

Friday 19 May 2017

EUROPEAN REFERENCE NETWORKS  
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES

Share. Care. Cure.

# ERN



## GUARD-HEART



ERN GUARD-HEART BIMONTHLY NEWSLETTER

YEAR 2017 NUMBER 1

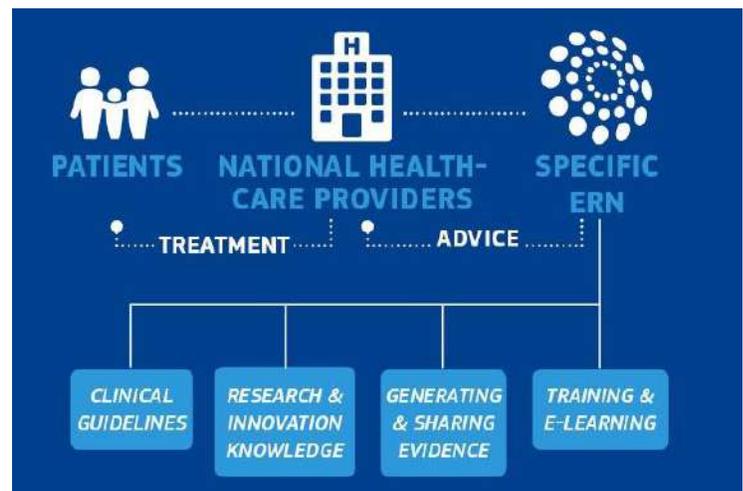
## The Launch of the European Reference Networks

9 March 2017 – Vilnius, Lithuania

European Reference Networks (ERNs) are virtual networks involving healthcare providers (HCPs) across Europe. They aim to tackle complex or rare diseases and conditions requiring highly specialised treatment and concentration of knowledge and resources. To review a patient's diagnosis and treatment, ERN coordinators convene 'virtual' advisory boards of medical specialists across different disciplines, using a dedicated IT platform and telemedicine tools. ERNs offer the potential to give patients and doctors across the European Union (EU) access to the best expertise and timely exchange of life-saving knowledge, without having to travel to another country.

Following the first call for proposals in July 2016, the first ERNs were approved in December 2016 and launched in March 2017. At their inception, the networks comprised more than 900 HCPs located in 313 hospitals in 25 Member States (plus Norway). 24 ERNs are working on a range of thematic issues. Over the next 5 years, ERNs are expected to reinforce their capacities to benefit thousands of EU patients suffering from a rare or complex condition.

The ERN initiative receives support from several EU funding programmes, including the Health Programme, the Connecting Europe Facility and Horizon 2020. EU Member States lead the ERN process: they are responsible for the recognition of centres at national level and endorse the applications. A Board of Member States is responsible for developing the EU ERN strategy and approving the Networks.



Activities of an European Reference Network (ERN)



Coordinators of all 24 ERNs at the kick-off meeting in March 2017 in Vilnius, Lithuania



# ERN on Rare and Complex Diseases of the Heart

## ERN GUARD-HEART



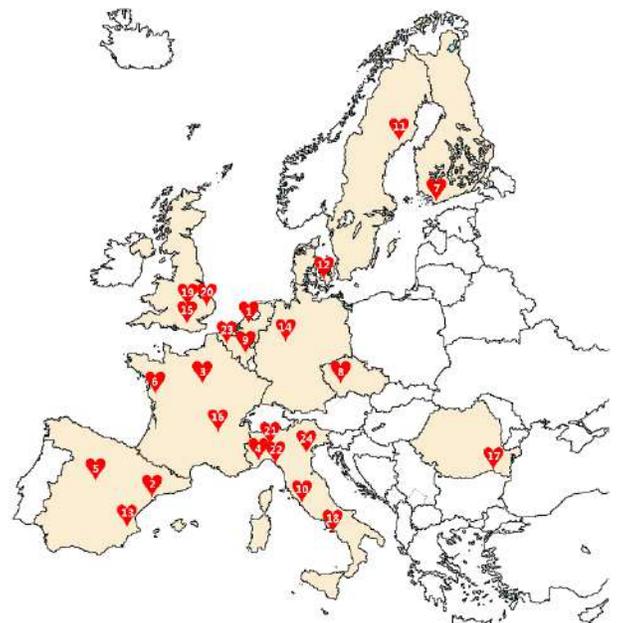
↑ Certificate of the ERN GUARD-HEART. → Dr. Arthur Wilde, Coordinator of ERN Guard-HEART, receives the certificate from Dr. Vytenis Andriukaitis, European Commissioner for Health and Food Safety.



