

Friday 31 May 2019

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 2019 NUMBER 3

Long QT Syndrome & Sports

Amsterdam, 31 May 2019

In this project of the ERN GUARD-Heart, healthcare professionals are invited to seek advice for their patients with long QT syndrome (LQTS) with regard to participation in sports. Healthcare professionals can do this by entering their patients with LQTS diagnosis who (wish to) participate in sports into the Clinical Patient Management System (CPMS)* and invite members of an advisory panel to provide recommendations.

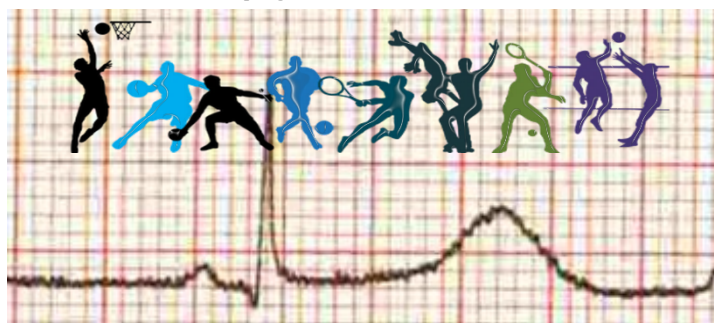
Current members of ERN GUARD-Heart advisory panel for this project are: Elijah Behr, Lia Crotti, Antoine Leenhardt, Anita Hiippala, Pier Lambiase, Jacob Tfelt-Hansen, and Arthur Wilde.

For each patient entered into the CPMS, the referring physician should upload at least a 12-lead ECG at baseline (and preferably during an exercise test). In addition, the referring physician is requested to address the following questions:

- Has genetic testing been performed in the patient? If yes, which genes have been tested and what was the result of genetic testing (please include affected gene, variant and assignment).
- Has the patient experienced any symptoms? If yes, please specify (e.g., cardiac arrest, syncope, documented arrhythmia, palpitations)?
- Does the family history include (aborted) sudden death or cardiac arrest at age <45 years? If yes, did this occur during sports activity?

- Does the patient use (non-cardiac) drugs? In case of a beta-blocker, please provide the name of the beta-blocker and the dosage (mg/kg)?
- Which sports does the patient primarily perform?
- Does the patient perform any secondary sports (>3 hours per week)? If yes, which?
- How many hours per week does the patient train & compete?
- How many years has the patient been training at a comparable level?
- The patient trains at the following level:
 - Recreational
 - Recreational with competitions
 - Competitive national
 - Competitive international
- On which treatment is the patient (e.g., beta-blocker, ICD, left stellate ganglion block)?
- What is the maximal heart rate of patient during a maximal exercise test (on treatment)?
- What is the maximal capacity of patients during a maximal exercise test? (answer numbers and unit, i.e. Watts, km/h, VO2max, and METS).

Continued on next page.





ERN News

Participation of patients with long QT syndrome in sports

There are no criteria with regard to age, sex, and/or ethnicity for patient inclusion.

The recommendations of members of the advisory panel should at least address the next question:

- should the patient with the LQTS diagnosis limit or restrict his/her participation in sports? If yes,
 - If yes: to what limit should participation in sports be restricted (answer in minutes or hours per week)?
 - which type of sports is the patient yet allowed to perform (check white box for appropriate sports category in the table)?

All cases entered into the CPMS for this project will be collected in a database for future research. Patient enrolment will start on 1 June 2019. ❤

Increasing Static Component ↑	III. High (>50% MVC)	Bobsledding/Luge*†, Field events (throwing), Gymnastics*†, Martial arts*, Sailing, Sport climbing, Water skiing*†, Weight lifting*†, Windsurfing*†	Body building*†, Downhill skiing*†, Skateboarding*†, Snowboarding*†, Wrestling*	Boxing*, Canoeing/Kayaking, Cycling*†, Decathlon, Rowing, Speed-skating*†, Triathlon*†
	II. Moderate (20-50% MVC)	Archery, Auto racing*†, Diving*†, Equestrian*†, Motorcycling*†	American football*, Field events (jumping), Figure skating*, Rodeoing*†, Rugby*, Running (sprint), Surfing*†, Synchronized swimming†	Basketball*, Ice hockey*, Cross-country skiing (skating technique), Lacrosse*, Running (middle distance), Swimming, Team handball
	I. Low ($<20\%$ MVC)	Billiards, Bowling, Cricket, Curling, Golf, Riflery	Baseball/Softball*, Fencing, Table tennis, Volleyball	Badminton, Cross-country skiing (classic technique), Field hockey*, Orienteering, Race walking, Racquetball/Squash, Running (long distance), Soccer*, Tennis
		A. Low ($<40\%$ Max O ₂)	B. Moderate (40-70% Max O ₂)	C. High ($>70\%$ Max O ₂)
Increasing Dynamic Component →				

