

Thursday 28 November 2019

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

Share. Care. Cure.

ERN

GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart



ERN GUARD-HEART BIMONTHLY NEWSLETTER

YEAR 2019 NUMBER 6

Fellowship Clinical Genetics Laboratory of Brussels

Brussels, 25 November 2019

One of the goals of ERN GUARD-Heart is to share and spread knowledge and evidence in the diagnosis and treatment of rare and complex heart diseases within and outside the Network. To achieve this goal, the Network will organize on an annual basis two specialized training programmes for healthcare professionals. One of these programmes is a 4-6 months fellowship (in 2020, exact time is flexible) which will provide residents or young specialists the opportunity to gain knowledge and expertise in the interpretation of next generation sequencing (NGS) and OMICs data in the field of cardio-genetics. The fellowship will be hosted by the Clinical Genetics Laboratory of the University Hospital in Brussels, and will be coordinated by Professor Sonia Van Dooren (Clinical Laboratory Geneticist and representative member of Univ. Hosp. Brussels in the ERN GUARD-Heart) and Professor Carlo De Asmundis (cardiologist and representative member of Univ. Hosp. Brussels in the ERN GUARD-Heart). Funding is foreseen for maximum of 5.000,- EURO (housing/ traveling). The salary of the fellow will have to be funded by his/her own institute.

Healthcare professionals, for example cardiologists, cardiologists in training, clinical geneticists and scientists, clinical geneticists or scientists in training, related disciplines working in a medical centre in the European Union and who are **interested in this fellowship program, are requested to write a motivation letter and CV (resume)** to the Project Management Office of ERN GUARD-Heart (n.hofman@amsterdamumc.nl) **before 21 December 2019**. Candidates should master the English language and have an official degree in Medicine, and a degree (or training) in Cardiology, Human Genetics or a related field. ♥

7th ERN GUARD-Heart Board Meeting

21 February 2020 (8.30 – 14.30)

ROME

*Parlamentino Room (Bambino Gesù) in
Palazzo Alicorni*





European Conference on Rare Diseases & Orphan Products (ERCD)

Amsterdam, 15 November 2019

THE 10th EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS

Stockholm, 15-16 May 2020

THE JOURNEY OF LIVING WITH A RARE DISEASE IN 2030

JOIN US AT THE LARGEST PATIENT-LED RARE DISEASE EVENT TAKING PLACE IN EUROPE

Leading, inspiring and engaging all rare disease community stakeholders to take action

+850 attendees
including patient advocates, healthcare professionals, healthcare industry, academics, regulators, payers and policy makers

2 days
15-16 May 2020
Stockholm, Sweden

Worldwide attendance

#ECRD2020

Over 100
expert chairs, speakers and panelists

PROGRAMME COMMITTEE CO-CHAIRS

Prof. Milan Macek
Professor of Medical and Molecular Genetics, Motol University Hospital and Charles University Prague, Czech Republic

Maria Montefusco
President, Rare Diseases Sweden

Violeta Stoyanova-Beninska
Chair, Committee of Orphan Medical Products, European Medicines Agency

Organised by **EURORDIS**

Co-organised by **orphanet** and **Inserm**

With the support of **AFMTELETHON** and the **European Union**

LEARN - NETWORK - INSPIRE

- Co-create the policies and services of the future to support measurable and improved health outcomes for people living with a rare disease
- Learn about how care for rare diseases is organised across Europe, what are the trends, innovations and emerging models of care
- Share/exchange your knowledge and experience with other fellow clinicians from all around Europe and beyond to create collaborations
- Meet patients from other countries and learn from their experiences
- Showcase and share your work with a wide, multi-stakeholder audience as a poster presenter or as a speaker in the open microphone plenary session
- Exchange views on technological developments that are changing the rare disease landscape and contribute to debates on the opportunities, ethical, legal and social implications in areas such as data sharing and artificial intelligence
- Learn about the important progress made to advocate for rare diseases as a health and human rights priority at the United Nations and international agencies
- Learn more about growing efforts to advocate for organised and integrated social support and patient-centred care at the national level
- Gain insight into European and national rare disease policies
- Obtain medical credits by attending ECRD 2020

LANGUAGES
Simultaneous interpretation and live streaming of the Opening and Plenary sessions on 15 May 2020 will be available in:

ENGLISH | FRENCH | GERMAN

For more information, please visit www.rare-diseases.eu

The ERN GUARD-Heart is an Associate Partner of the 10th ECRD, taking place on 15-16 May 2020 in Stockholm, Sweden. The overarching theme is the rare disease patient journey in 2030. The website is now live: <https://www.rare-diseases.eu/>

