Thursday 01 December 2022

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 2022 NUMBER 7

5-years Evaluation Process ERNs

Amsterdam, 28 November 2022

An important activity for the upcoming months is the 5 years evaluation process of the ERNs. This has officially started after the signature of the contract with the independent evaluation body on 9 November 2022. **All centers (HCPs) which were included from the beginning (March 2017) will be evaluated (21 HCPs in the ERN GUARD-Heart).**

The Independent Evaluation Body (IEB) is the consortium IDOM/ACSA. **On 12 December 2022 the self-assessment tool and guide to the evaluation platform will be made available.**

The evaluation calendar is demanding and for orientation you may wish to note that future steps will include:

- self-assessment of clinical units and of the ERNs (to be completed by 30 January 2023);
- on-site audits to a sample of clinical units (expected to take place from March to May 2023);
- ERNs and HCPs draft evaluation reports will be prepared by the IEB. ERNs and HCPs will be asked to provide comments, if any from April to June 2023.

→ To prepare the following documents are available on the website:

1. Evaluation manual
2. Tool-box
3. List of operational criteria.

https://guardheart.ern-net.eu/experts/5-years-evaluation-process/ ♥

AEPC Webinar Series

Leiden/ Amsterdam, 28 November 2022

In addition to the ERN webinars which started this summer; the Association for European Paediatric and Congenital Cardiology, in collaboration with ERN GUARD-Heart, is initiating a series of teaching webinars on paediatric cardiology and cardiac surgery topics, serving towards preparation for the AEPC exam. The sessions will cover topics summarised in the "AEPC Recommendations for Training in Paediatric and Congenital Cardiology" (CITY, 2020).

The webinars will take place biweekly on Wednesdays between 17:00-18.30 CET, starting on 7 December 2022 and will generally consist of a case presentation, followed by 2 lectures, and will allow sufficient time for Q&A and interaction with the speakers.

Topics & most speakers for the first 4 webinars are now confirmed and you are invited to sign up to attend these webinars!

Find out more & register here:
https://www.aepc.org/aepc-webinars

Registration for the webinars is open to members & non-members, and we encourage you to share this information with anyone who would be interested to join these free teaching sessions.

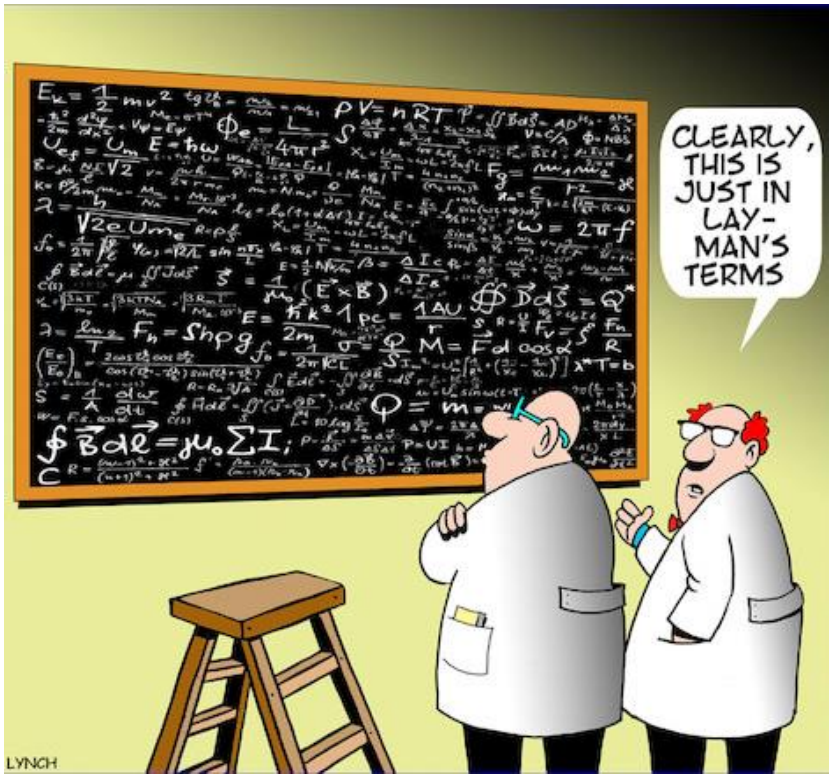
We look forward to welcoming you to our first session on 7 December 2022! ♥

Translating ERN publication abstracts into laymen language

Amsterdam, 28 November 2022

Within the ERN, patient engagement has a major focus. With that in mind we believe it is important to 'unlock' our research output for the lay public more systematically. We would therefore like to launch a dedicated patients page on our website where all of us commit to write an abstract on our mutual research papers in laymen language. Corresponding authors from 'official ERN papers'* will therefore be contacted by e-mail with the request to 'translate' the abstract into laymen language. This will be added to the website with a direct link to the full-publication. See below the first translation as an example. ♥

*a publication which includes member HCPs from at least two different EU Member States.