### 9 March 2017 – Vilnius, Lithuania

ERN GUARD-HEART is an ERN on rare and complex diseases of the heart and on cardiac conditions that require highly specialised interventions. The Network, as the ERN is also referred to, was officially launched on 1 March 2017, and the kick-off meeting took place in Vilnius on 9 March 2017. During this meeting, on behalf of the Network, its Coordinator, Dr. Arthur Wilde from the Academic Medical Centre (Amsterdam, Netherlands) gave an introductory presentation to inform and update the Representatives of participating Healthcare Providers (HCPs) and other stakeholders on the mission, structure, governance, the intended methods, and pre-defined deliverables of the Network, and pointed to future opportunities, concerns and possible pitfalls of the ERN initiative.

The size and geographical diversity of ERN GUARD-HEART, which comprises 24 multidisciplinary highly specialised HCPs in 12 EU Member States, guarantees an adequate scale to enable the sharing of expertise and the establishment of cross-border patient pathways. Based on the clinical guidelines of the European Society of Cardiology (ESC), the International Classification of Diseases (ICD10) and ORPHANET, the Network has identified 3 thematic areas: familial electrical diseases, familial cardiomyopathies, and paediatric special conditions. The Network is seeking to strengthen coordination of expertise and resources to >>



BE	CZ	DK	DE	ES	FR	IT	NL	RO	FI	SE	UK
2	1	1	1	3	3	6	1	1	1	1	3

Healthcare Providers (HCPs) in ERN GUARD-HEART & the numbers of HCPs per Member State



>> facilitate pooling of multidisciplinary knowledge. By fostering closer cooperation between experts, new scientific knowledge will be acquired and shared to support the development of new diagnostic and therapeutic procedures.

During the kick-off meeting, Dr. Elijah Behr, the Representative of the HCP St. George's University Hospitals NHS Foundation Trust (London, UK) presented a potential model for the ERN GUARD-HEART, based on the activities of the Association for Inherited Cardiac Conditions (AICC), to deliver education and training programs to European clinicians, interact with national patient interest groups, and to collect data for research and to interact with existing working groups where objectives are shared.

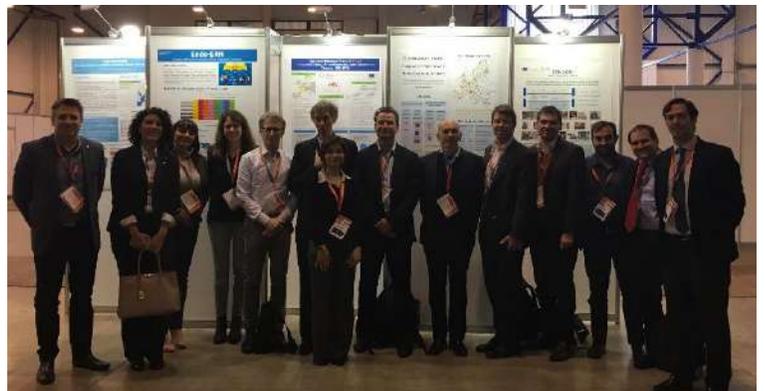
At the meeting in Vilnius, Representatives of the participating HCPs agreed on the importance of constructing a team of patient-representatives for the Patient Advocacy Board, as specified in the ERN grant application, and decided to provide their suggestions with regard to relevant national patient organizations and names of candidate patient representatives in an online google spreadsheet that was made by the Management Office of the Network. In addition to this, to arrange a 2<sup>nd</sup> meeting for the HCP Representatives another Google-spreadsheet was created to identify the availability of the HCP Representatives for future international cardiology conferences. Finally, as establishment of ERN-wide registries of patients with rare and complex heart diseases is one of the main objectives of the Network and the EU, the HCP Representatives agreed upon the initiation of a registry of patients with long QT syndrome types 4 -16, for the purpose of a grant application led by Dr. Georgia Sarquella-Brugada, the Representative of the Hospital Sant Joan de Déu (HSJD) - Barcelona's Children Hospital (Barcelona, Spain) and leader of the thematic area 'paediatric special conditions'.