Deadline for poster submission is 10 February 2020. Registration for this event will open soon. EURORDIS-Rare Diseases Europe is offering patient fellowships for 40 patients advocates to attend this conference. These fellowships aim to empower patient advocates by offering a platform for networking opportunities, access to information and sharing experiences. More information (and the link to apply for this fellowship) can be found on the website of EURORDIS. ❤️

Survey on approach to postmortem genetic testing

Amsterdam, 13 November 2019

We would like to ask your participation in a survey which aims to explore post-mortem genetic testing practices internationally and how healthcare professionals think about the approach implemented in their country. This study is initiated by Lieke van den Heuvel, psychologist and PhD student in inherited heart disease in Amsterdam, and Jodie Ingles, associated professor/genetic counsellor in the Centenary Institute, Sydney, Australia. This survey will also count as an ERN benchmark study, therefore a complete response is of importance. Please follow the link and include your valuable information before the 1st of January <https://redcap.sydney.edu.au/surveys/?s=9KKX8F3PH3> ❤️



ERN expansion: procedure after call is closed

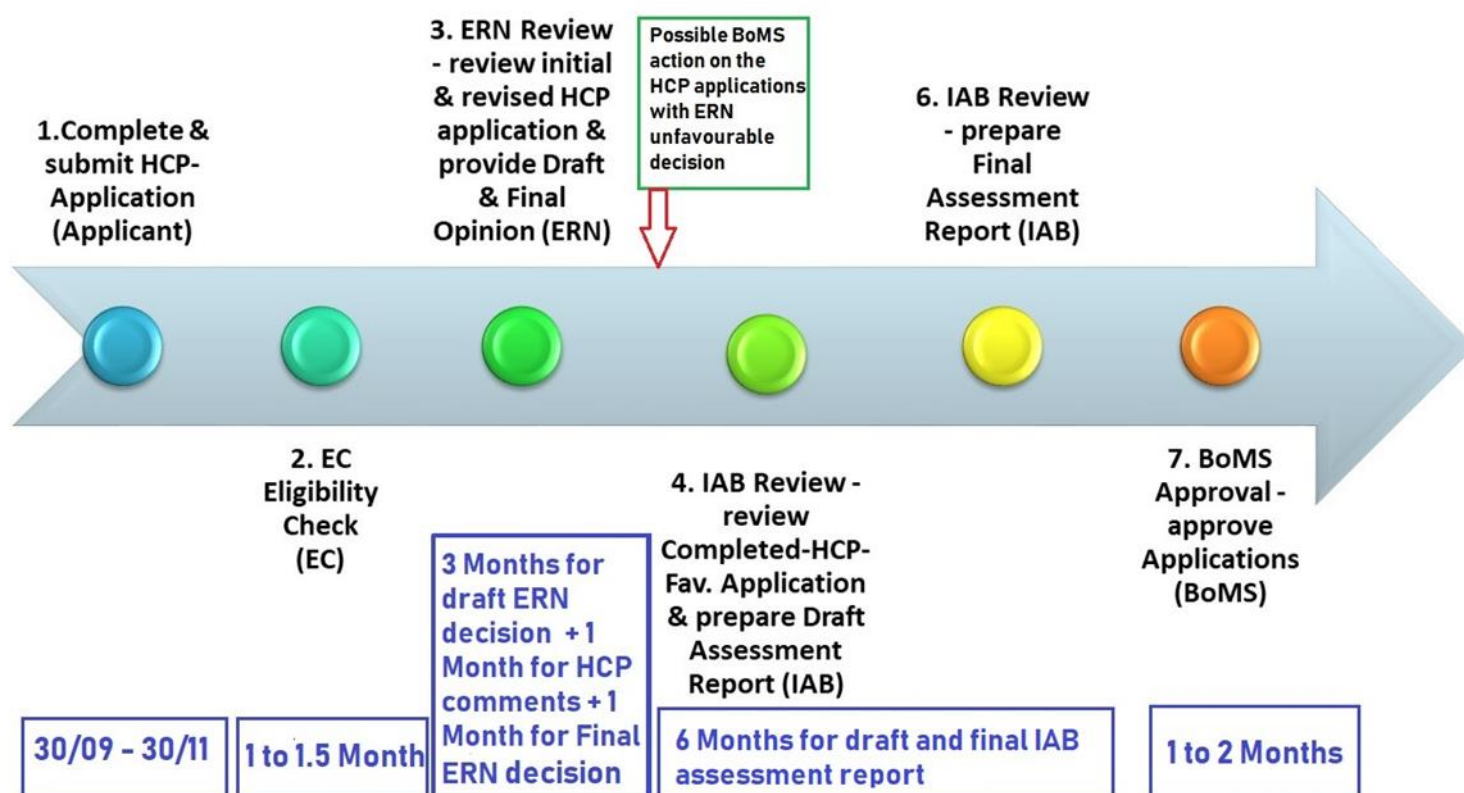


Amsterdam, 26 November 2019

The call for HCPs to join existing ERNs will close on 30 November 2019. The procedure to final approval of candidate HCPs by the Board of Member States is summarized in the workflow diagram below. The board of the ERN GUARD-Heart is involved in this procedure by reviewing the applications (step 3 in the workflow below), of which a large number is expected. Officially, the board consists of a representative of all full members. To facilitate this process and make it more efficient, we propose to mandate the coordinator and the thematic area leaders (and Eric Schulze Bahr as the only representative of Germany, in light of the expected large number of applications from Germany) to provide the draft and final opinion of the ERN. According to EU regulations, all board members need to agree with this proposal and therefore we will forward a separate email on this topic to the respective representatives of each full member HCP, which subsequently needs a reply from each full member representative. ♥

WORKFLOW

The following diagram provides an overview of the process from the call for interest to the final approval by the Board of Member States (BoMS).





ERN monitoring: complete 2019 year data collection

Amsterdam, 26 November 2019

