Wednesday 4 June 2025

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 2025 NUMBER 2

ERN Board Meeting in Madrid

Amsterdam, 2 June 2025

The next ERN Board Meeting will take place in Madrid, during the ESC Congress, on **Monday, 1 September 2025.** The meeting will begin at 12:00 with a light lunch, followed by the official program from 12:30 to 16:30. The venue is the **Maternal and Child Hospital Gregorio Marañón** (an ERN full member centre), located at **Calle de O'Donnell 48, Retiro, 28009 Madrid**. We will meet in the **'Salón de Actos'**, which is located close to the hospital's main entrance. The hospital is centrally located, near the main hotels, and approximately 35 minutes by metro or 20 minutes by taxi from the congress centre. ♥

ERN Coordinators Group & Board of Member States (BoMS) Meeting

Rome, 21-22 May 2025

The 22nd ERN Coordinators Group Meeting and the Joint Session with the ERNs Board of Member States, shaping the future of rare and complex disease care across Europe, was recently held in Rome. The meeting was organised by the European Commission and hosted by the Italian Ministry of Health. Our ERN was represented by Arthur Wilde. Key topics included funding mechanisms, evaluation procedures, the restructuring of working groups, and the integration of ERNs into national healthcare systems. The agenda also addressed the use of Orphacodes and the scope of disease coverage within ERNs, highlighting the importance of retaining appropriate expertise at the ERN level.

Working groups of the ERN Coordinators provided updates on their ongoing activities, covering areas such as monitoring, knowledge generation, and legal and ethical frameworks. Additionally, the Board of Member States held a dedicated session where representatives actively contributed to discussions on ERN reporting and strategic planning.

Looking ahead, the focus will shift toward promoting research initiatives and strengthening ERN collaboration, paving the way for continued progress in rare disease care across Europe. ♥

Timothy Syndrome Alliance won People's Choice Award

Cardiff, May 2025

Connections, a short public engagement film created by the Timothy Syndrome Alliance in collaboration with the Cardiff University, won the People's Choice Award at the Smiley Charity Film Awards. It shares the lived experiences of families affected by CACNA1C rare variants and celebrates the value of partnership across research, care, and community. Here is the link to the winning film: https://smileycharityfilmawards.com/films/connections

Congratulations to Sophie and her team!

As an ePAG member in the ERN GUARD-Heart, Sophie and the Timothy Syndrome Alliance carry out that meaningful progress in rare disease care starts with connection. If you are working with patients who have confirmed or suspected CACNA1C-related diagnoses, don't hesitate to get in touch: sophie@timothysyndrome.org

Sophie Muir, People's Choice winner at the Smiley Charity Film Awards

Support for the Heart Institute of Ukraine

UK, 8 May 2025

Amid ongoing conflict, the Heart Institute of Ukraine in Kyiv continues its vital work in providing life-saving care. To sustain operations, the Arrhythmia Alliance has facilitated the delivery of a significant shipment of medical supplies. The Heart Institute has issued an urgent appeal for specific medical equipment required to maintain and enhance its clinical services.

Donations of gently used equipment would be greatly appreciated to help meet these needs.

Requested equipment includes:

•Laparoscopic surgical instruments (Karl Storz)
•Endoscopic system for bronchoscopy, gastroscopy, duodenoscopy, colonoscopy (Evis X1, Olympus)
•Ultrasound diagnostic system of expert class with 3D transesophageal probe (Epic Elite, Philips)
•High-resolution ultrasound diagnostic system of expert class (Philips)
•Aortic balloon catheter (MAQUET)
•Surgical instruments for cardiac surgery
•Defibrillator with monitoring capabilities
•Patient monitoring system
•Central monitoring system for patients
•Infusion pumps (Perfusor compact plus, BBraun)

For further information or to offer support, please contact Andrea Baer a: **a.baer@heartrhythmalliance.org**.

All contributions and consideration are sincerely appreciated.

Arrhythmia Alliance
www.heartrhythmalliance.org

Joint Action on the integration of ERNs into national healthcare systems

Amsterdam, 02-06-2025

Launched in 2024, JARDIN is a Joint Action (JA) with the aim to integrate ERNs into national healthcare systems and to improve the accessibility of the ERNs for patients across Europe for better rare disease care. This EU funded 3-year project involves all Member States(MS) plus Norway and Ukraine. JARDIN will produce recommendations, and implementation pilots in the main fields of action, such as patient pathways, national reference networks, and data management for rare diseases or complex conditions. Activities will include identification and exchange of best practices, development of concrete recommendations, guidelines, and toolboxes suited for the needs of all Member States (taking into account the different preconditions, such as size and population, economy, and structure of the respective healthcare system), as well as support of capacity building and performing pilot implementation steps on different levels in MS in the proposed fields of action. JARDIN will develop strategies for systematic dissemination of information on the ERNs, with a specific emphasis on people living with rare diseases as well as the healthcare professional's community.

