

Monday 3 December 2018



GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart



ERN GUARD-HEART BIMONTHLY NEWSLETTER

YEAR 2018 NUMBER 6

4th Conference on European Reference Networks “ERNs in Action”

Brussels, 22 November 2018

The 4th Conference on the European Reference Networks “ERNs in action” was held on 21 and 22 November 2018 in Brussels. **ERN GUARD-Heart** was presented to the Conference with 9 delegates. The event gathered more than 400 participants – EU institutions, National authorities, healthcare providers, hospital managers, ERN members, and patients’ representatives – to share and discuss the results achieved so far and challenges ahead. The Conference opened a new stage in the ERNs’ lifecycle: the deployment phase. It also presented the main clinical & organisational outcomes of the ERNs and discussed the challenges & margins for future improvements.

The event consisted of a series of plenary sessions addressing the state of play of the ERNs, the tools & resources for their deployment, the EU political and institutional commitment, and the challenges and integration of ERNs into national healthcare systems. Moreover, 8 parallel sessions focused on specific aspects, including ERN showcases, quality assessment, monitoring & evaluation of the ERNs, research, hospital’s management & ERNs, ERNs sustainability, patient-healthcare professional partnerships in ERNs, clinical guidelines, & new medicaments, medical devices, clinical trials & stakeholders support. Links to the presentations will become available soon on the [IT Platform](#). ❤



ERN members at the ‘ERNs at the Action’ Conference. FLTR: Ahmad Amin & Nynke Hofman (project managers), Arthur Wilde (ERN coordinator), Evelien van der Schaaf-de Wolf (CPMS helpdesk), Dayenne Zwaagman (patient representative), Jacob Tfelt-Hansen (ERN representative), Hidde Bleijendaal (CPMS helpdesk) & Jaana Pihkala (ERN representative). Georgia Sarquella-Brugada & Philippe Charron (ERN representatives) were also present at the event.



ERN News



Long QT syndrome & Exercise

Amsterdam, 29 November 2018

Currently, evidence-based guidelines for patients with long QT syndrome who engage in (competitive) sports are lacking. ERN GUARD-Heart aims to establish an advisory panel of experts who wish to provide their opinion on patients with long QT syndrome who are referred to the ERN for online consultation through CPMS, and who participate in (competitive) sports. Ultimately, all cases will be collected in a database for future research studies. We would like to ask all ERN members to respond who are interested to participate in this small project. Shortly we will circulate a study set-up. ♥

ERN GUARD-Heart Patient Groups Meeting

Barcelona, 9 December 2018

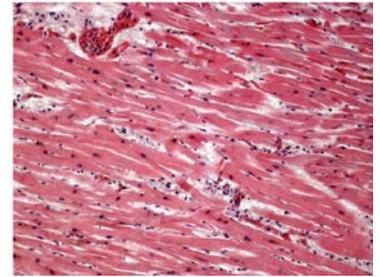
The seven patient advocacy groups in the ERN GUARD-Heart (see [ERN website](#) for details) have been granted 7 travel grants of €500,- each by the EURORDIS to organise a face-to-face meeting. The meeting will held on Sunday 9 December 2018 in Barcelona. The agenda of the meeting includes the following topics: tasks and responsibilities of the patient advocacy groups, organisation of the Patient Advocacy Board of the ERN GUARD-Heart, patient reported outcome measures (PROMS), patient pathways, and clinical guidelines. ♥



European Reference Network

for rare or low prevalence complex diseases

Network
Heart Diseases
(ERN GUARD-HEART)



Digital European Pathology Network of ERN experts

Amsterdam, 29 November 2018

Histopathology expertise across the European Union in the diagnosis of rare diseases is an eminent task across all ERNs. Two ERNs (ERN EpiCare for epilepsy and ERKNet for kidney diseases) have joined forces with the aim of developing a common (cross-ERN) web-based pathology network.

The 2 ERNs invite all other ERN experts engaged in the histopathology & tissue-based molecular-genetic work-up of rare diseases to participate and join this new cross-ERN Task Force. For further information, please contact Dr Jan Becker (jan.becker@uk-koeln.de) (nephropathology, Cologne, Germany) & Dr Ingmar Blümcke (Bluemcke@uk-erlangen.de) (neuropathology, Erlangen, Germany). ♥

5th ERN GUARD-Heart Board Meeting



13 February 2019

Paris - France



Co-funded by
the European Union





ERN News

Continuous monitoring and assessment of ERNs

Quality indicators

Brussels, 20 November 2018

ERN GUARD-Heart has seven overarching goals to achieve within 5 years (2017-2022): 1) to set up, consolidate and extend the ERN, 2) to concentrate knowledge/expertise to improve clinical practice using best available evidence, 3) improve quality and safety of healthcare services, 4) guarantee accessibility to healthcare, 5) promote research and innovation both in diagnosis and treatment, 6) put the patient and their family in the centre of healthcare services, and 7) foster transparency in the delivery of healthcare through a global communication strategy.