1 Arthur Wilde (Amsterdam, NL)	● ● ●	13 Juan Ramon Gimeno (Madrid, ES)	● ● ●
2 Georgia Sarquella-Brugada (Barcelona, ES)	● ● ●	14 Eric Schulze-Bahr (Munster, DE)	● ● ●
3 Philippe Charron (Paris, FR)	● ● ●	15 Elijah Behr (London, UK)	● ● ●
4 Silvia Priori (Pavia, IT)	● ● ●	16 Philippe Chevalier (Lyon, FR)	● ● ●
5 Pablo Garcia-Pavia (Madrid, ES)	● ● ●	17 Ruxandra Jurcut (Bucharest, RO)	● ● ●
6 Vincent Probst (Nancy, FR)	● ● ●	18 Guiseppe Limongelli (Naples, IT)	● ● ●
7 Jaana Pihkala (Helsinki, FI)	● ● ●	19 Juan Kaski (London, UK)	● ● ●
8 Jan Janousek (Prague, CZ)	● ● ●	20 Perry Elliot (London, UK)	● ● ●
9 Rik Willems (Leuven, BE)	● ● ●	21 Peter Schwartz (Milan, IT)	● ● ●
10 Fabrizio Drago (Rome, IT)	● ● ●	22 Eloisa Arbustini (Pavia, IT)	● ● ●
11 Annika Rydberg (Umea, SE)	● ● ●	23 Pedro Brugada (Brussels, BE)	● ● ●
12 Jacob Tfelt (Copenhagen, DK)	● ● ●	24 Sabino Iliceto (Padua, IT)	● ● ●

### Thematic Area Expertise

- Familial electrical diseases in adults
- Familial electrical diseases in children and special electrophysiology conditions
- Familial cardiomyopathies in adults and children

↑ List of HCP Representatives of ERN GUARD-HEART with the thematic area expertise of their HCPs. Thematic area leaders are Dr. Georgia Sarquella-Brugada (paediatric special conditions), Dr. Silvia Priori and Dr. Carlo Napolitano (familial electrical diseases), and Dr. Philippe Charron (familial cardiomyopathies).



HCP Representatives of the ERN GUARD-HEART: ↑ in front of the Network poster and ↓ celebrating the launch of the Network. March 9, 2017, Vilnius, Lithuania.





# First Meeting of the ERN Coordinators Group

26 April 2017 – Brussels, Belgium

The first meeting of the Coordinators of the 24 ERNs with representatives from the European Commission and the Board of Member States (representatives from across the EU Member States) was held in Brussels, Belgium, on 26 April 2017. One of the major topics of this meeting was the working methods of the ERN Coordinators Group and to reach an agreement on the chairing of the future Coordinators meetings and to outline the working procedures of the ERN Coordinators Group. During this meeting, the Coordinators agreed to organize 7 Working Groups based on important topics for future discussions with the European Commission and the Board of Member States.

The ERN Coordinators agreed to select a Chair (Spokesman) from each of the Working Groups for future discussions with the European Commission and the Board of Member States. On behalf of ERN GUARD-HEART, the Coordinator, Dr. Arthur Wilde, will participate in the Working Groups on Research and Brexit. The ERN Coordinators Working Group on Research will participate in the three Action Groups of the Horizon 2020 Joint Programme on transnational research on rare diseases (Action 1: research), virtual platform (Action 2: data and information), and data management (Action 3: capacity building). The ERN Coordinators Working Group on Brexit

identifies the need of future discussions with the European Commission about the importance of Healthcare Providers (HCPs) from UK to remain in the ERNs after the completion of Brexit.