Document: European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases. *Europace (2022) 24, 1307–1367. Translated by Arthur Wilde.*

In the last three decades it has become increasingly clear that genetics play a very important role in a variety of diseases of the heart. Indeed, in the early nineties of the previous century the first genes underlying specific cardiac diseases were discovered and the years thereafter many more genes, often for the same conditions, were identified. In addition, it also became clear that for some conditions with more complex inheritance patterns are pertinent.

With the discovery of different genes underlying the same conditions it appeared that decisions regarding (future) therapy were influenced by the gene responsible for the disease (i.e. gene-specific treatment). Also, the future risk of developing the disease or specific symptoms (not rarely the risk of sudden death) appeared to be dependent on the underlying gene defect. This knowledge, in addition to the technical developments regarding genetic testing, has led to increasing availability of more comprehensive genetic testing. And after a genetic variant has been found in an affected person, so-called 'presymptomatic screening' of his or her family members has also become popular even if they are free of symptoms. By detecting carriers of the gene variant in the family, individuals at risk can be identified and followed up appropriately. And with the genetic substrate available within affected families, targeted screening, before family members develop symptoms, directed towards the identified variant causing the disease, has also become popular.

This consensus document reviews the state of genetic testing in 2022, and addresses the questions of what tests to perform and when to perform them. Hence, for every inherited heart condition, specific details are given on who to test (i.e. which criteria are needed before a patient should be tested for a specific diagnosis), which genes to screen and when (for example from which age onward). The document also includes a section on when and how to perform genetic testing after sudden cardiac death and, for the first time, a section on genetic testing in congenital heart disease. The document was written by international experts, including doctors and scientists, in the fields, and was endorsed by the major professional societies from around the world, spanning Europe, North and South America and Asia).

The main recommendations include an emphasis on appropriate genetic counselling prior to every genetic test, working in a specialized multidisciplinary team with involvement of geneticists and (pediatric) cardiologists, and on targeted, rather than random, genetic screening. It is also made very clear that in the case of potentially lethal and treatable conditions such as catecholaminergic polymorphic ventricular tachycardia (CPVT) or long QT syndrome (LQTS), it is the responsibility of the physician, in conjunction with an expert genetics team, to communicate to the patient/family the critical importance of family screening, encompassing both clinical testing (i.e. ECG, exercise-ECG, echo, etc.), and predictive genetic testing. The ultimate goal of this document is to help guide clinicians on the effective use of genetic testing to improve the diagnosis and care of patients, and their families, with inherited heart diseases.

Update 15-16-17 March 2023: ERN GUARD-Heart Board Meeting and EJP-RD Networking Symposium

Amsterdam, 28 November 2022

On Wednesday 15 March 2023, the ERN project management organizes a hybrid ERN GUARD-Heart Board meeting in Amsterdam (13:00 – 17:00) and dinner in the evening (19:00 – 22:00).

Practical information about the ERN Board meeting

Date: Wednesday 15 March 2023

Time: 13:00 – 17: 00

Location: Trippenhuis, Kloveniersburgwal 29, 1011 JV Amsterdam (city centre)

Registration: Registration for the Board meeting and dinner is completed now. If there are any changes or requests: please contact the project manager (n.hofman@amsterdamumc.nl). Please book your travel tickets by yourselves. For those who indicated this, one hotel night will be booked by the ERN office. For ERN members who come to Amsterdam there will be travel costs reimbursed up to 300,- EURO, and a one night stay in a hotel room will be covered as well (for one person per HCP, max. 175,- EURO). Online attendance of the meeting will be possible as well.

Trippenhuis
National monument in Amsterdam

Update 15-16-17 March 2023: ERN GUARD-Heart Board Meeting and EJPRD Networking Symposium

Amsterdam, 28 November 2022

Practical information about the Symposium

The symposium is organized by the Amsterdam UMC and ERN GUARD-Heart and is funded by the EJP RD Networking Support Scheme. It will include talks on cutting edge research from some of the world's leading investigators in this field. It will also include discussions on developing the new and existing networks required to further progress research in this field and translate recent advances into clinical applicability - the development of multicentre patient registries, national and international databases and future collaborative projects and grant applications. The detailed programme is available in the former ERN-newsletter (no.6) and behind the link below.

