

Sunday 30 September 2018

ERN

GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart



ERN GUARD-HEART BIMONTHLY NEWSLETTER

YEAR 2018 NUMBER 5

4th Meeting of ERN GUARD-Heart Board of Representatives

Munich, 28 August 2018

The 4th Board meeting of ERN GUARD-Heart took place on Tuesday 28 August in Munich, Germany. The meeting was attended by members of several representatives, i.e. Anwar Baban, Cristina Basso, Elijah Behr, Alida Caforio, Philippe Charron, Lia Crotti, Carlo De Asmundis, Fernando Dominguez, Fabrizio Drago, Pablo Garcia Pavia, Juan Gimeno, Tiina Helio, Ruxandra Jurcut, Jaana Pihkala, Vincent Probst, Tomas Robyns, Annika Rydberg, Georgia Sarquella-Brugada, Eric Schulze-Bahr, Peter Schwartz, and Arthur Wilde. From the ERN Patient Advocacy Board, Ester Costafreda and Patricia Vlasman were present.

During the meeting several general topics were discussed, including the activities of ERN in its 1st year of its existence (3/2017-3/2018), plans for the 2nd ERN year (3/2018-3/2019), extension of the ERN with new centres and additional thematic areas, the European Joint Programme for Rare Diseases (EJPD-RD), and ERN patient registries. Moreover, plans for a number specific objectives for 2018-2019 were proposed, i.e., to establish uniform European cross-border patient pathways for rare cardiac diseases, to set up exchange programs for health professionals in ERN GUARD-Heart, to organize patient-physician meetings, inclusion of dedicated patient advocacy groups,

translation of patient folders in different European languages, identification and selection of patient reported outcome measures (PROMs), and the performance of bench mark studies between the different ERN centres. At the end of the meeting a workshop was provided for introduction and training in the Clinical Patient Management System (CPSM), the e-based 2nd opinion service to discuss patient within the ERN.

The notes of the meeting have been disseminated among ERN members by the management office, and are also available on the [EU IT Platform](#). ♥





ERN News



Assessment of ERN GUARD-Heart Members by the European Commission

Amsterdam, 27 September 2018

As previously announced in the ERN GUARD-Heart Newsletter of September 2018, the Andalusian Agency for Healthcare Quality (ACSA), commissioned by the European Commission, has carried out a technical assessment of two members of the ERN GUARD-Heart, i.e.: “*Emergency Institute for cardiovascular disease: Prof dr. C. C. Iliescu*” (Bucharest; technical assessment and on-site visit) and “*University Hospital Virgen de la Arrixaca*” (Murcia; technical assessment). Both centres have received favourable feedback from ACSA and will receive a final assessment report in the near future. Next year, another sample of ERN members will be assessed by DG SANTE European Commission. DG SANTE has reassured all ERNs that their members will be selected in an objective way respecting a ratio between the magnitude of the Networks and a geographical balance. ♥



Ruxandra Jurcut
*Emergency Institute for
cardiovascular disease:
Prof dr. C. C. Iliescu*



Juan R. Gimeno
*University Hospital
Virgen de la Arrixaca*



Ruxandra Jurcut (*Emergency Institute for cardiovascular disease: Prof dr. C. C. Iliescu*) and members of the Andalusian Agency for Healthcare Quality (ACSA) during the on-site visit.



Cross-border and uniform patient care pathways

Munich, 28 August 2018

ERN GUARD-Heart aims to establish uniform cross-border patient care pathways to provide equal and high quality care for all patients with rare cardiac diseases across Europe. A care pathway is one of the main tools used to manage the quality in healthcare concerning the standardisation of care processes. The project management office plans to collect care pathways, either for the diagnostic or therapeutic processes of patients with rare cardiac diseases, that are currently available in each ERN member centre. Available care pathways will be reviewed and uniform care pathways will be developed. ERN members who wish to participate in this project can contact the management office.

