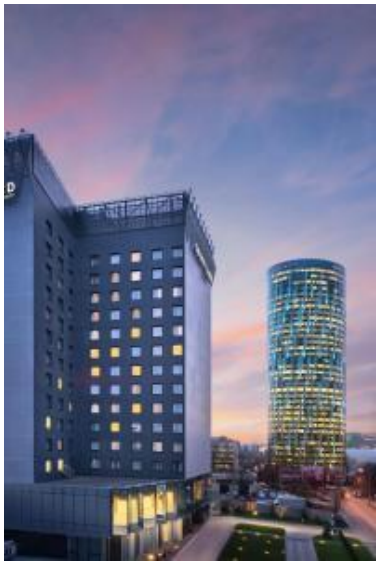
EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES
Share. Care. Cure.

ERN GUARD-HEART BIMONTHLY NEWSLETTER YEAR 2026 NUMBER 1

19th ERN GUARD-Heart Board Meeting in Bucharest

Amsterdam, 05-02-2026

All Board members have received the invitation to register in the upcoming ERN GUARD-Heart board meeting. Participation is foreseen one representative per HCP, as well as two ePAG delegates. For those unable to join on site, an online option will be available. The meeting will take place at the Bucharest Courtyard by Marriott Floreasca Hotel (14:00 – 18:30) and we will start with lunch at 13:00. The agenda is available already at the website (https://guardheart.ern-net.eu/ern-events/future-events/) . ♥



Courtyard by Marriott Floreasca
Boulevard Dimitrie Pompeiu 2A, Sector 2; 0771190 Bucharest

10th Hereditary Cardiovascular Disease Course

Bucharest, January 2026

The Hereditary Cardiovascular Disease Course in Bucharest celebrates it's 10th edition. Register and join this in-person course, where national and international experts will present highly relevant and up-to-date topics in the field. The course is addressed to all physicians from the specialties of cardiology, pediatric cardiology, medical genetics, internal medicine, and family medicine. Registration for this meeting is possible via: https://www.cardioportal.ro/hereditary-cardiovascular-diseases-course/ ♥

ERN GUARD-Heart Webinars

Amsterdam, January 2026

The ERN continues its monthly educational webinar series for healthcare professionals, covering all thematic areas within the Network. Each live 60-minute ZOOM session features expert-presented clinical cases followed by open discussion. Recordings are available on the website. If you would like to receive the programme and registration details, please subscribe to https://forms.gle/gRn6d9Zw77V5yoGv9 ♥

Please: sign the Declaration on a European Innovation and Care Ecosystem for Rare and Complex Diseases

Luxembourgh, 04-02-2026

Dear ERN- representatives,

We are excited to invite you to officially endorse and sign the Declaration on a European Innovation and Care Ecosystem for Rare and Complex Diseases.

Launched at the first-ever High Level Meeting for Rare and Complex diseases (HLM Rare) in December 2025, this Declaration marks a pivotal moment for rare disease care and innovation across Europe, building on a vision to strengthen the collective impact of the European Reference Networks.

This Declaration is a political and strategic commitment born from a multi-stakeholder coalition, including patients, healthcare professionals, ERNs, academia, and industry representatives. It outlines a shared vision with 8 Priority Actions designed to address fragmentation and accelerate progress in rare and complex diseases.

These actions include enhancing diagnostics and treatment access, fostering clinical trials, establishing comprehensive infrastructure (CoRDICs), boosting real-world evidence generation, exploring new business models, and critically, securing ringfenced funding for ERNs under the upcoming 2028–2034 Multiannual Financial Framework.

By endorsing the Declaration, you will help the European rare diseases community:

- Mobilise political commitment for a comprehensive EU Action Plan on Rare Diseases.
- Secure long-term sustainability and recognition for the vital work of ERNs and the broader rare disease ecosystem.
- Strengthen our collective voice to influence EU policy, legislation, and funding priorities.

The Declaration serves as a powerful reference for policy positions, an advocacy tool at national levels, and a robust framework for future initiatives and funding. With over 50 organisations already onboard, including EURORDIS-Rare Diseases Europe, EUCOPE, ERDERA, and ERN Coordinators, we urge you to join this growing movement and help us make the difference.

For a comprehensive overview of the Health Leadership Mission for Rare Diseases (HLM4RARE), its objectives, and the detailed recommendations, please find the presentation attached to this email.

To sign the Declaration and become a formal endorser, please follow *this link* .

When signing, you will have to indicate any European Reference Networks (ERNs) to which you are affiliated. This is for our internal tracking purposes and will help us better monitor the collective endorsement from the ERN community. If you are not affiliated with an ERN, please feel free to indicate "not applicable"

Thank you for your dedication to advancing rare and complex disease care!”

