Wednesday 7 April 2021

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

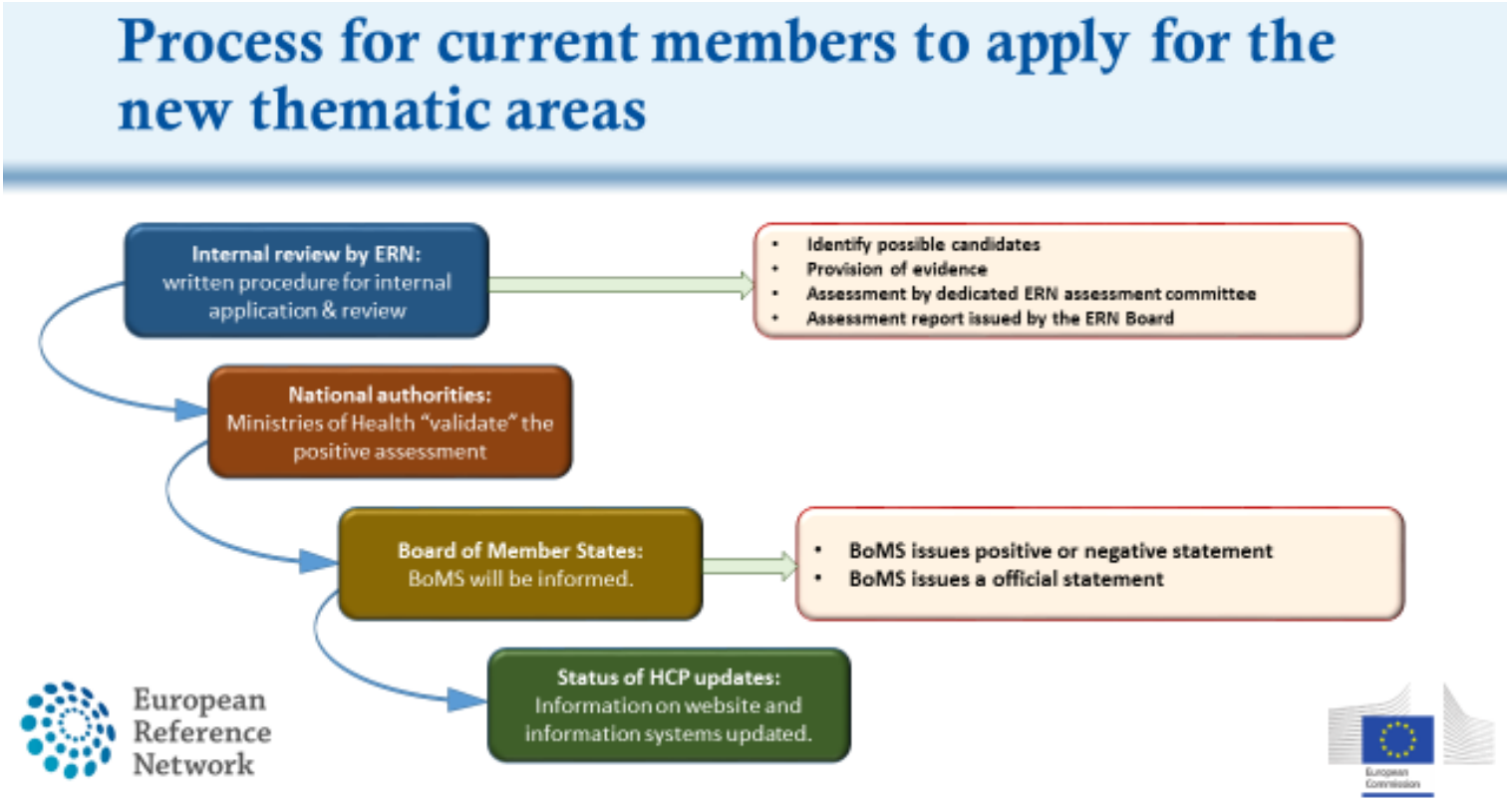
Gateway to Uncommon And Rare Diseases of the Heart

ERN GUARD-HEART BIMONTHLY NEWSLETTER YEAR 2021 NUMBER 2

New thematic area's: where are we now?