* For instructions how to gain access to CPMS and enter a patient into the CPMS, healthcare professionals can contact the ERN GUARD-Heart CPMS helpdesk (cpmshelpdesk@guardheart.ern-net.eu; phone +31645153319) or management office (<http://guardheart.ern-net.eu/about/contact/>). A similar project will be soon announced in this Newsletter for patients with presymptomatic arrhythmogenic cardiomyopathy and their participation in sports.

Solve**RD**
Solving the Unsolved Rare Diseases

“Solve-RD - Solving the unsolved Rare Diseases” is a large European research project funded by the EU for five years (2018-2022) under Horizon 2020. The consortium is collaborating with ERNs to improve the diagnosis of rare diseases. Solve-RD is establishing an European Rare Disease Models & Mechanisms Network (RDMM-Europe) using the successful Canadian blueprint as a role model. The goal of RDMM-Europe is to link clinicians discovering new genes in patients with rare diseases with basic scientists, who can functionally analyse equivalent genes and pathways in model organisms. Solve-RD will provide 50 Seeding Grants (€ 20.000 each) to fund projects that will allow rapid confirmation of potentially disease-causing genes and elucidation of the underlying molecular disease mechanisms. European model organism investigators are encouraged to register in the [RDMM-Europe registry](#) and enter the genes/pathways they are working on for the respective model organism(s). Please contact the model organism investigators you are collaborating with to register! Further information can be found [here](#).



ERN News



UK withdrawal from the EU extended until 31 October

Brussels, 10 April 2019

The European Council decided on Wednesday 10 April 2019, in agreement with the United Kingdom (UK), to further extend the Article 50 deadline until 31 October 2019 to allow for the ratification of the Withdrawal Agreement by both Parties. The European Council will review the Brexit progress in June.

Therefore, the UK experts and representatives will participate as usual in all the meetings organised by the European Commission. UK Healthcare providers members of the ERN GUARD-Heart will participate in all the activities, boards, etcetera (including CPMS access and use). ♥



NEW CALL FOR PROPOSAL

Upcoming call for European centres to join existing ERNs

Brussels, 21 May 2019

The European Commission is currently preparing a new call for Centres of Excellence within the EU to join the existing ERNs. The call is expected to be launched later this year. This call will not be for proposals to establish new ERNs. It is yet not clear whether a call for new ERNs will be launched in the future.

Any updates regarding the future call for Centres of Excellence will be posted on [DG Sante ERN](#) and on the [website](#) of ERN GUARD-Heart. ♥

An European platform for rare disease registration

Brussels, 21 May 2019

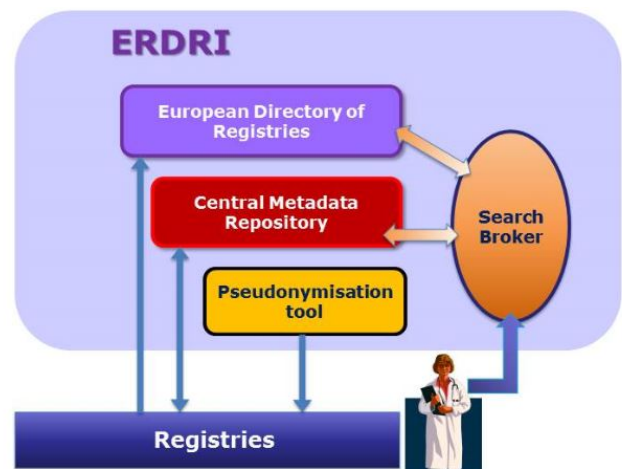
The European Platform on Rare Disease Registration (EU RD Platform), developed by the Commission's Joint Research Centre (JRC) in collaboration with DG SANTE, has been launched and is accessible online at the link: <https://eu-rd-platform.jrc.ec.europa.eu/>

The EU RD Platform will be essential in creating an overview of all existing rare disease registries and make them visible at European level. The Platform's tools will be available to all registries, free of charge. By using these tools, which are contained in the European Rare Disease Registry Infrastructure (ERDRI), the Platform will tackle the fragmentation of rare disease patients' data existing in hundreds of registries across Europe. It's important to note that the registries will remain the owners of their data.