Thank you once again for your continued support and engagement in strengthening the European rare and complex disease ecosystem. ♥

Who are our ePAGs?

France, 05-02-2026

One of the European patient advocates of ERN GUARD-Heart in the spotlight: *Sophie Pierre, vice-president of patient organization AMRYC.*

Céline's Journey: Perseverance, Resilience, and Hope

I'm Sophie Pierre, vice-president of AMRYC (France) which aims at supporting all patients suffering from inherited cardiac rhythmic diseases (long and short QT syndromes, Brugada syndrome, Catecholaminergic Polymorphic Ventricular tachycardia, early repolarization syndrome..). Today, I am honored to share with you the story of Céline, our beloved daughter and the third child in our family. When I earned my PhD in Virology and Molecular Biology in 2002, I never could have imagined that my academic pursuit would one day turn into such a deeply personal journey. The knowledge and experience I gained have not only shaped my professional life but have also become essential in navigating the challenges our family has faced, especially with Céline's condition. Our journey with Céline has been marked by extraordinary challenges, unwavering love, and deep involvement with the Association for Hereditary Cardiac Rhythm Disorders.

Céline's story began on January 23, 2009, at her birth—a moment that should have been filled with celebration but instead was overshadowed by concern. Born 2 months before her term with a severe bradycardia (Doctors thought of foetal distress at that time), Céline's arrival was abrupt and fraught with uncertainty. The medical team was worried, and so were we, as parents. I couldn't see her before 3 days as she was immediately transferred to an intensive care unit where she stayed one full month; We didn't know what the future would hold, or if Céline would survive her first year.

Our journey took turn in May 2009 when we met a dedicated inherited rhythmic disease specialist at Necker Hospital. She discovered Céline was suffering from a rare hereditary cardiac rhythm disorder known as short QT syndrome type 2. This disease was unfamiliar to most doctors and terrifying in its uncertainty.

This meeting was decisive for Céline's care and our family's future. The dedicated pediatric cardiologist not only provided expert medical guidance but also learned about the disease alongside Céline as she grew. Together, we navigated unknown territory, building a partnership based on trust, learning, and hope.

After 17 years, she still takes care of Céline now in Marie Lannelongue Hospital and we express to her our deepest gratitude for everything that has been done for Céline over the past seventeen years. Her ongoing care, attention, and willingness to accommodate Céline's unique needs have made an immeasurable difference in her daily life. With time and experience, we have learned that there are only 19 published cases of short QT syndrome type 2 worldwide. Our family's journey is not just personal, it is part of a global story of rare diseases, medical discovery, and hope for the future.

Throughout these years, our family has learned to cope with uncertainty, fear, and the ever-present possibility of sudden loss. We have discovered reserves of strength we never knew we had. Living with short QT syndrome type 2 meant adapting every day to the unknown. We managed the evolving challenges of the disease, always mindful of the risk of sudden cardiac events. By the time Céline was three years old, we got a defibrillator at home—a constant reminder of the fragility of life. Céline's courage has inspired us all to face each day with hope, gratitude, and a determination to cherish every moment together.

Céline's journey was complicated further by bilateral vestibular areflexia, another rare condition linked to the same genetic mutation. This brought additional challenges to her development and daily life, demanding even greater adaptation and resilience from our family. Our personal experience led us to become deeply involved with AMRYC. Through the association, we strive to raise awareness about inherited cardiac diseases, support other families facing similar challenges, and advocate for research and better care & Quality of Life. This commitment is a way to transform our journey into a source of hope and help for others.

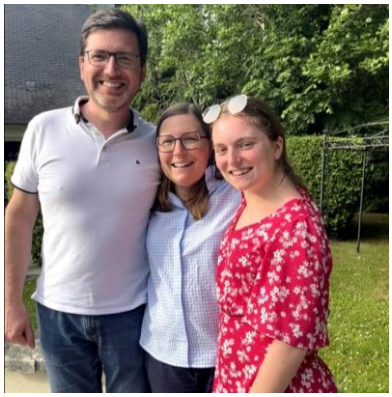
-- See next page --

Who are our ePAGs?

***Sophie Pierre** (continued)*

AMRYC commitment led us to actively promote cardiac emergency awareness and first aid training in schools and sports facilities, empowering communities to respond swiftly during crises. We also advocated for installing AEDs at local sports venues to enhance safety and protect children with rare cardiac conditions. Alongside Céline's medical journey, our family also faces the challenges of Céline's sister's psychiatric disease, which adds another layer of complexity to our daily lives and anxiety for Céline as well. Additionally, I carry the familial BRCA2 mutation inherited from my mother, a genetic risk that exposes us to breast, ovarian, and pancreatic cancers. Céline also has one chance out of 2 to have this additional deleterious mutation. Before concluding, I would like to mention the "3 days in one challenge" I face every day as other mothers taking care of disabled children : balancing my full-time role as Director of Scientific Affairs in a Diagnostic Company, caring for my beloved family, and managing the triplicate combination of cancer, cardiac disease, and psychiatric illness in the current challenging French medical care environment. Navigating these responsibilities requires constant adaptation and resilience, but I am determined to do my best for those I love and for helping other patients.

