

Tuesday 24 December 2024



GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart



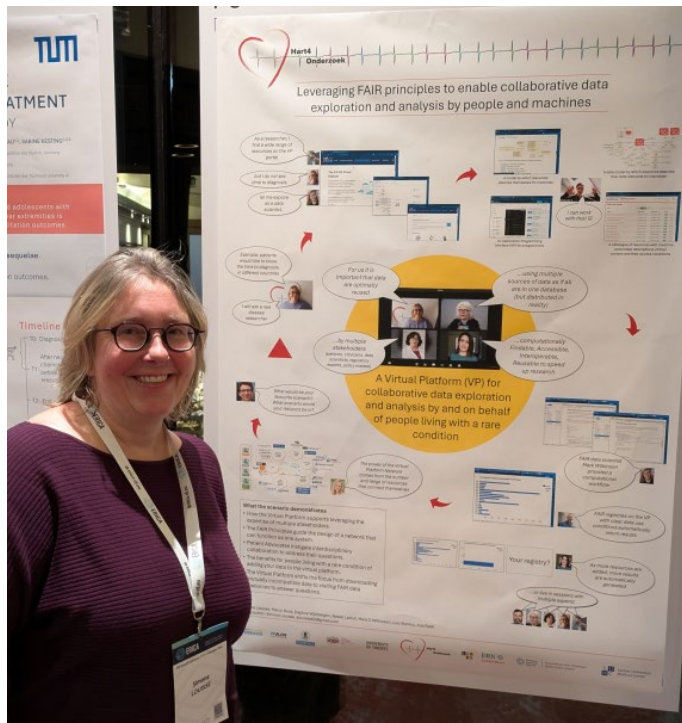
ERN GUARD-HEART BIMONTHLY NEWSLETTER

YEAR 2024 NUMBER 6

ERICA Conference in Udine

Udine, 11-12-2024

The ERICA conference in Udine, took place from December 11th – December 13th. The ERICA Conference serves as a great opportunity to ‘meet and greet’ the active ERN research community, to present the research projects involving ERNs and to discuss the future of the ERN related Research. Three abstracts of ERN GUARD-Heart were accepted, so there was an active participation from our ERN. Congratulations to all these researchers. ❤️