Amsterdam, 31 March 2021

The internal applications from the existing full members of ERN GUARD-Heart to join the new thematic areas 'Congenital Heart Disease' and 'Other Rare Heart Disease' have been assessed. All applicants have received the outcome of this assessment. When an application has received a favorable opinion, the HCP has to send their 'National Certificate/Letter for Centre of Excellence' in the specific thematic area to the project management office of the ERN (n.hofman@amsterdamumc.nl). The new thematic areas will start their work as soon as the assessment procedure of new full ERN members has been completed by the European Commission. Presently, the new candidate full members are in the process of a documental review by the indendent assessment body "Andalusian agency for Healthcare Quality". ♥



Online Board meeting

Amsterdam, 9 March 2021

Our 9th board meeting was held online on Tuesday 9 March 2021. Although it is not always easy and exciting to meet online, and it may be challenging to brainstorm on new ideas, the project management team was grateful with the participation of 37 HCP representatives (including 20 representatives from the 21 full ERN members, 6 representatives from 11 affiliated partners, and 6 patient representatives). The notes will be soon available on the IT-platform. ♥

AMEQUIS - project

Amsterdam, 31 March 2021

The AMEQUIS project, which aims to develop an integrated Assessment, Monitoring, Evaluation and Quality Improvement system (AMEQUIS) for ERNs, has started recently. The project is being conduced by an external research group. Last week, this AMEQUIS group has distributed a survey among ERN coordinators and a selection of HCPs to collect their input on the assessment, the monitoring and the evaluation of ERNs and their individual HCPs.
On Monday 12 and Tuesday 13 April, an online stakeholder conference will be organized in which the AMEQUIS has invited two representatives (one 'general' and one 'patient' representative) per ERN. The representatives are expected to provide input to the different steps of the quality improvement system for ERNs. ♥

Patient folders

Amsterdam, 01 April 2021

So far, ERN GUARD-Heart has produced 6 different informative folders for patients affected with on the most common rare diseases (i.e., LQTS, CPVT, BrS, HCM, DCM, ACM). These diseases fall in the scope of the 3 existing thematic areas of the ERN. The folders, at least some of them, are currently available in the following languages: Czech, Danish, Dutch, English, Finnish, French, German, Italian, Romanian, Spanish, and Swedish. The folders are available on the website of the ERN GUARD-Heart. One of our affiliated partners from Lithuania has shown interest to translate the folders in the Lithuanian language. It would be great if we could expand the number of translations with the next languages: *Bulgarian, Croatian, Estonian, Greek, Hungarian, Irish, Latvian, Maltese, Polish, Portuguese, Slovak, and Slovenian*. There is a modest financial budget available (€100,- per folder). For more information and coordination of the translations, please contact the ERN management office (n.hofman@amsterdamumc.nl). ♥

FHL1-registry in production

Bucharest, 15 March 2021

The FHL1 cardiomyopathy registry is now open. The registry, initiated by Ruxandra Jurcut, aims to gather more solid data on a gene with mutations leading to a specific type of hypertrophic cardiomyopathy, generally associated with muscular disease.
As each centre within the ERN may provide care to one or a few families with a mutation in the *FHL1* gene, it will be of extreme value to join forces, put together the data, and characterize this population.

→ To add your patients to this REDCap registry, the registry should be added to your REDCap account, or if you yet don't have an account, a new account should be created for you. This can be done very easily. Please send an email to Ruxandra Jurcut (rjurcut@gmail.com) and provide the number of patients you may wish to add to the registry please put Erik van Iperen in the c.c. as he will create the account for you: e.p.vaniperen@amsterdamumc.nl ♥

Activities of ERN members: educational video's

Amsterdam, 31 March 2021

As discussed during the latest Board meeting, the ERN GUARD-Heart aims to continue with the production of educational video's for healthcare professionals and patients. According to what has been proposed and discussed, the project management team will contact the HCP representatives for the details of the format and the time frame. Please check the planned activities in the list below and contact us (n.hofman@amsterdamumc.nl) with questions, additional information or additional ideas. All the delivered videos are available on the website and the YouTube channel of the ERN. This list only includes the videos for healthcare professionals. In the following Newsletter, the educational videos for patients will be discussed. ♥