To timely assess whether the ERN GUARD-Heart is performing adequately to achieve its overarching goals, a continuous monitoring system has been officially set up. The system uses structure, process, and outcome indicators to:

- allow a closer follow-up and assessment of the activities performed by the Network,
- ensure consistency across the assessment of all member centres within the ERN, and support the self-assessment processes,
- build an ongoing quality improvement system for timely identification of areas for improvement,
- if necessary, when objectives are not met, foster organisational change or adjustments in the strategy of the ERN or request further support from Member States or the European Commission,
- promote patient empowerment by enabling educated choices by the patients,
- provide transparency & demonstrate the value of the ERN to the European Commission, Board of Member States, healthcare authorities, patients, health care providers, and clinical and research experts (accountability), and,
- facilitate periodic evaluation of the ERN and its Members by an evaluation body appointed by the European Commission.



The continuous monitoring system for ERNs has been established through a collaboration between the European Commission, ERNs, and patients. The system uses both 'common' indicators to monitor and assess the activities and progress of all ERNs (i.e., indicators applicable to all ERNs), as well as 'specific indicators' to monitor the progress of one specific ERN based on the diseases addressed by the ERN and technical aspects specific to the ERN.

A total of 18 common indicators (structure, process, or outcome indicators) have been selected to monitor the activities and the progress of the ERNs. All ERNs (including ERN GUARD-Heart) need to show that the Networks are functioning and are adding value compared to what existed before the ERNs. The 18 common indicators have therefore been chosen with regard to their ability to capture the added value after the establishment of ERNs. Moreover, the common indicators capture successes and failures in trying to fulfil the ERN objectives.

In addition, a total of 11 specific indicators will be used to assess the progress and the activities of the ERN GUARD-Heart and its individuals members. A full list of all common and specific indicators will be disseminated soon by the ERN project management office among the 24 representative members of the ERN.

It is important to highlight that ERNs could be terminated by the European Commission if they have not achieved the common ERN indicators. Similarly, membership of a specific ERN may be terminated if a member centre has not achieved the specific indicators. It may also important and relevant to note that many of the common and specific indicators focus on the use of the Clinical Patient Management System (CPMS) by ERNs to discuss patients. ♥



ERN PATIENT REGISTRIES

ERN GUARD-Heart members have established the following patients registries (ongoing or new)

Registry	Registry leader	Registry coordinator	Link - Host	Minimal no. of patient to include
CALM genes	Peter Schwartz & Lia Crotti <i>Milan, IT</i>	Carla Spazzolini (c.spazzolini@auxologico.it)	https://redcap.heart-institute.nl AMC, Amsterdam, NL	1
CPVT	Arthur Wilde <i>Amsterdam, NL</i>	Krystien Lieve (k.v.lieve@amc.nl)	https://www.openclinica.nl AMC, Amsterdam, NL	10
Short QT syndrome	Silvia Priori & Andrea Mazzanti <i>Pavia, IT</i>	Maira Marino (maira.marino@icsmaugeri.it)	https://redcap.heart-institute.nl AMC, Amsterdam, NL	1
PRKAG2 gene	Philippe Charron <i>Paris, FR</i>	Gabriel Laurent (gabriel.laurent@chu-dijon.fr)	https://redcap.heart-institute.nl AMC, Amsterdam, NL (under development)	1
Type 5 long QT syndrome	Jason Roberts <i>Ontario, CA</i>	Jason Roberts (jason.roberts@lhsc.on.ca)	https://redcap.lawsonresearch.ca Lo London Health Sciences Centre, Ontario, CA	1
Long QT syndrome (GWAS)	Connie Bezzina <i>Amsterdam, NL</i>	Najim Lahrouchi (n.lahrouchi@amc.nl)	https://redcap.heart-institute.nl AMC, Amsterdam, NL (under development)	10
ARVC	Peter van Tintelen <i>Amsterdam, NL</i>	Peter van Tintelen (p.vantintelen@amc.nl) Nynke Hofman (n.hofman@amc.nl)	https://redcap.heart-institute.nl AMC, Amsterdam, NL (under development)	5

How to participate

For including patients in the registries, please check the minimal number of patients that are required for inclusion (see table) and contact the specific study-coordinator by e-mail. He/she will request for a log-in account. Only registry leaders and coordinators have access to the complete data of their registry. As a participant you will receive access to the patients of your own HCP.

Consent form

Consent of the patient is required. For new patients and families the new designed ERN-consent form should be used and stored at the local HCPs. This consent form is available on the [IT-platform](#).

Questions

For general questions about existing registries, please contact Nynke Hofman (project/research manager of the ERN GUARD-Heart; e-mail: n.hofman@amc.nl).