At the end of October 2019, ERN GUARD-Heart submitted the required data for the year 2018 to the IT-tool of the European Commission. The data were required for the data collection process for the indicators in ERN monitoring system. 22 of 24 (92%) full member HCPs returned the data in time. The complete data for 2019 has to be submitted before the end of February 2020. Soon, all full member HCPs will receive forms for this. Please prepare for this where possible. ♥

European Joint Programme on Rare Diseases (EJP RD)

A pre-announcement for the call for proposals for 2020 was received from EJP RD. The aim of the call is to enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with a clear future benefit for patients. Proposals should focus on pre-clinical research to develop effective therapies for rare diseases. The call is scheduled to open on December 13, 2019. Further information and the official publication of the call will soon be available on the website of EJP RD: <http://www.ejprarediseases.org> ♥

Latest ERN GUARD-Heart Publications

1. Towbin JA, McKenna WJ, Abrams DJ, Ackerman MJ, Calkins H, Darrieux FCC, Daubert JP, de Chillou C, DePasquale EC, Desai MY, Estes NAM 3rd, Hua W, Indik JH, Ingles J, James CA, John RM, Judge DP, Keegan R, Krahn AD, Link MS, Marcus FI, McLeod CJ, Mestroni L, Priori SG, Saffitz JE, Sanatani S, Shimizu W, van Tintelen JP, Wilde AAM, Zareba. 2019 HRS expert consensus statement on evaluation, risk stratification, and management of arrhythmogenic cardiomyopathy. *Heart Rhythm*. 2019;16(11):e301–e372. doi:10.1016/j.hrthm.2019.05.007
2. Padrón-Barthe L, Villalba-Orero M, Gómez-Salineró JM, Domínguez F, Román M, Larrasa-Alonso J, Ortiz-Sánchez P, Martínez F, López-Olañeta M, Bonzón-Kulichenko E, Vázquez J, Martí-Gómez C, Santiago DJ, Prados B, Giovinazzo G, Gómez-Gaviro MV, Priori S, García-Pavia P, Lara-Pezzi E. Severe Cardiac Dysfunction and Death Caused by Arrhythmogenic Right Ventricular Cardiomyopathy Type 5 Are Improved by Inhibition of Glycogen Synthase Kinase-3β. *Circulation*. 2019;140:1188-1204. doi:10.1161/CIRCULATIONAHA.119.040366.

Upcoming Cardiology Events

Patient and Experts PLN Meeting

28 November 2019

Murcia – Spain

Boardmeeting ERN GUARD-Heart

21 February 2020

Rome - Italy

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



ERN GUARD-Heart
Project
Management
Office



Arthur A.M. Wilde
Network Coordinator
Academic Medical Center
Amsterdam, Netherlands
Email: a.a.wilde@amc.nl



Nynke Hofman
Project Manager
Academic Medical Center
Amsterdam, Netherlands
Email: n.hofman@amc.nl
Mobile: +31622046435



Ahmad S. Amin
Project Manager
Academic Medical Center
Amsterdam, Netherlands
Email: a.s.amin@amc.nl
Mobile: +31614471713