Disease / topic	Title	Speaker	Available
LQTS	*Diagnosis and Risk of LQTS*	*Schwartz*	✓✓
LQTS	*Treatment of LQTS*	*Schwartz*	✓✓
LQTS	*Genetic basis of LQTS*	*Schwartz*	✓✓
CPVT	*Diagnosis and Risk of CPVT*	*Wilde*	✓✓
CPVT	*Treatment of CPVT*	*Wilde*	✓✓
CPVT	*Genetic basis of CPVT*	*Wilde*	✓✓
BrS	**Brugada syndrome in children**	**Sarquella-Brugada**	
BrS	**Genetic aspects of BrS**	**Behr / Probst**	
BrS	**Clinical aspects of BrS**	**Behr / Probst**	
SQTS	**…**	**Priori / Mazzanti?**	
SND	**…**	**Schülze-Bahr**	
HCM	*Definition, aetiology, diagnostic work-up*	*Garcia Pavia*	✓✓
HCM	**management / therapy**	**Gimeno**	
DCM	*Definition, aetiology, diagnostic work-up*	*Charron*	✓✓
DCM	**management / therapy**	**Henning Bundgaard?**	
ARVC	**Definition, aetiology, diagnostic work-up**	**Corrado (Padua)?**	
ARVC	**management / therapy**	**Estelle Gandjbakhch?**	
LV non-compaction	**…**	**Jurcut?**	
Neuromuscular disorders	**…**	**Karim Whabi?**	
Fabry diseases	*…*	*Jurcut*	✓
TTR amyloidosis	**…**	**Garcia Pavia?**	
Devices	**…**		
NGS and interpretation of variants	**…**	**Arbustini?**	
Sudden Cardiac Death	*…*	*Tfelt-Hansen*	✓✓
Genetic counselling	**…**	**Baban**	
Predictive testing	**…**		
Management of pregnancy in channelopathies	**…**	**Denjoy**	
Management of pregnancy in cardiomyopathies	**…**	**Bauce**	
Management of sport	**…**	**Crotti**	
Role of psychology support	**…**		
Role of pathologist in SCD	**…**		
Myocarditis	**…**	**Caforio**	
Cardiac Tumours	**…**	**Ereminiene**	