ERN SURVEY

Survey on the use of genetic testing in ERN GUARD-Heart

Amsterdam, 29 November 2018

In November 2018, a survey on the use of genetic testing was conducted among representative members of ERN GUARD-Heart. Twenty out of 24 representative members responded to the survey (no response was received from Clinical Scientific Institutes Maugeri Pavia, Foundation Policlinico San Matteo Pavia, Louis Pradel Cardiovascular Hospital Lyon, and University Hospitals Brussels). ERN GUARD-Heart centres performed genetic testing by using Sanger sequencing or Next-Generation sequencing (gene panels and/or whole-exome sequencing). The purposes of genetic testing were for diagnosis (20/20), therapy (13/20), family screening (20/20) or prognosis (13/20). There were large differences in some aspects of genetic testing between ERN centres, e.g., whether the costs of genetic testing is covered by insurance (fig. 1), number of genes included in panels (fig. 2), and how long it takes before test results are known. Full results of the survey will be published on the ERN GUARD-Heart [website](#). ❤

Fig. 1: Reimbursement of genetic testing

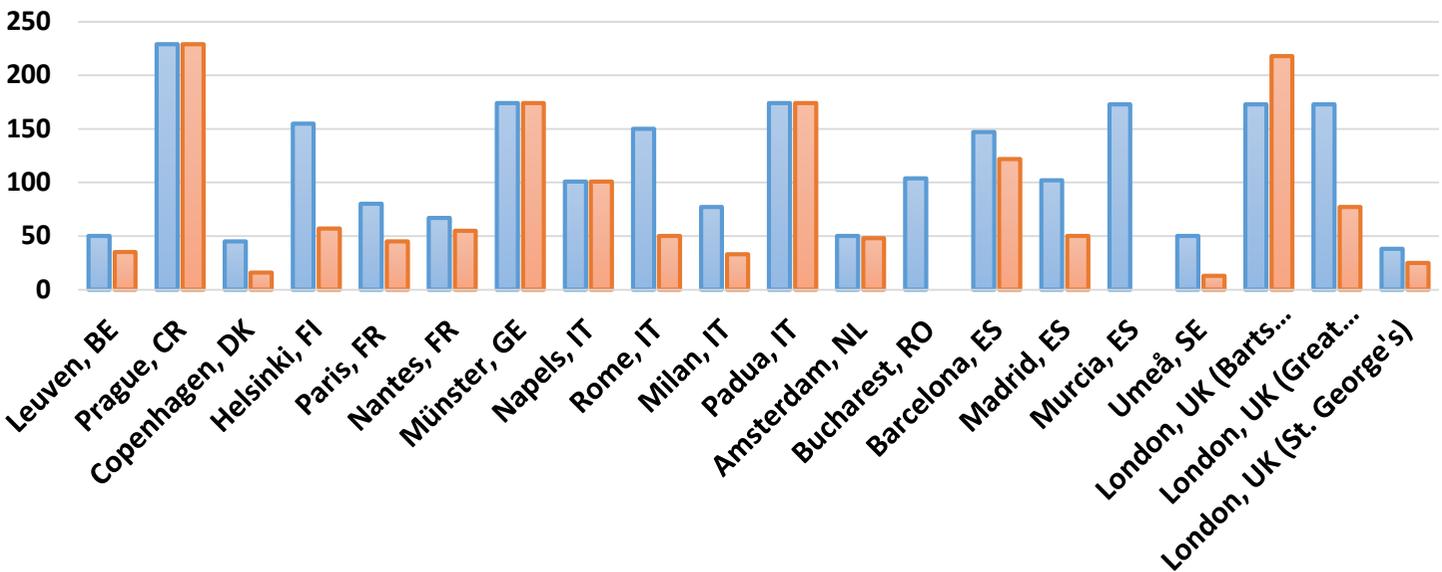
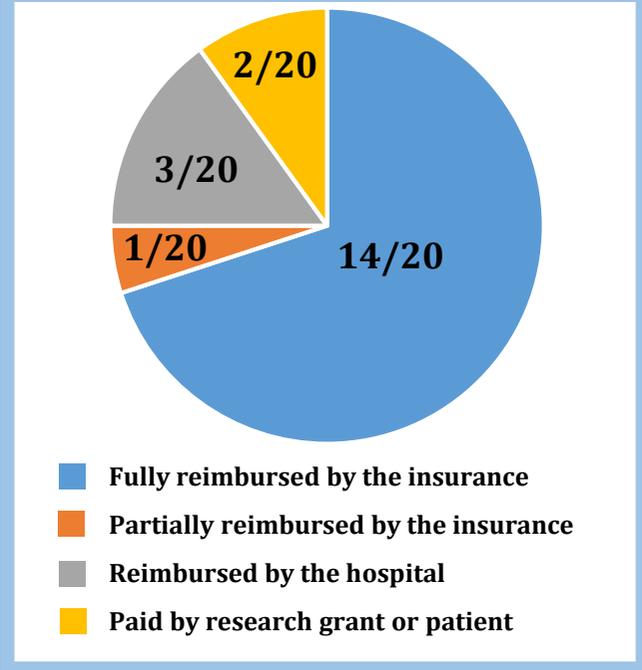


Fig. 2: Number of genes included in cardiomyopathy (blue bars) or arrhythmia panel (red bars) per centre.

Follow ERN GUARD-Heart on: *(click on logo to proceed)*



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