Monday 1 February 2021

ERN GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart

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

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ERN GUARD-HEART BIMONTHLY NEWSLETTER YEAR 2021 NUMBER 1

Vaccination against SARS-CoV-2 in specific groups of patients with rare cardiac diseases

Amsterdam, 29 January 2021

The ERN coordinators recently discussed within their ERNs which patients should have priority in the vaccination program against the corona virus, and in which group of patients vaccination may be contraindicated. The lists of patient groups that were provided by the ERNs were presented on the coordinators meeting (27 January 2021). The final list will be sent to the Board of Member States.
This issue has also been discussed internally in the ERN GUARD-Heart, following an email by the project management to all representative members of member HCPs. Based on the comments received and the discussions made, the ERN GUARD-Heart recommends a priority for vaccination against SARS-CoV-2 for the following patient groups:

- Patients with the Brugada syndrome (including patients diagnosed by a positive drug challenge test without additional diagnostic criteria).
- Patients who carry a (probable) pathogenic variant in the cardiac sodium channel gene *SCN5A*.
- Patients with primary arrhythmia syndrome and a history of symptomatic arrhythmias.
- Patient with cardiomyopathy (or congenital heart disease) and risk factors (reduced left ventricular systolic function, heart failure N.Y.H.A. III/IV or pulmonary hypertension).

The board of the ERN GUARD-Heart recommends patients with Brugada syndrome and a spontaneous type 1 ECG and patients with a (probable) pathogenic variant in *SCN5A* to take paracetamol 1 hour before administration of the vaccine (1000 mg in adults; adjusted dosage according to prescription in children) and continue using paracetamol in a total of 3 days after vaccination (1000 mg 3 times a day in adults; adjusted dosage according to prescription in children).

The board of the ERN GUARD-Heart recommends patients with primary arrhythmia syndrome and a history of arrhythmias and patient with cardiomyopathy and risk factors (as described above) to measure their temperature regularly and take paracetamol in case of fever (>38.5˚C).

If, despite the measures described above, fever (>38.5˚C) occurs, patients are recommended to contact and/or follow the recommendations of their treating physician.

Finally, it must be noted that, due to lack of data in extremely rare inheritable cardiac diseases, it is yet unknown whether unanticipated and undesired reactions may occur due to vaccination (for example an abnormal immune response in patients with the Timothy syndrome). However, although additional caution is required, the board of the ERN GUARD-Heart advises that these groups should not be routinely withheld from vaccination against the SARS-CoV-2. This official recommendations of the ERN GUARD-Heart are available on the website of the ERN: https://guardheart.ern-net.eu/covid-19/ ♥

IX JORNADA DE CARDIOGENÉTICA

Dña. Ester Costafreda
Presidenta SAMS

Juan Ramón Gimeno
CSUR Cardiopatias Familiares
HUVA

Activities of ERN members: online patient meetings

Amsterdam, 26 January 2021

The Spanish colleagues in the ERN have been active in organising (patient) meetings. On 27 November 2020, Juan Gimeno from Murcia organized the *IX Jornada de Cardiogenetica*. The program included 5 online sessions on topics such as COVID-19 and inherited cardiac disease, new therapies (mavacamten for hypertrophic cardiomyopathy), sports, pre-implantation diagnosis and pregnancy. In total, 57 invited speakers completed the 11 hours program, which was recorded and is available to date for more than 860 people registered. There were 209 patients/relatives, who participated both as speakers and attendants, 42 researchers, 39 pharma-technological company delegates, on top of a majority of medical staff from different disciplines. ERN GUARD-Heart patient advocates Ruth Biller and Ester Costafreda participated and joined as well to represent the voice of the patients. https://jornadacardiogenetica.com/#home ♥

Georgia Sarquella-Brugada and Ester Costafreda from Barcelona have organized the 1st online edition of the *Jornada SAMS* on 16 January 2021. The day was a great success with >300 inscriptions and >190 people connected for the entire duration of the event (which was 5 and half hours; almost two hours more than planned because of the big success of the Q&A through the chat). The talks were divided into two blocks: one for channelopathies and one for cardiomyopathies. Cardiologists, geneticists, gynaecologists, a heart transplant surgeon and sexologists presented on genetics, long QT syndrome, Brugada syndrome, cardiac devices, pregnancy and heart transplantation. Experts from 3 well-known hospitals (Clinic, Sant Joan de Deu and Bellvitge) were included.♥