ERN Research – ERICA Project

European Rare Disease Research Coordination and Support Action

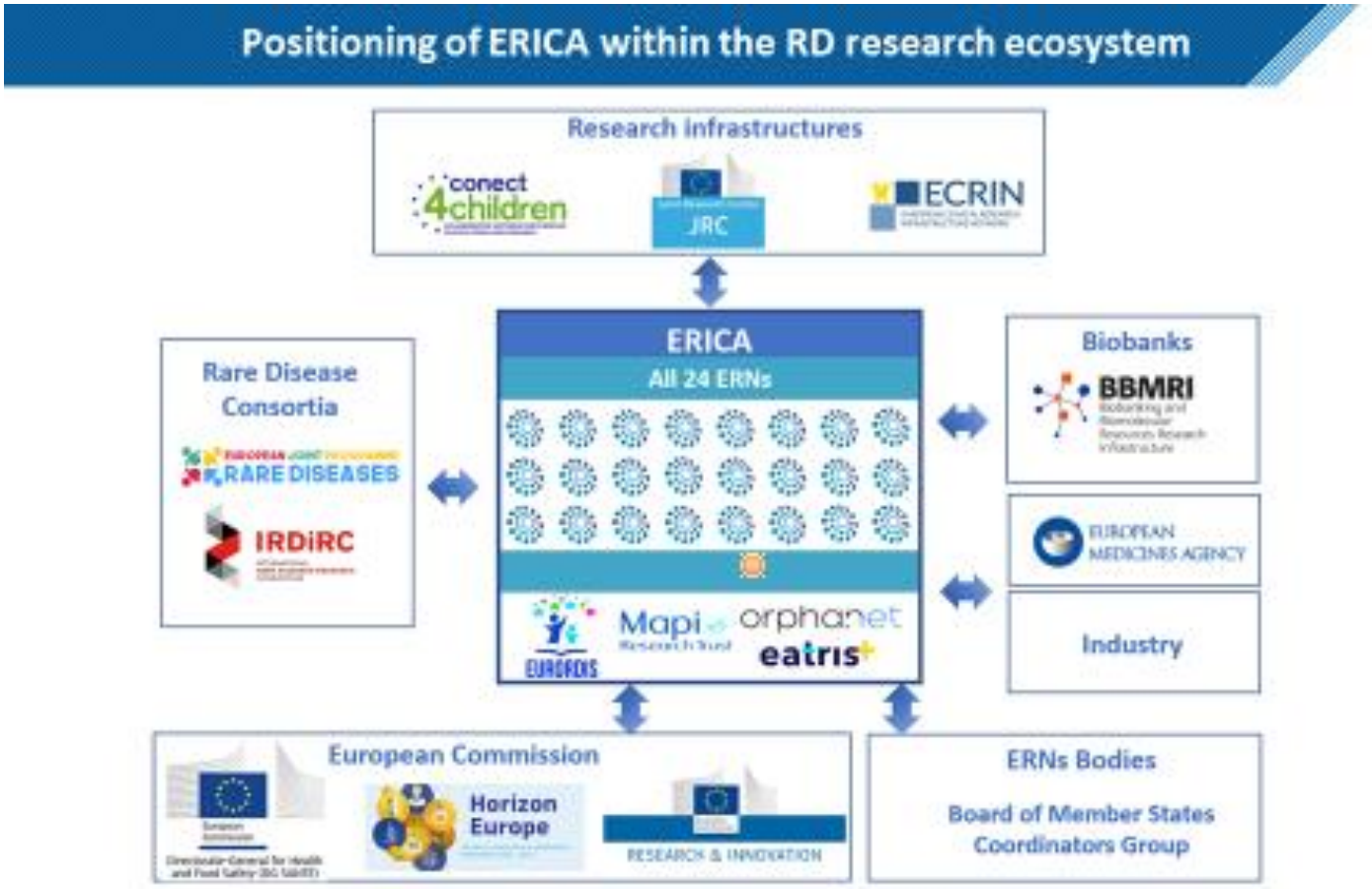
Leiden, 01 March 2021

The ERICA project (European Rare Disease Research Coordination and Support Action) started on 1 March 2021. ERICA is a platform dedicated to coordinating clinical research among the ERNs and their interaction with the EJP RD, the international rare disease research consortium (IRDiRC) and the EU-based research infrastructures. The ERICA consortium consists of 29 partners: all ERNs, EJP RD, Orphanet, Map Trust Research, and EATRIS. The overall objective is to increase the research and innovation potential of ERNs through facilitating inter-ERN collaboration and increase the visibility and impact of ERNs. Through knowledge sharing, engagement with stakeholders in the rare disease domain and assembly of transdisciplinary research groups working across the global health spectrum, ERICA strives to reach the following goals:

- new intra- and inter-ERN rare disease competitive networks;
- effective data collection strategies;
- better patient involvement;
- enhanced quality and impact of clinical trials;
- increased awareness of ERNs innovation potential.

ERICA will strengthen research and innovation capacity by the integration of ERN research activities, outreach to European research infrastructures to synergistically increase impact and innovation. This will result in efficient access and safe therapies for the benefit of patients suffering from rare diseases and complex conditions.An ERICA Kick-off meeting with the first General Assembly will be held on Thursday 27th – Friday 28th May 2021. All the ERNs are invited to assign representatives for the ERICA working groups (see figure). Please contact the project management (n.hofman@amsterdamumc.nl) if you are interested to participate on behalf of the ERN GUARD-Heart. Soon all information will be available on: www.erica-rd.eu ♥