Simone Louisse, e-PAG Hart4Onderzoek at the Poster presentation in Udine

Next board-meeting in March

Maarsssen, 23-12-2024

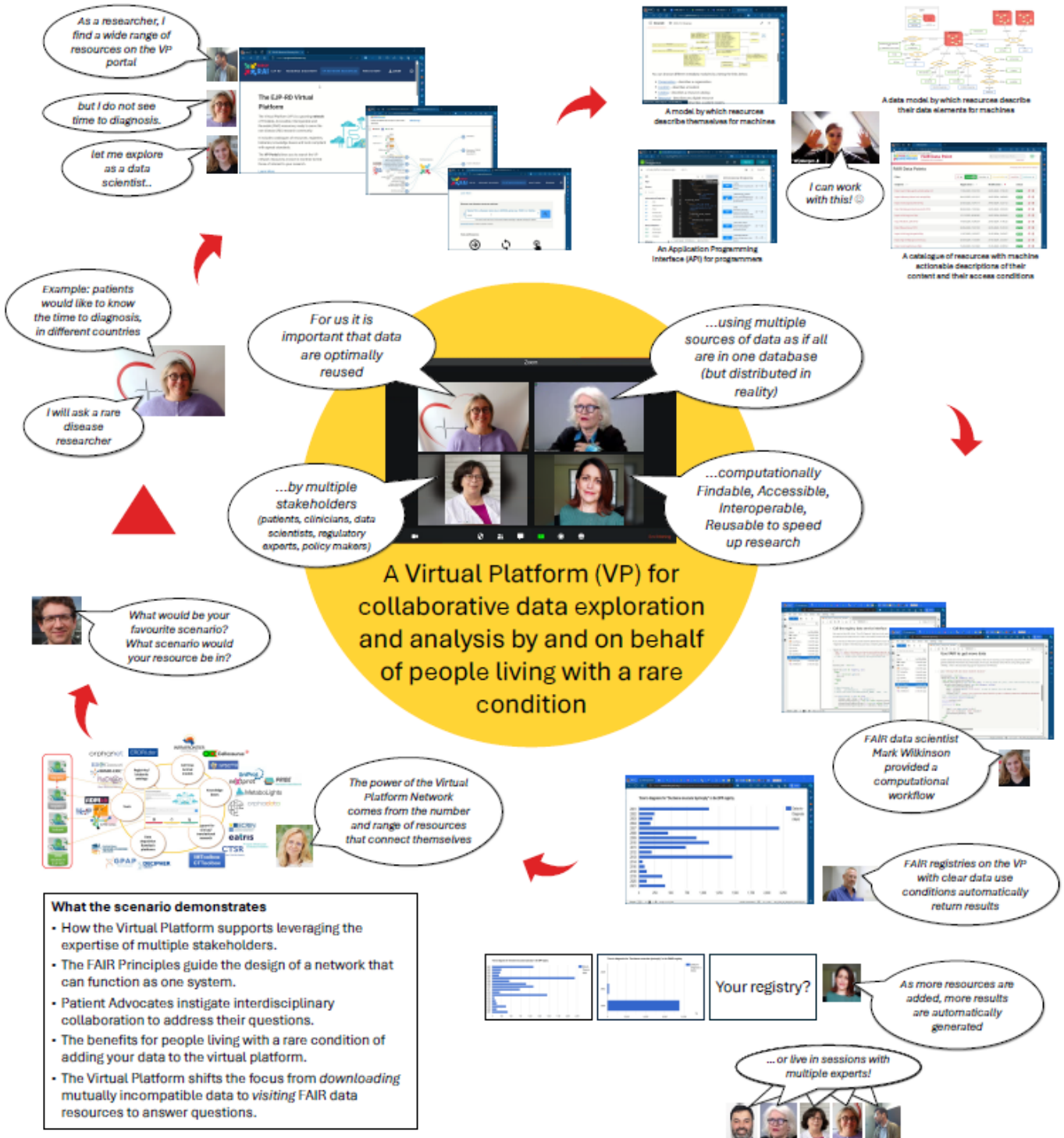


The next board meeting of ERN GUARD-Heart will be held – Friday 14 March 2025 - on site in the Netherlands, in Maarsssen at the Country Estate Doornburgh. We will meet at 9:00 AM in the chapel of the former monastery and we will have a nice lunch in the restaurant of the same building. There will be some extra time planned for the different topics (such as CPMS and registries). The meeting will be finished probably around 15:30.

For efficient organization it is important to know who will be able to attend. A registration link was sent by e-mail. We will confirm your registration early January. ❤️



Leveraging FAIR principles to enable collaborative data exploration and analysis by people and machines



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Online: ERDERA website!

The rare diseases landscape has dramatically changed over the past decade in Europe, thanks largely to the work done by the European Joint Programme for Rare Diseases (EJPRD) and previous partnerships in advancing basic research and clinical applications, fostering patient engagement and enabling important international collaborations.

More specifically, EJP RD mobilized 101 million euros (55 million came from the EU) to fund collaborative research projects. The partnership has also developed a cutting-edge virtual platform that now provides coordinated access to rare disease data, tools and resources. To build the capacity of all stakeholders involved in rare disease research, EJP RD has provided training, mentoring and clinical studies support services, as well as resources such as the Innovation Management Toolbox or the Rare Diseases Clinical Trials Toolbox. Finally, the partnership has proved crucial for aligning international research policy agendas with national strategies. However, work is far from done. There are still considerable gaps that affect millions of people living with a rare disease

and, indirectly their families, carers and society as a whole. To leave no one behind, over 170 organisations championed by the European Union and members states are working hand in hand to make Europe a world leader in rare diseases research and innovation. The European Rare Disease Research Alliance (ERDERA) takes over EJPRD to deliver concrete health benefits to rare disease patients in the next decade by advancing prevention, diagnosis and treatment research.

Take a look on the website for the staggering figures of today: [ERDERA - European Rare Diseases Research Alliance](#)

ERDERA Joint Transnational Call 2025: Apply Now!

ERDERA Joint Transnational Call (JTC) 2025, invites innovative research proposals focusing on “Pre-clinical therapy studies for rare diseases using small molecules and biologicals – development and validation.” This call aims to foster international collaborations to advance therapeutic solutions for rare diseases, benefiting millions of patients worldwide. Click [on the link](#) for more information. ❤

THE 2025 JOINT TRANSNATIONAL CALL IS OPEN!

CALL
OPENS

December 10, 2024

PRE-
PROPOSAL
DEADLINE

February 13, 2025

LEARN MORE:
JOIN THE
WEBINAR

December 17, 2024

FULL
PROPOSAL
DEADLINE

July 9, 2025



ERNs evaluation results report

Luxembourg, december 2024

The following report is a comprehensive evaluation of **the first 5 years** of the European Reference Networks (ERNs) and its members, the Healthcare Providers (HCPs), across Europe, focusing on their role in addressing complex and rare conditions. 24 ERNs and 836 HCPs from 24 EU Member States and Norway participated in the evaluation. The evaluation included self-evaluations, document reviews, ERN interviews, on-site HCP audits and stakeholder interviews. The results showed a high level of commitment of ERNs to their objectives, with a significant proportion, 100% of ERNs and 89.7% of their members, achieving satisfactory results in the evaluation. Strengths were identified in the areas of Education and Training, Networks and Dissemination, Competence, Experience, and Patientcentred care, while Quality and Patient Safety, and Organization and Management are areas for improvement for both, ERNs, and HCPs respectively.