ACTION REQUEST: ERN centres are requested to send their patient care pathways to the project management office. ♥

SAVE THE DATE

Educational Course on

Sudden Cardiac Death

& Familial Electrical Diseases

6 December 2018 – London, UK



Co-funded by
the European Union





Education

Exchange programs (fellowships) for healthcare professionals

Amsterdam, 27 September 2018

The ERN GUARD-Heart aims to organize specialized training programmes for healthcare professionals in the Network. At the 4th Board Meeting, several ideas were proposed. For the 2nd ERN year (i.e., from March 2018 till February 2019), the Network plans to set up two fellowships at 2 member centres: one fellowship at a specialized cardiovascular pathology unit and one fellowship at a specialized molecular genetic diagnostic unit. A budget of €5.000,- per fellowship will be available for the hosting centre to arrange a fellowship for a duration of 3-6 months.

ACTION REQUEST: ERN centres that are willing to organize a fellowship are requested to contact the management office.

In the next years, the project management office, together with the cross-cutting action group on Teaching and E-learning, will elaborate the idea to produce online video materials (e.g., PowerPoint presentations with voice-over by ERN experts) to educate healthcare professionals on rare cardiac diseases covered by the ERN. ♥

Educational meetings for patients: one meeting per ERN country in 2018-2019

Amsterdam, 27 September 2018

To promote patient self-knowledge and self-control, ERN GUARD-Heart member centres are encouraged to organize educational meetings for patients with rare cardiac diseases and conditions. According to the plans of the Network, which have been approved by the EU, 12 topic meeting should be organized for patients before 1st of March 2019 (one meeting per country). A maximum budget of €4000,- will be available for each patient meeting, for example for renting a conference room, catering, or travel costs for ~50 patients or more

ACTION REQUEST: ERN centres are requested to present their plans for a patient meeting to the project management office, preferably before 1st of December 2018.

Meeting agendas and programmes will be published on the website and the bimonthly Newsletters of the ERN. After the patient meeting, a reimbursement form, together with of all the payments, can be sent to the management. ♥

Translation of patient information folders

Amsterdam, 27 September 2018

Patient information folders are available on the ERN [website](#) for the next diseases:

- Brugada syndrome
- Catecholaminergic polymorphic tachycardia
- Long QT syndrome
- Arrhythmogenic right ventricular cardiomyopathy
- Dilated cardiomyopathy
- Hypertrophic cardiomyopathy

These folders are only available in English language. Translation of the folders to other EU languages has to be done by the ERNs themselves. Unfortunately, the quality of translation to the German language by

official translation offices has been disappointing. Juan Ramon Gimeno (*University Hospital Virgen de la Arrixaca*) has successfully translated the folders to Spanish. Eric Schulze-Bahr (*University Hospital Munster*) will translate the folders to German. Both Spanish and German versions of the patient folders will soon become available on the [website](#) of the ERN. A small budget of €600,- is available for ERN members who wish to translate the available folders to another European language.

ACTION REQUEST: ERN GUARD-Heart members who wish to translate the patient folders in an European language are requested to contact the project management office. ♥



Quality of Care

Patient Reported Outcome Measures (PROMs)

Amsterdam, 28 September 2018

Patient reported outcome measures (PROMs) are measures of health status or health-related quality of life as reported by patients. Patient experience and perceptions of their health and wellbeing are important indicators of the quality of service provided by healthcare provider (HCP). Measuring patient experience through standardized and validated PROMs will enable the Network to compare the quality of the services provided by its member HCPs and would support ongoing quality improvement within the Network.

PROMs can generally be divided in two categories: generic or condition-specific. Generic PROMs measure the health status or health-related quality of life of patients regardless of their disease. Condition-specific PROMs aim to capture aspects of health pertinent to specific diseases and conditions, and improvements in those health aspects due to treatment. Generic PROMs aim to measure health status or improvements of health on several domains, including the general health, physical health, mental health, and health-related quality of life. The advantage of generic measures is that they enable comparisons of health status and/or improvements in a commensurate manner across all conditions, therapies and HCPs. The main disadvantage of generic PROMs is that they may sometimes fail to detect changes that are disease- or condition-specific. The main advantage of condition-specific PROMs is that they are usually sensitive to detect change in the disease or condition that is being addressed by the measures. The disadvantage of these measures is that they restrict comparisons of health status and/or improvement to HCPs of similar services. These aspects of generic and specific PROMs provides the basis for the rationale that ideally a HCP should incorporate both generic and condition-specific PROMs to measure patient experiences and perceptions of their health status and wellbeing.