In line with the general objective of the call in the framework of the European Union Programme for Action in the field of Health (EU4Health), area of action "Enhanced European Reference Networks", JARDIN aims to improve the accessibility and support the long-term sustainability of the ERN system, also strengthening the resilience of the national health systems.

To disseminate JARDIN's mission and vision the JARDIN's website is created. Beside the relevant info over its work packages, it also acts as a bridge to easily access information on ERNs and offers useful resources in the area of health affecting people living with rare diseases and complex conditions. Among other tools, the ERNs dissemination toolkit can be found.
To share the latest news and to inform about the progress of this Joint Action, the JARDIN publishes external newsletters. We like to inform you that the JARDIN's #2 Newsletter is Out!

In this second edition of the external newsletter, you will find highlights from recent events and a look at what is coming next, as well as an update on the progress of Joined Actions so far. If you like to stay informed, you can subscribe for the JARDIN newsletter, click here.
Check out the operational WEBSITE and the most recent NEWSLETTER. ♥

Survey EURORDIS for ePAGs and Clinicians

Barcelona, 15 May 2025

EURORDIS Rare Diseases Europe, the patient driven alliance of patient organisations and individuals active in the field or fare diseases, published their yearly survey to assess the usefulness of the tools and resources developed by EURORDIS to support Patient Partnership.
They collect responses from ePAGs, Networkmanagers and clinicians.

It takes 5-10 minutes to complete, and open to receive responses, until June 30
Please, find the link to the survey form below
https://form.jotform.com/250683470680359

For more information about all activities of EURORDIS: https://www.eurordis.org/

ERDERA

European Rare Diseases Research Alliance

The European Rare Diseases Research Alliance (ERDERA) kicked off last September, with an estimated budget of 380 million euros and the aim of improving the lives of 30 million rare disease patients in Europe and beyond. To address these important issues, the ERDERA has been set up to build on the advancements made by former EU funded projects such as SOLVE-RD, ERICA and the European Joint Programme for Rare Diseases (EJP RD), the previous partnership which has run for the past 5 years. To leave no one behind, over 170 organizations championed by the European Union and member states are working together to make Europe a world leader in rare diseases research and innovation. The ERDERA takes over EJPRD to deliver concrete health benefits to rare disease patients in the next decade by advancing prevention, diagnosis, and treatment research. ERDERA will continue developing a robust and comprehensive data and expertise infrastructure and innovative clinical research services with a focus on advanced therapies, funding new research projects, providing training and expediting. translation of findings into tangible solutions for patients. It will also ensure alignment on international and national rare diseases research strategies. The multidisciplinary team of more than 1400 professionals involved in ERDERA will contribute their expertise in preclinical, clinical and translational research; drug development and diagnosis innovation; biostatistics; data and regulatory science; research funding; social sciences and humanities; patient engagement and education.

Read ERDERA's press release & Visit website www.erdera.org to learn more.

Research (funding) opportunities for ERNs:

ERDERA's funding research strategy is built upon a diversity of calls: Joint Transnational Calls, Clinical Trials Calls and the already active **Networking Support Scheme.** ♥

Multinational Clinical Trials Call

A call for multinational clinical trials is expected in 2026.

Yearly Joint Transnational Calls

These calls support multinational research projects that are of broad scope. Topics include improving diagnosis, developing new therapies, and understanding the social impact of rare diseases.

Call for Proposals 2025: Preclinical Therapy Studies for Rare Diseases

Networking Support Scheme

This scheme offers small grants (30 000 €) to support events or workshops aimed at fostering knowledge sharing and community building.

Through this constantly open call, ERDERA supports the organisation of transnational networking events that promote knowledge sharing, research uptake and collaborations among clinicians, researchers, and patients/patient advocacy organizations (PAOs).

Scheme is open from May 2025! Find out more

ERDERA

European Rare Diseases Research Alliance

Direct Collaboration Opportunities

Collaborate directly with ERDERA's research teams involved in "in-house" research.

Clinical Research Network – Encompassing all ERDERA's in-house research activities, this network will enhance diagnostics and clinical trial readiness, help assess the impact of rare diseases and support the development of advanced therapies. It will also build on the clinical expertise of the 24 European Reference Networks (ERNs).