The EU RD Platform's website contains instructions about how to use the following tools:

- the European Directory of Registries,
- the Central Metadata Repository, and,
- the Pseudonymisation tool which provides pseudonyms to the registries at local level.

The JRC will organise training courses for registries' staff and they can express interest in upcoming training sessions directly on the website of EU RD Platform. The possible use of the EU RD Platform by the ERN GUARD-Heart will be discussed during the next Board Meeting of the Network in September. ♥





ERN News

Call for expression of interest for members of Horizon Europe Mission Boards

Brussels, 17 May 2019

Following the partial political agreement reached in April 2019 between the European Parliament and the Council on Horizon Europe, the 2021 - 2027 EU Framework Programme for Research & Innovation, the European Commission is now preparing for the implementation of this programme, pending agreement on its budget as part of the general agreement on the Union's multi-annual financing framework and adoption of this programme.

One of the main novelties of 'Horizon Europe'2 is the development and implementation of ambitious missions to maximise the impact of EU support to research & innovation & demonstrate its relevance for society and citizens.

Five areas agreed for the implementation in the first years of the Horizon Europe implementation are:

- Cancer
- Adaptation to Climate Change
- Healthy Oceans, Seas, Coastal and Inland Waters,
- Climate-neutral and Smart Cities
- Soil Health and Food

On 13 May 2019 the Commission launched a [call for expression of interest](#) for members of five Mission Boards to help specify, design and implement missions for Horizon Europe. It will be open until 11 June 2019. The Commission is looking for a broad mix of experts from industry & innovation, research, policy making, practitioner organisations & from end users, which are appointed in a personal capacity and have a deep commitment to the success of the missions.

You can find more details about the call under link http://ec.europa.eu/transparency/regexpert/index.cfm?do=calls.calls_for_app

For any further information regarding this call please contact RTD-HORIZON-EUROPE-MISSIONBOARDS@ec.europa.eu. ♥

Education

Educational videos by ERN GUARD-Heart experts

Amsterdam, 31 May 2019

ERN GUARD-Heart is planning to produce educational videos for healthcare professionals who wish to gain more detailed knowledge on rare cardiac diseases & different aspects of the multidisciplinary management of these diseases (e.g., next-generation sequencing, sports, pregnancy). The videos will be in powerpoint format with voice over of ERN GUARD-Heart experts. The videos (with an ideal duration of 15 min.) will be published on the website and social media channels of the ERN (i.e., LinkedIn and Facebook). In the 1st slide, the speaker will be introduced, followed by the slides with educational content about the specific disease or topic. In collaboration with the thematic area leaders, the management office is currently creating a list of topics & candidate speakers. Next, candidate speakers will be asked whether they are willing to participate. The videos will be produced and disseminated by the management office. An exemplary educational video on diagnosis & risk stratification in catecholaminergic polymorphic ventricular tachycardia by Arthur Wilde is currently in production. ♥

Grants

Call for rare diseases registries

Brussels, 14 May 2019

The European Commission (EC) has published a call to support the development of rare disease registries by ERNs (see [link](#)). Five ERNs already received financial support from the EC and are developing a comprehensive approach for rare disease registries covering their entire ERN, following standards and tools developed by the EC's Joint Research Centre. The remaining 19 ERNs can now apply to this call. The budget is max € 6.000.000 per ERN with deadline for submission on 10 September 2019.

ERN GUARD-Heart management office is preparing a strategy on how to apply for this grant, and next will contact the thematic area leaders for discussions & approval. ♥



Cardiac Patient Trial & Registry Tool (Cardiac Patrol)

*now available in
app stores*

*sponsored by
ERN GUARD-Heart*

Cardiac Patrol

Cardiac Patient Trial and Registry Tool. The app contains relevant scientific studies and database information (registries) in the field of rare and congenital heart diseases.