This report, drawing on a wide range of data, increases the understanding of the essential role of the ERN system in addressing rare diseases and conditions across Europe. It also presents relevant information on the structure, maturity, activity, and impact of the ERN system, and identifies opportunities for improvement based on the findings of the stakeholder interviews and the Evaluation to further enhance the evaluation process and ensure the continued sustainability of the ERN system. The information and views set out in this report are those of the Independent Evaluation Body and do not necessarily reflect the official opinion of the Commission/Executive Agency. ♥

Click [here](#) for the link to the report .





Reminder: HCP Performance indicators, 31 December 2024!

Amsterdam, 15 December 2024,

All the ERNs must evaluate the performance of the separate member-HCPs in the ERN-network. This performance-indicator evaluation will take part each 6 months. This is the second period of evaluation. All the full-members received the evaluation form by e-mail. Many of you returned it already. The form was sent by e-mail.

After this evaluation all the HCP's will receive feedback, based on the results of both 6-months evaluation forms. This file will be shared with all the member centers. Centers with a score < 20% (on the yearly basis) will be invited to write an improvement plan.

Please send back your form to the management (n.hofman@amsterdamumc.nl), **finally on the 31st of December 2024.** ♥



Merry Christmas and a Happy New Year!

Amsterdam, 24-12-2024

Thanks a lot for your collaboration and participation in 2024.

Merry Christmas and Best Wishes for a healthy, and peaceful New Year.

Let's make 2025 another good one. ♥

Arthur, Amin, and Nynke





Latest ERN GUARD-Heart Publication (s)

1. Novel risk predictor of arrhythmias for patients with potassium channel related congenital Long-QT Syndrome.

Juarez CK, Proost VM, Tanck MW, Dittmann S, Bos JM, Crotti L, Barc J, van den Berg MP, Mujkanovic J, Rickert C, Lopes Neves RA, Musu G, Dagradi F, Giovenzana FL, Clédel A, Thollet A, Giudicessi JR, Tfelt-Hansen J, Probst V, Schwartz PJ, Ackerman MJ, Schulze-Bahr E, Bezzina CR, Wilde AAM. Heart Rhythm. 2024 Dec 13:S1547-5271(24)03660-9. doi: 10.1016/j.hrthm.2024.12.015. Online ahead of print. PMID: 39675651

2. Key priorities for the implementation of the 2023 ESC Guidelines for the Management of Cardiomyopathies in low resource settings.

Jurcut R, Barriaes-Villa R, Biagini E, Garcia-Pavia P, Olivotto I, Protonotarios A, Arbustini E, Mogensen J, Elliott P, Arbelo E, Pablo Kaski J; Members of the Task Force for the 2023 ESC Guidelines for the management of cardiomyopathies. Eur Heart J Qual Care Clin Outcomes. 2024 Dec 9:qcae103. doi: 10.1093/ehjqcco/qcae103. Online ahead of print. PMID: 39657995

3. Publisher Correction: Genome-wide association study reveals mechanisms underlying dilated cardiomyopathy and myocardial resilience.

Jurgens SJ, Rämö JT, Kramarenko DR, Wijdeveld LFJM, Haas J, Chaffin MD, Garnier S, Gaziano L, Weng LC, Lipov A, Zheng SL, Henry A, Huffman JE, Challa S, Rühle F, Verdugo CD, Krijger Juárez C, Kany S, van Orsouw CA, Biddinger K, Poel E, Elliott AL, Wang X, Francis C, Ruan R, Koyama S, Beekman L, Zimmerman DS, Deleuze JF, Villard E, Trégouët DA, Isnard R; FinnGen; VA Million Veteran Program; HERMES Consortium; Boomsma DI, de Geus EJC, Tadros R, Pinto YM, Wilde AAM, Hottenga JJ, Sinisalo J, Niiranen T, Walsh R, Schmidt AF, Choi SH, Chang KM, Tsao PS, Matthews PM, Ware JS, Lumbers RT, van der Crabben S, Laukkanen J, Palotie A, Amin AS, Charron P, Meder B, Ellinor PT, Daly M, Aragam KG, Bezzina CR. Nat Genet. 2024 Dec;56(12):2843. doi: 10.1038/s41588-024-02047-4. PMID: 39633063 **Free PMC article.** No abstract available.