Date: Thursday 16 March 2023 and Friday 17 March 2023

Location: Trippenhuis, Kloveniersburgwal 29, 1011 JV Amsterdam (city centre)

How to register for the symposium?
Many ERN-members registered already for the symposium, and **the tickets for in-person attendance are sold out.** However, there is an option for online only attendance (for Day 1 only), follow the link for registration. https://www.eventbrite.com/e/ejprd-networking-symposium-complex-genetics-of-inherited-arrhythmias-tickets-399392793597

Invitation for Abstracts
Abstracts are invited for this symposium, especially from early career researchers. A number of submitted abstracts will be selected for talks during Session 6. There will also be poster viewing for all submitted abstracts. Please send abstracts to genetics_abstracts@amsterdamumc.nl (abstract deadline: 01/02/2023) **For abstract presenters there will be become extra (in-person attendance) tickets available.**

For speakers at the symposium: the travel tickets and a two night stay will be arranged. They have been contacted personally about this.

For more information, please contact the meeting organizers:
Connie Bezzina (c.r.bezzina@amsterdamumc.nl)
Nynke Hofman (n.hofman@amsterdamumc.nl)
Roddy Walsh (r.t.walsh@amsterdamumc.nl). ♥

ePAG's presentation in Zürich

Zurich, 22 September 2022

The 5th Zurich International Symposium on Arrhythmogenic Cardiomyopathies took place in Rüschlikon, Switzerland on September 22nd and 23rd. It was organized by Universitätsspital Zürich (USZ). Worldwide experts from Europe, USA, Canada and Australia participated in several sessions on etiopathogenesis, diagnosis, risk stratification, therapy and update on registries and research on ACM. Two research prizes for basic and clinical ACM science (15.000 Swiss Francs each) were granted by the Georg and Bertha Schwyzer-Winiker Foundation.

For the first time in the history of this symposium (the first edition was 10 years ago), a patient representative was invited to speak as well. Ruth Biller, Chair of ERN GUARD-Heart ePAG and member of the Cardiomyopathy Council in Global Heart Hub presented very passionately ACM from the patient's perspective and asked for:

- an official nomenclature;
- shorter times to diagnosis;
- task force criteria for left dominant types;
- genetic testing in recurrent myocarditis;
- clear recommendations to practice sports and taking contraceptives;
- special programs for rehab and psychological support;
- more participation from patients in congresses, studies, and guidelines.

Her presentation received lots of compliments from the participants. ♥

Start with Podcast Series

Amsterdam, 30 November 2022

In January 2023, ERN GUARD-Heart will start with a weekly podcast series in which all members will be requested to participate. The idea is that in each recording, the main representative of each member HCP and one of his/her colleagues (preferably an early career professional in the field of rare cardiac diseases) tell about their HCP and their experiences and challenges in the field of rare cardiac diseases. A standard template will be used for the questions, so that everyone can prepare well before the recording. Additional detailed information will follow soon by e-mail. ♥

ERN monitoring data

IMPORTANT NOTICE

Amsterdam, 28 November 2022

As explained during the last Board meetings, all the ERNs have a common monitoring process. As ERN, we have to collect and submit our activities each year to the European Commission.

Soon you (ALL ERN members) will be invited to deliver your HCP activities. Early January 2023, you will receive a template in which you can fill in your activities. Please start to list all the organized courses/ webinars, presentations at conferences etc. during the year, this will help you to complete the document easily.

The included indicators are available in a manual (which was sent by e-mail in April 2022). The deadline for sending your information will be mid-February 2023.

Our experience is, that the most difficult indicator is the one which asks for the number of NEW patients referred to the HCPs participating in the ERN with the diagnosis of a disease/condition that fall within the scope of the ERN.

* we will ask for the numbers of NEW patients per disease.

* this is really about NEW patients (who were not in the hospital system before), seen in 2022.