Recently, two online meetings have been organized for patients at the Amsterdam University Medical Centers (UMC). The first meeting was held on 16 December 2020 for patients with an increased risk for idiopathic ventricular fibrillation due to a Dutch founder haplotype on chromosome 7 (comprising *DPP6* gene). The second meeting was held on 21 January 2021 to provide information on the different vaccines against the corona virus and possible precautions to be taken in patients with Brugada syndrome and long QT syndrome (350 participants). ♥

The Romanian Society of Cardiology, the Expert Center of Rare Cardiovascular Genetic Diseases and the University of Medicine and Pharmacy of Bucharest will organize an online course with live transmissions on April 16-17th 2021, aclled the *Cardiac Hereditary Diseases.* Course directors are Ruxandra Jurcuţ, Carmen Ginghină, and Bogdan A.Popescu from the Emergency Institute for Cardiovascular Diseases "Prof. dr. C.C. Iliescu" (Bucharest, Romania). The full program of the course will be soon published on the website of the ERN (https://guardheart.ern-net.eu/ern-events/future-events/). *See poster of the event on page 4.* ♥

European Reference Networks

EUROPEAN JOINT PROGRAMME RARE DISEASES

RESEARCH TRAINING WORKSHOP

CALL OPEN: 25. JANUARY-7. MARCH 2021

• Submit workshop topics to receive funding for the organization of 2-days workshop

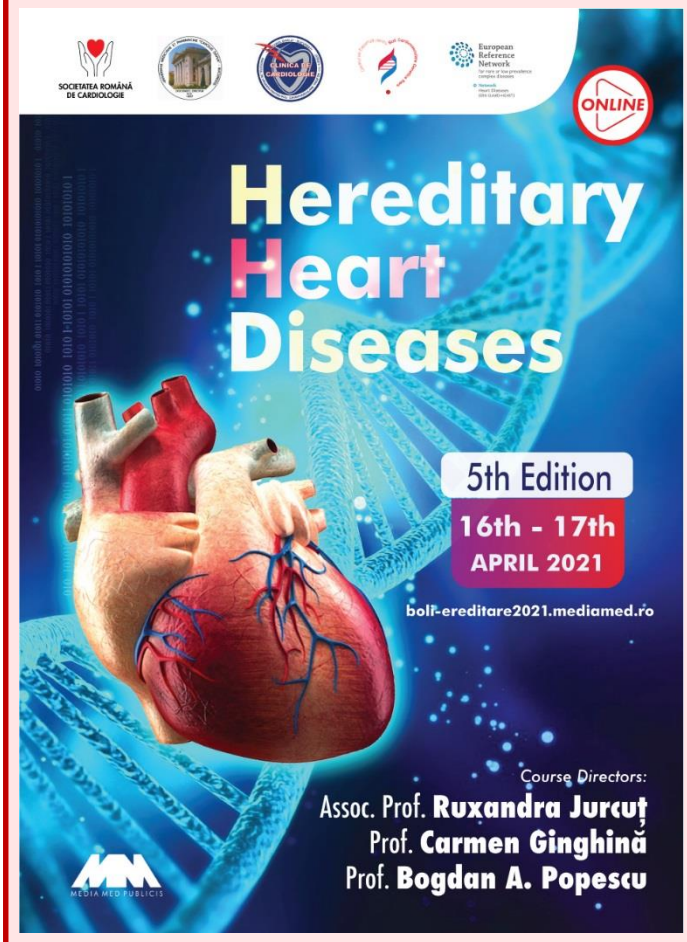
• Training topics may include among others: diagnostic research topics, interdisciplinary treatment approaches (e.g. gene therapy,transplantation)

• Budget: 25.000€/ workshop (e.g. venue, catering & travel and accommodation of speakers & participants)

• Applicant's profile:

- Affiliated to an EJP RD beneficiary institution or
- Affiliated to ERN-Full Member or ERN-Affiliated Partner