4. Inappropriate Therapy and Shock Rates Between the Subcutaneous and Transvenous Implantable Cardiac Defibrillator: A Secondary Analysis of the PRAETORIAN Trial.

Olde Nordkamp LRA, Pepplinkhuizen S, Ghani A, Boersma LVA, Kuschyk J, El-Chami MF, Behr ER, Brouwer TF, Kääb S, Mittal S, Quast ABE, van der Stuijt W, Smeding L, de Veld JA, Tijssen JGP, Bijsterveld NR, Richter S, Brouwer MA, de Groot JR, Kooiman KM, Lambiase PD, Neuzil P, Vernooy K, Alings M, Betts TR, Bracke FAL, Burke MC, de Jong JSSG, Wright DJ, Jansen WPJ, Whinnett ZI, Nordbeck P, Knaut M, Philbert BT, van Opstal JM, Chicos AB, Allaart CP, Borger van der Burg AE, Dizon JM, Miller MA, Nemirovsky D, Surber R, Upadhyay GA, Weiss R, de Weger A, Wilde AAM, Knops RE; PRAETORIAN Investigators. Circ Arrhythm Electrophysiol. 2024 Dec;17(12):e012836. doi: 10.1161/CIRCEP.124.012836. Epub 2024 Dec 3. PMID: 39624908 Clinical Trial.

5. State of the art and perspectives of gene therapy in heart failure. A scientific statement of the Heart Failure Association of the ESC, the ESC Council on Cardiovascular Genomics and the ESC Working Group on Myocardial & Pericardial Diseases.

Van Linthout S, Stellos K, Giacca M, Bertero E, Cannata A, Carrier L, Garcia-Pavia P, Ghigo A, González A, Haugaa KH, Imazio M, Lopes LR, Most P, Pollesello P, Schunkert H, Streckfuss-Bömeke K, Thum T, Tocchetti CG, Tschöpe C, van der Meer P, van Rooij E, Metra M, Rosano GMC, Heymans S. Eur J Heart Fail. 2024 Nov 22. doi: 10.1002/ejhf.3516. Online ahead of print. PMID: 39576264

6. aTrial arrhythmias in inhEriTed aRrhythmla Syndromes: results from the TETRIS study.

Conte G, Bergonti M, Probst V, Morita H, Tfelt-Hansen J, Behr ER, Kengo K, Arbelo E, Crotti L, Sarquella-Brugada G, Wilde AAM, Calò L, Sarkozy A, de Asmundis C, Mellor G, Migliore F, Letsas K, Vicentini A, Levinstein M, Berne P, Chen SA, Veltmann C, Biernacka EK, Carvalho P, Kabawata M, Sojema K, Gonzalez MC, Tse G, Thollet A, Svane J, Caputo ML, Scrocco C, Kamakura T, Pardo LF, Lee S, Juárez CK, Martino A, Lo LW, Monaco C, Reyes-Quintero ÁE, Martini N, Oezkartal T, Klersy C, Brugada J, Schwartz PJ, Brugada P, Belhassen B, Auricchio A. Europace. 2024 Dec 3;26(12):euae288. doi: 10.1093/europace/euae288. PMID: 39527076 **Free PMC article.**

7. Clinical Features and Outcomes of Pediatric MYH7-Related Dilated Cardiomyopathy.

de Frutos F, Ochoa JP, Webster G, Jansen M, Remior P, Rasmussen TB, Sabater-Molina M, Barriaes-Villa R, Girolami F, Cesar S, Fuentes-Cañamero ME, Alvarez García-Rovés R, Wahbi K, Limeres J, Kubanek M, Sliker MG, Sarquella-Brugada G, Abrams DJ, Dooijes D, Domínguez F, Garcia-Pavia P; European Genetic Cardiomyopathies Initiative Investigators. J Am Heart Assoc. 2024 Nov 5;13(21):e036208. doi: 10.1161/JAHA.124.036208. Epub 2024 Nov 4. PMID: 39494569

8. Long-term prognosis of patients with an SCN5A loss-of-function variant and progressive cardiac conduction disorder or Brugada syndrome.

Tuijnburg F, Proost VM, Thollet A, Barc J, Groffen AJA, Veerman CC, van der Crabben SN, van der Pas VR, Kyndt F, Jurgens SJ, Tanck MWT, Postema PG, Peter van Tintelen J, Bezzina CR, Probst V, Wilde AAM, Gourraud JB, Amin AS. Heart Rhythm. 2024 Nov 2:S1547-5271(24)03518-5. doi: 10.1016/j.hrthm.2024.10.057. Online ahead of print. PMID: 39491571 7.

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