Other important topics of this first meeting of ERN Coordinators involved future calls, eHealth tools, cooperation and interaction of ERN with the ERN Board of Member States, and patient informed consent for ERNs, which are discussed elsewhere in this Newsletter.

## The Working Groups of ERN Coordinators

1. IT and data sharing
2. Research
3. Monitoring and assessment system of ERNs
4. Conflict of interest and legal healthcare
5. Capacity building, education, and guidelines
6. Cross-border healthcare
7. Brexit

## First ERN GUARD-HEART Board Meeting at the ESC Congress

As specified in the ERN grant application, Meetings of the Network Board will take place every six months with all members required to attend and discuss strategic and scientific issues. Based on the availability of the majority of the HCP Representatives, the first official meeting of the Network Board will take place on **27 August 2017 (from 13:00 to 17:00)** at the European Society of Cardiology Congress in Barcelona, Spain. Agenda for this meeting will be provided in the Newsletter of the ERN GUARD-HEART in July, 2017.





# ERN Core eHealth Services

## IT Collaborative Platform and Clinical Patient Management System

18 May 2017 – Amsterdam, Netherlands

### IT Collaborative Platform

The IT Collaborative Platform is a restricted online space for ERN communications and administrative document management. Each ERN has a specific work space for its Members to publish (Library), discuss (Forum), schedule (Agenda), vote (Poll), and endorse (Like) communications and documents. ERN Coordinators have the Moderator role, which means that they can contribute content directly or provide the ERN IT Expert Group with content to populate the online space by emailing the support mailbox (available from the link 'Support/Help' on the homepage). To access the IT Collaborative Platform, the next steps should be followed:

1. Create an EU Login using the link:  
<https://webgate.ec.europa.eu/cas/>
2. Go to the link:  
<https://webgate.ec.europa.eu/ern/>  
and use your EU Login to enter the Platform
3. Click on 'Access/request access to my network' Requests for membership will be authorized by the ERN Coordinator or your HCP Representative (or the Substitute Representative of your centre).

### Clinical Patient Management System

The Clinical Patient Management System (CPMS) is a system to enable specialists from ERNs to exchange information to diagnose and treat patients. The European Commission has contracted OpenApp/Vitro to provide Software as a Service (SaaS) for the CPMS and the delivery of the system is scheduled for September 2017. 24 Independent systems for each specific ERN with consistent underlying design will be provided. CPMS can be used to transfer patient files between separate ERNs and between HCPs within each ERN. Multiple ERN specialists (medical or non-medical) can contribute to the assessment of patient files (including ECGs, echocardiograms, microscopy and radiology files) through comments, formal contributions, and reviews of the outcome documents. Participation is possible through comments or panel meeting via live video conferences. When cases are closed, they can be anonymized for research.

The CPMS will be tested by the ERNs before roll-out. Patients will have no direct access to the CPMS (only authorized ERN Members). The EU Login system (see *IT Collaborative Platform*) will be used for authentication. Requests for access will be authorized by one designated ERN Member. The CPMS will have a mandatory minimum patient data set. Patient data will be pseudonymized before entering CPMS. A patient consent form, designed by the European Commission in collaboration with the ERN coordinators (for both treatment/care and/or research), will be required for patient data uploads. To use the CPMS, each HCP must obtain approval from its local ethics committee. Further details will be provided in advance to allow the ERN coordinators and ERN Members adequate time to prepare and cross-check with the hospital procedures.



The IT Collaborative Platform for the ERN GUARD-HEART



# One Patient Consent Form For ERN-Related Care & Research

26 April 2017 – Brussels, Belgium

To discuss a patient within a specific ERN with healthcare professionals of the participating Healthcare Providers (HCPs) for clinical purposes (e.g., to develop diagnosis and management plan), consent of the patient is required. Consent should be acquired before ERN(s) are consulted for advice and before patient data are shared through the ERN Clinical Patient Management System (CPMS). A consent form will allow the referring ERN healthcare professional to make available any of the data stored in the healthcare record that would help the ERN to discuss the patient. In addition, to improve future knowledge on rare diseases, ERNs are dependent on databases of patient information. Databases, or registries, should contain only coded patient information. Such databases can be used for current or future research projects by medical researchers in the ERN. To help build the databases, patients can provide consent to their data being added to such a database.