ERDERA WP25 Task 4 25.4 Alignment with the Research strategies of the European Reference Networks

This task will focus on the alignment of the research strategies within the ERNs and with ERDERA. An ERNs "living lab" will be created to foster new cross-fertilising avenues across ERNs to accelerate patient relevant advancements and discuss the alignment and implementation of the acquired knowledge and relevant parts of the strategic research roadmap in ERDERA activities and in global activities. A task force will be developed composed of the 24 ERN coordinators and relevant ERDERA Work Streams, and Work Package leaders. The task force will manage the knowledge transfer of ERICA 's valuable outputs and to identify potential gaps and forward-looking actions, thus ensuring productive continuity and avoidance of overlapping actions. The ERN coordinators will provide the ERDERA governance with information and advice on the research priorities distilled from thematic workshops aimed at the identification of potential solutions to patients' unmet needs. See the presentation by the WP-leads during the ERICA & ERN Research Conference Value of WP25.4 task for ERNs in ERDERA. ♥

May 2025

Arrhythmia Alliance has been working to support ERDERA's Networking Support Scheme opens to forge new rare disease and rare cancer alliances
The European Rare Disease Research Alliance (ERDERA) has launched its Networking Support Scheme (NSS), a continuously open call designed to knit together researchers, clinicians and patient advocates working on rare diseases and rare cancers. Proposals may be lodged at any time, but the first collection round will be assessed immediately after the 7 October 2025 deadline, allowing successful teams to start planning events without delay. Under the scheme, applicants can request up to €30 000 per networking event to cover meeting costs, travel costs, hybrid-meeting platforms and other essentials. Rounds will be held every six months until April 2029—or until the dedicated budget is exhausted—providing a response pathway for emerging collaborations of the partnership.
Eligibility is broad: researchers (including early-career investigators), clinicians, research managers and patient advocacy organisations may all act as principal applicants. Each event must gather applicants from at least three ERDERA countries and focus squarely on knowledge exchange in rare disease or rare cancer research. Meetings may run in person or in hybrid formats to maximise reach. A cornerstone of the call is inclusiveness. ERDERA "strongly encourages" convenors from 18 underrepresented countries—among them Bulgaria, Greece, Poland, Romania and Türkiye—to apply, aiming to widen Europe's rare disease network and ensure that expertise flows to and from regions that have historically lacked access.
Applications are screened for eligibility, then scored by an independent, transnational evaluation committee using transparent criteria aligned with Horizon Europe standards. Projects that clear the bar, have satisfactory ethics assessment and fit within the available budget—receive rapid confirmation so organisers can set dates, secure venues and invite speakers.
More information, such as guidance notes, evaluation criteria and the full list of eligible countries are available on the **ERDERA website.** ♥

Calling experts from underrepresented countries

May 2025,

Attention to all our ERN experts in the following EU Member States: Bulgaria, Cyprus, Czechia, Estonia, Greece, Georgia, Hungary, Ireland, Latvia, Lithuania, Morocco, Poland, Portugal, Romania, Serbia, Slovakia, Slovenia, Türkiye.

ERDERA is seeking experts from these so-called underrepresented countries (UCs) to join their exclusive pool of specialists. By expressing your interest, you may be invited to participate in ERDERA activities, expert panels, and networking opportunities. You will also receive valuable information on funding opportunities, available services, training, and education programs. This is an excellent opportunity to connect, contribute, and grow with a dynamic international community. Those interested, please fill in this short form about your expertise. ♥

Latest ERN GUARD-Heart Publications

1. Nitsche C, Dobner S, Rosenblum HR, Patel KP, Longhi S, Yilmaz A, Merlo M, Papathanasiou M, Griffin J, Oerlemans MIFJ, Gama F, Hamdan A, Kelion AD, Schuster A, Glaveckaité S, Akyol N, Porcari A, Schlender L, Capovilla T, Autherith M, Hauptmann L, Halavina K, Cavalcante JL, Fontana M, Scully PR, Moon JC, Mascherbauer J, Ristl R, Biagini E, Stortecky S, Maurer MS, Treibel TA; AS-Amyloidosis Consortium. Cardiac transthyretin amyloidosis treatment improves outcomes after aortic valve replacement for severe stenosis. Eur Heart J. 2025 Jun 2:ehaf362. doi: 10.1093/eurheartj/ehaf362. Epub ahead of print. PMID: 40452225.

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4. Reckman YJ, Haas J, van der Made I, Williams SG, Diaz IG, Akhtar M, Mogensen J, Rasmussen TB, Villard E, Charron P, Elliott P, Keavney BD, Monserrat L, Pinto YM, Meder B, Tijsen AJ. Rare DCM associated variants in pre-miR-208a disrupt miRNA maturation and function. Hum Mol Genet. 2025 May 6:ddaf069. doi: 10.1093/hmg/ddaf069. Epub ahead of print. PMID: 40327887.

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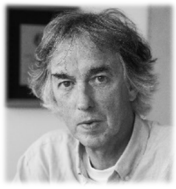

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