Céline's story is one of challenges, uncertainty, and adaptation—but above all, it is a story of love, hope, and family resilience. We are grateful for the support of our medical team, especially Céline's truly dedicated cardiologist and for the community we have found through the association, especially Françoise Pélissier, AMRYC's current president. By sharing our journey, we hope to inspire others to cherish their loved ones, support those facing rare diseases, and never lose sight of hope. ♥



Timothy Syndrome Alliance: CACNA1C Conference 22-23 July 2026

UK, January 2026

We are pleased to share an announcement from ePAG-linked organisation Timothy Syndrome Alliance (TSA), a charity supporting individuals with CACNA1C-related disorders. TSA will host a two-day in-person conference in Cardiff, UK, on 22–23 July 2026, bringing together patients, families, clinicians, and researchers to co-develop a patient-prioritised research agenda.
Registration is open for patients, families, researchers, and clinicians, and abstract submissions are welcomed until 16 February 2026. https://timothysyndrome.org/conference/♥

Sudden Cardiac Death Letter for families – Call for translations

Amsterdam, 05-02-2026

The letter for families following the (young) sudden cardiac death of a loved one, which was created by the E-PAGs members and approved by ERN experts, is translated in French. It would be great to have a translation in other European languages as well. The letter can be shared with hospitals, emergency physicians, paramedics, coroners, pathologists, family doctors, paediatricians, and others involved in sudden cardiac death in the young. Please contact the ERN-coordination office if you can help with the translations. The letter is available on the website:

https://guardheart.ernnet.eu/experts/forms/sudden-cardiac-death-letter/
♥

Medical Device Shortages: Watch the Cross-ERN Webinar on Regulatory Challenges

Amsterdam, 05-02-2026

On 21 January 2026, ERN eUROGEN hosted a joint webinar with ERNICA, ERKNet, and ERN GUARD-Heart to address a growing concern across rare and complex disease care: medical device shortages and regulatory constraints.
The webinar explored the real-world impact of reduced device availability, evolving EU regulatory requirements, and challenges around off-label and compassionate use of medical devices. Expert speakers (Elena Arbelo, Thomas Krasemann, Marc Gewillig from ERN GUARD-Heart)shared clinical, regulatory, and industry perspectives, highlighting how these issues affect patient access to life-saving technologies and the delivery of specialised care across Europe. The discussion also examined the current status of Medical Device Regulation (MDR) registrations and ongoing advocacy efforts within and beyond the ERN framework
This cross-ERN collaboration provides valuable insights for clinicians, researchers, patient representatives, and policymakers working in rare disease care, and remains highly relevant as regulatory and procurement challenges continue to evolve. The webinar can be re-viewed on You Tube: https://www.youtube.com/watch?reload=9&v=QGvGWXy7v80&feature=youtu.be ♥

Generated documents for ERN GUARD-Heart registries

Amsterdam, 10-02-2026

In collaboration with the Legal Office of Amsterdam UMC, a template was developed for a Joint Data Registry Agreement (JDRA). This template is an adapted version of the agreement developed by the Dutch Federation of Universities and is intended to facilitate arrangements regarding governance, data sharing, and data access for multicentre patient registries. The template can be used by centres that wish to establish a registry within the ERN and make use of the coordination and data hosting services provided by Amsterdam UMC. The JDRA consists of several appendices that need to be completed and attached. ♥

Registration Now Open for ECRD 2026 – European Conference on Rare Diseases & Orphan Products

Prague, 05-02-2026

ECRD is Europe's largest patient-led conference dedicated to shaping the future of rare disease care, research, and policy. It brings together clinicians, researchers, patient organisations, and stakeholders across sectors to advance solutions that improve the lives of people living with a rare disease. The overarching theme of ECRD 2026 is: "Rare Diseases in a Changing & Competitive Europe: Shaping policies to address the unmet needs of people living with rare diseases." ECRD 2026 will be a key milestone—five years after Rare2030 and midway to the WHO Global Plan 2028 target. The event will gather the rare disease community to assess progress and work collectively toward an EU Action Plan (or Strategic Framework) for Rare Diseases, often described as "planning for a Plan." Participants who register before 26 February 2026 will benefit from exclusive Early-Bird rates More info: https://www.rare-diseases.eu/programme-2026/ ♥