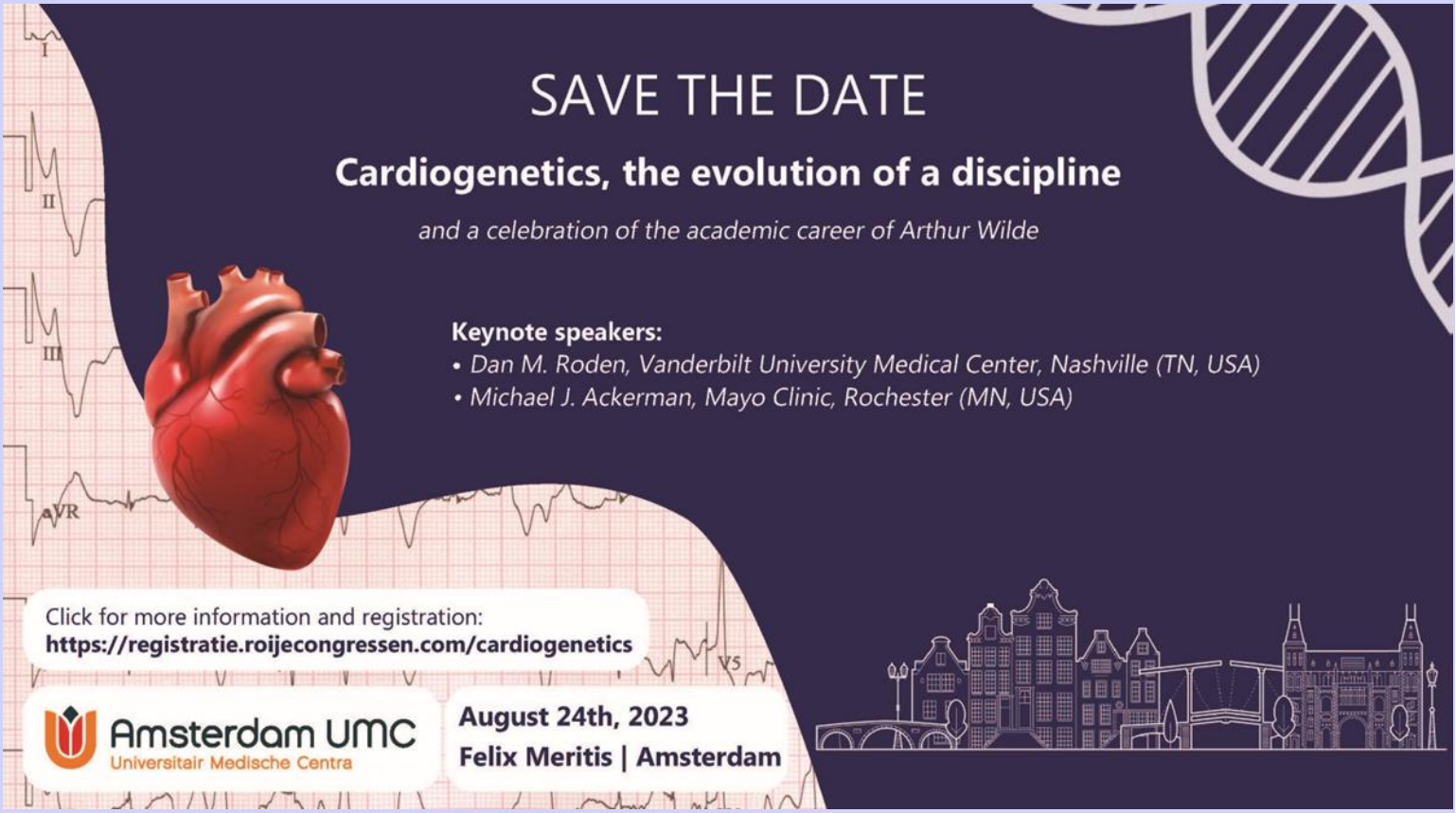
- it's about patients having a certified diagnosis of rare disease.

We would advice all (affiliated) members to think about the way to retrieve this information from your hospital systems, as this strongly depends of your local IT-services.

Only when we measure and count at the same way, we will have reliable numbers. . ♥

Latest ERN GUARD-Heart Publication(s)

1. **Syncope in hypertrophic cardiomyopathy (part II): An expert consensus statement on the diagnosis and management.** Brignole M, Cecchi F, Anastasakis A, Crotti L, Deharo JC, Elliott PM, Fedorowski A, Kaski JP, Limongelli G, Maron MS, Olivotto I, Ommen SR, Parati G, Shen W, Ungar A, Wilde A. Int J Cardiol. 2022 Oct 26:S0167-5273(22)01664-3. doi: 10.1016/j.ijcard.2022.10.153. Online ahead of print. PMID: 36309161.



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