In collaboration with the Coordinators of 24 ERNs, the European Commission has designed one single consent form for data sharing in ERNs for patient care and creation of rare disease registries. A template of this consent form was presented to the Coordinators at the first Coordinators meeting in Brussels, Belgium, in April 2017. At the next meeting of the Network Board of ERN GUARD-HEART, the consent form will be presented to all HCP Representatives for discussion.

*The 2<sup>nd</sup> page of the consent form template for ERN-related care and research. The 1<sup>st</sup> page contains relevant background information for patients.*

## ERN Websites

### A general ERN website

A single general public website has been launched for ERNs that seeks to raise awareness on the existence of the ERNs and to communicate outcomes. It is for the general public and will provide information about the ERNs, share knowledge with patients and general public, (listing members, characteristics, evaluation, activity, etc.) and to publish the public deliverables of the ERNs (e.g., clinical guidelines, patient guidance, etc.). To visit the website, use the link: <https://ec.europa.eu/health/ern/>

### ERN GUARD-HEART Website

The European Commission has created a web space for GUARD-HEART (<http://guardheart.ern-net.eu/>). The Project Management Office of ERN GUARD-HEART is currently designing the layout of the website. The website is expected to be launched in August 2017. It will be a public-facing website for the Network to publish content. After the launch, Network members can request a personal e-mail (e.g., [firstname.lastname@guardheart.ern-net.eu](mailto:firstname.lastname@guardheart.ern-net.eu)). These e-mails will be accessible via the webmail at <https://ern.awsapps.com/mail/> and also via Microsoft Outlook or smartphones.

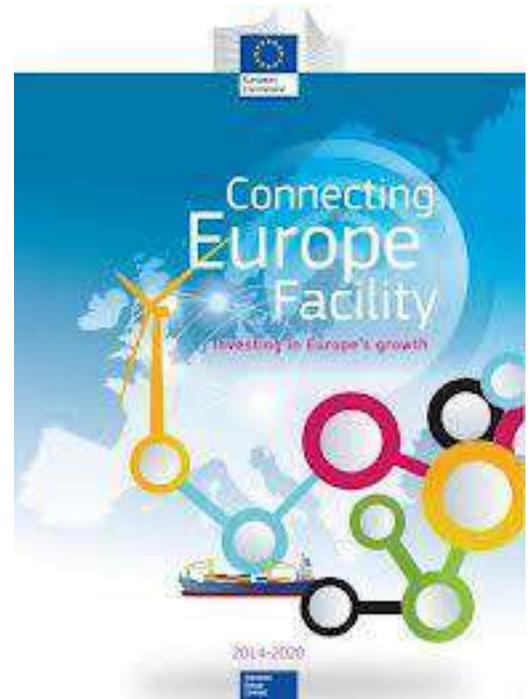
## Future EU Calls

26 April 2017 – Brussels, Belgium

### Connecting Europe Facility TELECOM 2017 Calls for Proposals

On 6 May 2017, the Innovation and Networks Executive Agency (INEA) and European Commission launched the second Connecting Europe Facility (CEF) Telecom call for 2017. The call makes funding available to stimulate and support the deployment of European Digital Service Infrastructures (DSIs). Within the eHealth area, the call makes €3 million available for facilitating the connection between members of ERNs by ensuring adequate and efficient use of the ERN core eHealth services (i.e., IT Collaborative Platform and the Clinical Patient Management System) by members of the ERNs. The maximum amount of funding for each proposal is € 125.000,- for a period of one year. The funding may be used for the establishment of an IT technical helpdesk, training in ERN eHealth services, purchase of hardware and/or software necessary for the implementation of the ERN eHealth services in an ERN member HCP. Interested applicants can submit proposals at the latest by 21 September 2017. Since only one application per ERN is accepted and the application should be signed by the Coordinator of the Network, applications should be first communicated with the HCP Representative and the Management Office ERN-GUARD-HEART. For more information on the call, please visit:

<https://ec.europa.eu/inea/en/connecting-europe-facility/cef-telecom/apply-funding/2017-cef-telecom-call-ehealth-cef-tc-2017-2/>



### Calls for new ERN Members

Calls for healthcare providers wanting to join existing ERNs will be launched by the European Commission for Health and Food Safety at the end of 2017.