RealiseD Webinars: Innovation in RD Clinical Trials

Amsterdam, 05-02-2026

The RealiseD project is hosting a four-part webinar series exploring how to improve clinical trial design and evidence generation in rare and ultra-rare diseases.
The series brings together experts from research, regulation, industry and patient organisations to address key challenges, including complex trial methodologies and the need for more patient centred approaches. Funded by the Innovative Health Initiative, RealiseD aims to establish new standards that support innovation, reduce inequalities, and improve patient access to effective therapies.
https://realised-ihi.eu/realised-webinars-driving-innovation-in-rare-disease-clinical-trials/ ♥

REMEDi4ALL Drug Repurposing Bootcamp for Academics

Cambridge, 05-02-2026

There are still places available for the REMEDi4ALL Drug Repurposing Bootcamp for Academics, taking place 11–12 March 2026 at Hinxton Hall, Wellcome Genome Campus (Cambridge, UK).
This in-person training is designed for early-career researchers and principal investigators who are leading, or planning to lead, a drug repurposing project. The programme supports effective translation of repurposing research through expert lectures, interactive discussions, and small-group working sessions, and offers the chance to engage with key stakeholders in the field.
Participation is fully funded: course fees, travel, and accommodation are covered for selected applicants.

Places are limited to 20, and applications close on 25 February 2026 .
More info & Application: Join the Repurposing Bootcamp for Academics: https://remedi4all.org/drug-repurposing-bootcamp-academics-2026/ .♥

Latest ERN GUARD-Heart Publications

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2. De Marco C, Asatryan B, Te Riele ASJM, Di Marco A, Gasperetti A, Delinière A, Roberts JD, Jensen HK, Davies B, Krahn AD, Tadros R, Svensson A, Castelletti S, Crotti L, Platonov PG, Borowiec K, Biernacka EK, Arbelo E, David LP, Saguner AM, Healey JS, Brunckhorst C, Cappelletto C, Stolfo D, Merlo M, Rootwelt-Norberg C, Haugaa K, Duru F, van Tintelen JP, Velthuis BK, Calkins H, Zimmerman SL, James CA, Bosman LP, Cadrin-Tourigny J. **Left Ventricular Late Gadolinium Enhancement for Arrhythmic Risk Prediction in ARVC**. Circ Arrhythm Electrophysiol. 2026 Jan 29:e014265. doi: 10.1161/CIRCEP.125.014265. Epub ahead of print. PMID: 41608798.

3. Mejren AHJ, Ladefoged B, Pedersen ALD, Clemmensen TS, Oerlemans MIFJ, Fensman SK, Vase H, Andersen MJ, Poulsen SH. **Distribution and prognostic implications of right and left ventricular systolic dysfunction in wild-type transthyretin amyloid cardiomyopathy**. Int J Cardiovasc Imaging. 2026 Jan 19. doi: 10.1007/s10554-026-03614-y. Epub ahead of print. PMID: 41549185.

4. Callegari A, Butera G, Krasemann T, Heying R, Michel-Behnke I, Bonnet D, Malekzadeh-Milani S. **Response to a letter from Meng Sun and Zhiqiang Zhao commenting on the article entitled "Antithrombotic approach in percutaneous pulmonary valve implantation (PPVI): What is our standard of care? A study endorsed by the Association for EuropeanPaediatric and Congenital Cardiology**". Arch Cardiovasc Dis. 2025 Dec 20:S1875-2136(25)00839-3. doi: 10.1016/j.acvd.2025.12.001. Epub ahead of print. PMID: 41539886.

5. De Raffele M, Casas G, Delgado V, Cediel G, Faggiano A, González-Santorum F, Anmad-Shihadeh LA, Subira-Ingla A, Juncà G, Kasa G, Conte C, Weerts J, Bertini M, Rodríguez-Palomares JF, Teis A. **Left atrioventricular coupling index and atrial fibrillation and stroke in hypertrophic cardiomyopathy: a CMR study.** Rev Esp Cardiol (Engl Ed). 2026 Jan 6:S1885-5857(26)00001-0. English, Spanish. doi: 10.1016/j.rec.2026.01.001. Epub ahead of print. PMID: 41506368.

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ERN GUARD-Heart Management Office

Arthur A.M. Wilde
Network Coordinator
Amsterdam UMC
Amsterdam, Netherlands
a.a.wilde@amsterdamumc.nl

Nynke Hofman
Network Manager
Amsterdam UMC
Amsterdam, Netherlands
n.hofman@amsterdamumc.nl
Mobile: +31622046435

Mari Murel
Network Manager
Amsterdam UMC
Amsterdam, Netherlands
m.murel@amsterdamumc.nl

Ahmad S. Amin
Registry Manager
Amsterdam UMC
Amsterdam, Netherlands
a.s.amin@amsterdamumc.nl
Mobile: +31614471713