ERN Research – ERICA Project



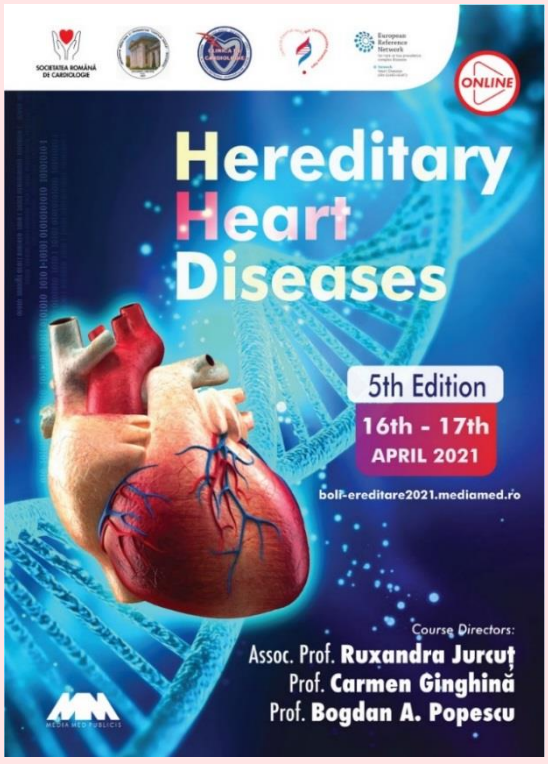
ERICA
European Rare Disease Research Coordination and Support Action

Latest ERN GUARD-Heart Publications

1. Sun B, Yao J, Ni M, Wei J, Zhong X, Guo W, Zhang L, Wang R, Belke D, Chen YX, Lieve KVV, Broendberg AK, Roston TM, Blankoff I, Kammeraad JA, von Alvensleben JC, Lazarte J, Vallmitjana A, Bohne LJ, Rose RA, Benitez R, Hove-Madsen L, Napolitano C, Hegele RA, Fill M, Sanatani S, Wilde AAM, Roberts JD, Priori SG, Jensen HK, Chen SRW. Cardiac ryanodine receptor calcium release deficiency syndrome. Sci Transl Med. 2021 Feb 3;13(579):eaba7287. doi: 10.1126/scitranslmed.aba7287. PMID: 33536282.

2. Ghidoni A, Elliott PM, Syrris P, Calkins H, James CA, Judge DP, Murray B, Barc J, Probst V, Schott JJ, Song JP, Hauer RNW, Hoorntje ET, van Tintelen JP, Schulze-Bahr E, Hamilton RM, Mittal K, Semsarian C, Behr ER, Ackerman MJ, Basso C, Parati G, Gentilini D, Kotta MC, Mayosi BM, Schwartz PJ, Crotti L. Cadherin 2-Related Arrhythmogenic Cardiomyopathy: Prevalence and Clinical Features. Circ Genom Precis Med. 2021 Feb 10. doi: 10.1161/CIRCGEN.120.003097. Online ahead of print. PMID: 33566628.

3. Asatryan B, Yee L, Ben-Haim Y, Dobner S, Servatius H, Roten L, Tanner H, Crotti L, Skinner JR, Remme CA, Chevalier P, Medeiros-Domingo A, Behr ER, Reichlin T, Odening KE, Krahn AD. Sex-Related Differences in Cardiac Channelopathies: Implications for Clinical Practice. Circulation. 2021 Feb 16;143(7):739-752. doi: 10.1161/CIRCULATIONAHA.120.048250. Epub 2021 Feb 15. PMID: 33587657. ♥



Lay journal publications

The objective 'external communication' of the specific grant agreement for the ERN GUARD-Heart includes the outcome indicator 'publication in scientific and non-scientific journals'. Currently, in our ERN the number of publications in lay (non-scientific) journals is too low. We would ask all our HCPs to publish this year at least one article in a lay (non-scientific) journal about the ERN (and the HCP). Examples of a lay journal can be a (hospital) magazine, a newspaper, a patient magazine, etc. If there are any local publications published already, please send a copy to project management office (n.hofman@amsterdamumc.nl). See for example the following link to a publication from Murcia (Juan Gimeno) in a lay journal: https://www.laverdad.es/ababol/ciencia/avances-genetica-cambian-20190504003322-ntvo.html ♥

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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