### Calls for new ERNs

Although not yet officially announced, the European Commission for Health and Food Safety is considering to launch calls for healthcare providers wanting to initiate an ERN on rare diseases outside the scope of existing ERNs in mid-2019.

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## Workshops for ERN-Related Activities

18 May 2017 – Amsterdam, Netherlands

ERN-related workshops are organized by RD-ACTION. RD-Action is an EU Health Programme funded Joint Action, promoting implementation of recommendations on policy, information and data for rare diseases. RD-ACTION is supported by the European Commission to rare diseases, through its Directorate General for Health (DG SANTE). A key objective of the RD-ACTION Policy is to provide support to rare disease community in conceptualising, implementing and evolving robust ERNs capable of meeting the needs and expectations of people living and working with conditions requiring a specific concentration of expertise.





## **Previous ERN-Related Workshops**

### **20 March 2017 - Valletta, Malta**

#### ***Integrating Research and Healthcare for Rare Diseases: a structured cooperation with high community added value.***

The launch of 24 ERNs is expected to have a radical impact on the provision of care for patients with rare diseases and the development of research. There is a major interest for collaboration between rare diseases researchers in Europe and beyond. The objectives of this meeting were 1) to outline strategic organisation and performance of research activities to achieve a higher level of integration, optimisation of resources and synergy between EU research infrastructure and healthcare, 2) to design operational proposals to address the challenges of prompt translation of innovative health technologies from research to care, cooperation for training of healthcare providers and establishing of disease registries, and 3) to identify instruments and solutions to ensure longer-lasting support to rare disease research in an integrated manner. At this workshop, synergies within and across the research and healthcare for rare diseases were presented, as well as gaps and existing challenges. Specific focus was paid on the infrastructural potential of ERNs where research for rare diseases can be developed and strategies for integrating research and healthcare activities in a sustainable manner can be designed.

*ERN GUARD-HEART was represented at this workshop by Dr. Connie R. Bezzina, molecular geneticist and ERN GUARD-HEART member from the Academic Medical Centre (AMC), Amsterdam, Netherlands.*

### **21-22 March 2017 - Madrid, Spain**

#### ***Rare Diseases Registries***

The workshop will brought together ERNs and other key stakeholders for the development of patient registries for rare diseases. The workshop was a unique chance to foster knowledge exchange and best practice of the European rare disease registries, to reinforce research and epidemiological surveillance through setting up of shared registries, to maximize integration and interoperability of European and national rare disease registries, and to highlight the registries linked to ERNs.

*ERN GUARD-HEART was represented at this workshop by Dr. Juan Ramón Gimeno, cardiologist and Representative of the University Hospital Virgen de la Arrixaca, Murcia, Spain.*

### **26-27 April 2017 - Brussels, Belgium**

#### ***Using standards and embedding good practices to promote interoperable data sharing in ERNs***

ERNs are first and foremost dedicated to care. Once established and connected by IT Platform and Clinical Patient Management System, ERNs will be able to exchange knowledge and expertise between their Member HCPs. It is important to emphasise that wherever possible (and appropriate), expertise will travel rather than the patients themselves. Data therefore sits at the centre of this, and there are certain standards, ontologies and principles one can apply to rare disease data to optimise its value by making it interoperable and able to serve a multitude of additional purposes, besides enabling a virtual consultation for the most complex patients. ERN communities will be key generators and handlers of rare disease data, and as such it is important for ERN Members to be aware of the potential to embed 'approved' and highly recommended rare disease ontologies for the capture and exchange of certain types of data. Shared consensus guidance is important to support ERNs and ensure a baseline compatibility and interoperability between the ERNs. This workshop introduced ERN Members to 1) the ORPHANET nomenclature and ORPHACODE classification system for rare diseases, 2) ORPHANET Knowledge Management System to enable ERN clinicians to contribute to the future population of ORPHANET database, 3) the Human Phenotype Ontology (HPO) as an optimal ontology for phenotypic descriptions for rare diseases, and 4) the concept of Privacy-Preserving Record Linkage (PPRL) as an example of good practice. Finally, the FAIR data approach was introduced for future research activities by the ERNs. In the FAIR data approach, data should be: Findable (easy to find by both humans and computers), Accessible (stored such that they can be easily accessed with well-defined license and access conditions), Interoperable (ready to be combined with other datasets by humans or computer systems), and Reusable (ready to be used for future research).