However, currently, standardized & internationally validated PROMs do not exist for the rare diseases and conditions covered by the 3 thematic areas of the Network, i.e., familial electrical diseases in adults and children (thematic area 1), familial cardiomyopathies in adults and children (thematic area 2), and special electrophysiology conditions in children (thematic area 3). In addition, not all member HCPs of ERN GUARD-Heart cover all three thematic areas; this is associated with differences in the services provided by different member HCPs. It is for these reasons that the Network has decided to start measuring patient experience using a core set of standardized and validated generic PROMs.

To select a set of standardized and validated generic PROMs that is best suitable to measure patient experience within ERN GUARD-Heart, the project management office, in collaboration with representatives from the Patient Advocacy Board, used psychometric reviews of generic PROMs, and reviewed a Summary Report of the Organisation for Economic Co-operation and Development (OECD) on "*Measuring generic patient reported outcome measures for health system improvement*". Based on the volume of evaluations, good measurement and operational characteristics, three instruments were presented to ERN Board for further consideration, i.e. SF-36, EQ-5D, and PROMIS GHS.

On the basis of appraisal of evidence, the aspects of health measured & applicability in thematic disease areas covered by the Network, the Network Board selected PROMIS GHS as a suitable instrument to measure patient experiences. In the near future, the project management office will disseminate PROMIS GHS in different European languages to the member HCPs.

In the future, the ERN will add a supplemental set of condition-specific PROMs. To design and study such PROMs, a meeting for experts from different ERN centres will be organized by the ERN in March 2019.





Invitation for ERN members to join RD-Connect



An international platform for rare disease data analysis and data sharing

Amsterdam, 23 August 2018

[RD-Connect](#) is an EU-funded integrated platform which connects databases, registries, biobanks & clinical bioinformatics for rare disease research. It provides user-friendly tools for data analysis and links different data types - omics (e.g., genomics), clinical information, patient registries and biobanks - into a common resource. By enabling scientists & clinicians around the world to analyse and share data & biosamples, RD-Connect speeds up research, diagnosis and therapy development to improve the lives of patients with rare diseases. RD-Connect has built 3 integrated online systems open to any rare disease:

- [Genome-Phenome Analysis Platform \(GPAP\)](#) for analysis & sharing of omics data to diagnose patients and discover new disease genes.
- [Registry & Biobank Finder](#), a global directory of rare disease patient registries and biobanks.
- [Sample Catalogue](#), which helps researchers find rare bio-samples stored in biobanks

RD-Connect is open for data submissions from ERNs and already holds thousands of secure, pseudonymized datasets. Data are linked at an individual per-patient level. Researchers can analyse data, find similar cases and related information such as availability of biomaterials. In collaborations with other EU projects, RD-Connect has already contributed to the discovery of dozens of novel disease genes.

Opportunities for ERNs to use RD-Connect tools

RD-Connect is working together with the ERNs to support them in their research & diagnostic goals. In particular, the RD-Connect Genome-Phenome Analysis Platform can be used by ERN clinicians themselves to solve their unsolved cases. In 2018 & 2019, as part of [Solve-RD](#) project, the RD-Connect Genome-Phenome Analysis Platform will be used by clinicians and researchers from four ERNs for reanalysis of 19.000 undiagnosed exomes towards diagnosis and gene discovery. GENTURIS, EURO-NMD, ITHACA, & ERN-RND are full partners in Solve-RD, with six more ERNs as associated partners. However, all ERNs are invited to submit their sequencing data and analyse them in RD-Connect. If you have a cohort of undiagnosed patients with NGS data you would like support in reanalysing, RD-Connect would love to hear from you!

Join the RD-Connect Community

In July 2018 the new RD-Connect Community was launched. This is an international association of individuals and organizations who would like to participate in shaping the rare disease field, promote data sharing and reuse and advance research. Membership is free of charge and open to organisations, research groups and individuals from diverse backgrounds, engaged in rare disease research anywhere in the world. Several ERNs have signed up as members, but we would be delighted to receive registrations from individual healthcare professionals HCPs and research groups. ♥

Interested in joining? Fill out the [registration form](#) to become a member! For more information about the RD-Connect Community, please [click here](#)



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