Further information at:
https://www.ejprarediseases.org/index.php/training-and-empowerment/ern-trainings/



Procedure and timeline for internal applications for new thematic areas

Amsterdam, 29 January 2021

The deadline for submitting internal applications for the thematic areas 'congenital heart disease' and 'other rare cardiac diseases' has passed. For both these new thematic areas we have received 10 applications. The applications will be assessed by the same expert team which has recently assessed the external applications (for new member to join the ERN GUARD-Heart), i.e.: Carlo Napolitano, Eric Schülze-Bahr, Philippe Charron, Georgia Sarquella-Brugada, and Arthur Wilde. The assessors will not be involved in the review and assessment of the application of their own centre. If needed, additional information may be requested before the 1 March 2021, and hopefully thereafter the results of the assessment will be shared within the ERN. ♥

Update PROMS-workshops

Amsterdam, 26 January 2021

The Heart Rhythm Management Centre (HRMC) and the Centre for Medical Genetics (CMG) of the University Hospital of Brussels (UZ Brussel), in collaboration with the ERN GUARD-Heart, has organized four online workshops between October 2020 and January 2021 in a joined attempt to assess the specific psychosocial issues encountered by individuals undergoing (genetic) counselling for inherited cardiovascular disease. These workshops were led by Saar van Pottelberghe, psychologist and PhD student in Brussels, with help of experts in this field and patient advocates Ruth Biller and Edward Callus. Each workshop was attended by around 20-25 participants (mainly cardiologists, psychosocial workers, genetic counsellors, clinical geneticists, and patients). With the workshops, opinions of different stakeholders in cardio-genetic health care were gathered. With the Delphi consensus technique, the project team wants to distil further relevant facilitators and barriers to implementing patient-centred care in genetic cardiology. Now they will start writing a paper on PROMs in cardio genetics based on the gathered data from the survey and literature and hopefully this will lead to the implementation of a PROMS tool in our field. ♥

The next ERN GUARD-Heart board meeting will be held online on Tuesday 9 March 2021 from 13.00 – 16.00 h.

ERN Research

Restart of the project on long QT syndrome and sports

Amsterdam, 29 January 2021

In 2019, the ERN GUARD-Heart launched a project in which medical doctors were invited to seek advice for their patients with long QT syndrome (LQTS) with regard to participation in sports. Physicians could 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. The advisory panel consisted of a group of LQTS experts from the ERN GUARD-Heart. Unfortunately, due to technical issues and the lack of time associated with the use of CPMS, only a few patient were included in the project.

ERN GUARD-Heart aims to re-establish this project. The panel of experts are still prepared to participate in the advisory panel and provide their advice. However, this time, a dedicated PhD student, Juliette van Hattum (MD and former professional hockey player) will help to upload cases and the required information.

For each patient entered into the CPMS, Juliette together with the referring physician will upload at a 12-lead ECG at baseline (and preferably also during an exercise test). In addition, the following data will be collected:

- 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 symptoms? If yes, please specify (e.g., cardiac arrest, syncope, documented arrhythmia, palpitations)?
- Does 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 any (non-cardiac) drugs?
- Which sports does the patient primarily perform?
- Does the patient perform any secondary sports (>3 hours per week)? If yes, which?

Juliette van Hattum

PhD student involved in the LQTS and sports project.

- 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).*

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 re-start on 1 February 2021. ♥

Amsterdam, 26 January 2021

Following the official Brexit date of 31 December 2020, the three UK centres, all from London, within ERN GUARD-Heart had to leave our ERN officially. We are grateful for the very active participation of Elijah Behr (St Georges University hospitals), Perry Elliott (Barts Heart Centre, St Bartholomew's Hospital) and Juan Kaski (Great Ormond Street Hospital). Although the UK and mainland Europe formally split we are sure that, in the interest of patients with rare cardiac diseases, we will continue collaborating on a very active basis. Besides, good friendship will not be influenced by what politicians in Brussels and Downing street decide. ♥