- 1 LOGIN**
Create an account or login with your BIG-number
- 2 LIST OF CONDITIONS**
Click the disease your patient has been diagnosed with and enter their age
- 3 STUDY INFORMATION**
Select 'line of treatment' and see if there is an 'open trial'
- 4 LINE OF TREATMENT**
Review the study scheme or enter the in- and exclusion criteria and see which hospitals participate
- 5 REFERRALS**
Refer the patient to the researcher(s)



TO ADD A REGISTER OR STUDY TO THE APP, THE RESEARCHER PROVIDES THE FOLLOWING COMPONENTS

- Disease
- Study name
- Study status
- Study start date
- Study completion date
- Principal Investigator name
- PI email address
- PI hospital + city
- Eligible ages
- Line of treatment
- Picture of study arms
- Participating sites + local PI
- Inclusion criteria
- Exclusion criteria



Hart4Onderzoek is the first platform that brings together scientific research in the field of congenital heart and vascular diseases, both nationally and within the EU.
www.hart4onderzoek.nl



Latest ERN GUARD-Heart Publications

1. Michowitz Y, Milman A, Andorin A, Sarquella-Brugada G, Gonzalez Corcia MC, Gourraud JB, Conte G, Sacher F, Juang JJM, Kim SH, Leshem E, Mabo P, Postema PG, Hochstadt A, Wijeyeratne YD, Denjoy I, Giustetto C, Mizusawa Y, Huang Z, Jespersen CH, Maeda S, Takahashi Y, Kamakura T, Aiba T, Arbelo E, Mazzanti A, Allocca G, Brugada R, Casado-Arroyo R, Champagne J, Priori SG, Veltmann C, Delise P, Corrado D, Brugada J, Kusano KF, Hirao K, Calo L, Takagi M, Tfelt-Hansen J, Yan GX, Gaita F, Leenhardt A, Behr ER, Wilde AAM, Nam GB, Brugada P, Probst V, Belhassen B. Characterization and Management of Arrhythmic Events in Young Patients With Brugada Syndrome. *J Am Coll Cardiol*. 2019;73:1756-1765.
2. Towbin JA, McKenna WJ, Abrams DJ, Ackerman MJ, Calkins H, Darrieux FCC, Daubert JP7, de Chillou C, DePasquale EC, Desai MY, Estes NAM, 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, Peter van Tintelen J, Wilde AAM, Zareba W. 2019 HRS Expert Consensus Statement on Evaluation, Risk Stratification, and Management of Arrhythmogenic Cardiomyopathy. *Heart Rhythm*. 2019. doi: 10.1016/j.hrthm.2019.05.007.
3. Norrish G, Jager J, Field E, Quinn E, Fell H, Lord E, Cicerchia MN, Ochoa JP, Cervi E, Elliott PM, Kaski JP. Yield of Clinical Screening for Hypertrophic Cardiomyopathy in Child First-Degree Relatives: Evidence for a Change in Paradigm. *Circulation*. 2019. doi: 10.1161/CIRCULATIONAHA.118.038846.
4. O'Mahony C, Akhtar MM, Anastasiou Z, Guttmann OP, Vriesendorp PA, Michels M, Magri D, Autore C, Fernández A, Ochoa JP, Leong KMW, Varnava AM, Monserrat L, Anastasakis A, Garcia-Pavia P, Rapezzi C, Biagini E, Gimeno JR, Limongelli G, Omar RZ, Elliott PM. Effectiveness of the 2014 European Society of Cardiology guideline on sudden cardiac death in hypertrophic cardiomyopathy: a systematic review and meta-analysis. *Heart*. 2019;105:623-631.

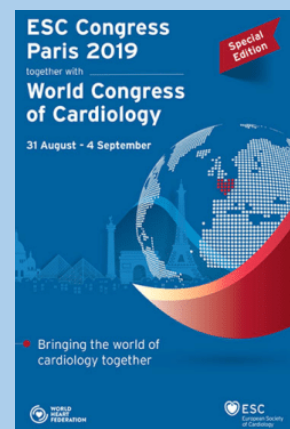


SAVE THE DATE

6th ERN GUARD-Heart Board Meeting

3 September 2019 (8.30 – 13.30)

*For (lunch) reservation send an email to
Nynke Hofman (n.hofman@amsterdamumc.nl)*



UPCOMING CARDIOLOGY EVENTS

EuroPrevent	11 April – 13 April 2019	Lisbon - Portugal
EuroCMR	02 May – 04 May 2019	Venice - Italy
Heart Rhythm Society Sessions	8 May – 11 May 2019	San Francisco – California
27th Nordic-Baltic Congress of Cardiology	10 June – 12 June 2019	Helsinki - Finland
Basic Science Summer School	16 June – 20 June 2019	Sophia-Antipolis - France
European Society of Cardiology	31 August - 4 September 2019	Paris – France
Update in Clinical Cardiology	16 October – 18 October	Boston - Massachusetts

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