*ERN GUARD-HEART was represented at this workshop by Dr. Ahmad S. Amin, cardiologist and project manager of ERN GUARD-HEART, from the Academic Medical Centre (AMC), Amsterdam, Netherlands.*



## **Future ERN-Related Workshops in 2017**

**1-2 June 2017 – Newcastle, UK**

### ***Indicators and Outcome Measures to demonstrate the impact and added value of ERNs***

*Context:* ERNs should identify relevant Quality and Outcome Indicators across their disease-specific Networks. Alongside, clinical Performance (Impact) Indicators common to the work of all ERNs should be collected. The selection of such key Indicators and how they are captured will be of crucial importance to the future success of ERNs. This workshop will address a number of key questions, including how ERNs can demonstrate their added value, what can be measured (in terms of Indicators), and how ERNs embed a culture of knowledge generation through the implementation of outcome measures that are centred upon learning through complex case studies? *ERN GUARD-HEART will be represented at this workshop by Dr. Ahmad S. Amin, cardiologist and project manager of ERN GUARD-HEART, from the Academic Medical Centre (AMC), Amsterdam, Netherlands.*

**27 September 2017 – Barcelona, Spain**

### ***Exploring structured collaboration between ERNs and Biopharmaceutical Industry to support increased access to orphan therapies***

*Context:* ERNs have the potential to make a meaningful contribution to improve access to rare disease therapies by stimulating the development of new therapies, supporting trials, enabling the collection of post-marketing data and real-world evidence, and streamlining the process by which existing therapies reach patients who need them. ERNs also possess the potential to favour the generation of less expensive, more reliable, quality EU-wide data. This workshop will focus on questions of data collection, i.e., how to create a system of information within ERNs which is sufficient and adequate for clinicians and the Industry to comply with their regulatory obligations, and on issues around working effectively with the Industry to optimise opportunities while avoiding any conflict of interest. *The member representing ERN GUARD-HEART at this workshop will be selected in the near future.*

**(2 days; either 4,5,6,7) December 2017 – Rome, Italy**

### ***How can ERNs generate, appraise and utilise clinical practice guidelines to enhance the impact and deployment of consensus guidelines in national health systems***

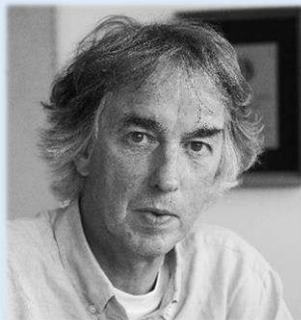
*Context:* The aim of this workshop is to create a harmonized 'dictionary' of terminology for ERNs to use in relation to guideline generation/implementation, to confirm the role ERNs should play in the generation, appraisal, use and dissemination of guidelines, what methodology should be used by the ERNs when elaborating guidelines for rare diseases, how to promote good practices in relation to generating patient-friendly guidelines, how ERNs can utilize existing relevant tools and resources, how often guidelines should be revised and updated, how ERNs may evaluate the impact of guidelines (e.g., in the healthcare system), and what does the 'status' of ERN best practice guidelines hold within individual Member States, i.e. how will national health systems utilise these outputs?

*The member representing ERN GUARD-HEART at this workshop will be selected in the near future.*

## **Project Management Office**

The Project Management Office of ERN GUARD-HEART coordinates the activities of the Network and the communication between Network members, European Commission, RD-ACTION, other ERNs and stakeholders. It also supports the Network in administrative tasks, organisation of meetings, designing and updating of the Network website, and creation and dissemination of the Newsletter.

For information and suggestions, and to provide content for the future issues of this Newsletter, please contact:



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