Monitoring numbers of 2020

Amsterdam, 29 January 2021

For the ERN monitoring system, we have to collect the activities of ERN members for 2020 (complete year). Although the online system for data collection is not open yet, we expect that we have to submit these numbers in March 2021. As soon as we have received the final instructions, we will send an e-mail with the requested indicators. This time the affiliated partners will be requested as well (for the first time) to report their activities and number of (new) patients. ♥

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Funded by European Union

European Reference Network for rare or low prevalen complex diseases

Network Heart Diseases (ERN GUARD-HEART)

EHRA

EHRA Survey on SUDY

London, 28 January 2021

Sudden death in the young (age 1-40) is a rare occurrence affecting around 2-3 in every 100,000 young people every year in Europe. Whilst this amounts to several thousand deaths per annum, the impact is more dramatic than when older people die suddenly. There is also a high likelihood of underlying genetic heart disease as the cause of death, and therefore genetic risk to other family members that requires identification in order to prevent further mortality. Historical studies have indicated, however, that there is an extreme heterogeneity in provision for investigation of genetic heart disease in decedents and their families across the continent despite several position statements and guidelines.

The aims of this survey are therefore as follows:

1. Gain an understanding of the provision across Europe of the following:
- services for families of decedents with sudden death in the young
- the current heterogeneity of autopsy practice and post-mortem genetic studies
- the availability of family service referrals and psychological support

2. The variation of family investigation protocols and the role for provocation studies.

Take the survey here:
https://www.surveymonkey.com/r/SUDY

Thank you for your consideration.

Elijah Behr
on behalf of the ECGen focus group of EHRA ♥

Latest ERN GUARD-Heart Publications

1. Behr ER, Ben-Haim Y, Ackerman MJ, Krahn AD, Wilde AAM. Brugada syndrome and reduced right ventricular outflow tract conduction reserve: a final common pathway? Eur Heart J. 2021 Jan 9:ehaa1051. doi: 10.1093/eurheartj/ehaa1051.

2. Tadros R, Francis C, Xu X, Vermeer AMC, Harper AR, Huurman R, Kelu Bisabu K, Walsh R, Hoorntje ET, Te Rijdt WP, Buchan RJ, van Velzen HG, van Slegtenhorst MA, Vermeulen JM, Offerhaus JA, Bai W, de Marvao A, Lahrouchi N, Beekman L, Karper JC, Veldink JH, Kayvanpour E, Pantazis A, Baksi AJ, Whiffin N, Mazzarotto F, Sloane G, Suzuki H, Schneider-Luftman D, Elliott P, Richard P, Ader F, Villard E, Lichtner P, Meitinger T, Tanck MWT, van Tintelen JP, Thain A, McCarty D, Hegele RA, Roberts JD, Amyot J, Dubé MP, Cadrin-Tourigny J, Giraldeau G, L'Allier PL, Garceau P, Tardif JC, Boekholdt SM, Lumbers RT, Asselbergs FW, Barton PJR, Cook SA, Prasad SK, O'Regan DP, van der Velden J, Verweij KJH, Talajic M, Lettre G, Pinto YM, Meder B, Charron P, de Boer RA, Christiaans I, Michels M, Wilde AAM, Watkins H, Matthews PM, Ware JS, Bezzina CR. Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. Nat Genet. 2021 Jan 25. doi: 10.1038/s41588-020-00762-2. Epub ahead of print. PMID: 33495596.

3. Mizia-Stec K, Charron P, Gimeno Blanes JR, Elliott P, Kaski JP, Maggioni AP, Tavazzi L, Tendera M, Felix SB, Dominguez F, Ojrzynska N, Losi MA, Limongelli G, Barriales-Villa R, Seferovic PM, Biagini E, Wybraniec M, Laroche C, Caforio ALP; EORP Cardiomyopathy Registry Investigators. Current use of cardiac magnetic resonance in tertiary referral centres for the diagnosis of cardiomyopathy: the ESC EORP Cardiomyopathy/Myocarditis Registry. Eur Heart J Cardiovasc Imaging. 2021 Jan 8:jeaa329. doi: 10.1093/ehjci/jeaa329. Epub ahead of print. PMID: 33417664.

All ERN members are invited to share news, activities and events from their centre with other members of the ERN GUARD-Heart and affiliated partners through this Newsletter.

Deadline for the next edition is 20 March 2021.

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