

Thursday, 03 October 2019

ERN



GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart



ERN GUARD-HEART BIMONTHLY NEWSLETTER

YEAR 2019 NUMBER 5

2019 call for membership to the existing ERNs is open

Brussels, 30 September 2019

The first call for new centres to join the 24 existing ERNs opened on the 30th of September and will run until the 30th of November 2019. For new centres wishing to join ERN GUARD-Heart, make sure to prepare well by reading the information on how to apply using the link:

https://ec.europa.eu/health/ern/consultations/2019_call_membership.en ♥

Cardiac Pathology Fellowship for Tereziá Tavačová from Prague

London, 17 September 2019

Tereziá Tavačová, resident in Pediatric Cardiology at Children's Heart Center and PhD Student in field of molecular and cell biology and genetics at Charles University in Prague, received the Cardiac Pathology Fellowship. This fellowship provides the opportunity to gain knowledge and expertise in the pathology of rare and complex heart diseases and in particular in the examination of the heart in young cases with sudden unexpected death. The fellowship will be hosted by the specialized CRY cardiovascular pathology unit in the St George's Medical School in London, United Kingdom, and will be coordinated by Professor Mary Sheppard (cardiac pathologist) and Professor Elijah Behr (cardiologist and representative member of St George's Healthcare NHS Trust in the ERN GUARD-Heart. ♥

LQTS – advice on sports activity – upload your cases

Amsterdam, 30 September 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. For each patient the consulting physician has to upload a 12-lead ECG at baseline and to fill in the digital Case Report Form (CRF) and upload it into CPMS. The CRF is available for download on our ERN website (see link). Then the advisory panel can provide their recommendations. Consulting specialists can use the general CPMS consent form for this. ♥

Thematic area's 'congenital heart disease' and 'other rare cardiac diseases' open for applicants

Amsterdam, 30 September 2019

In ERN GUARD-Heart, besides the thematic areas Familial Electrical Diseases in adults and children, Familial Cardiomyopathies in adults and children, and Paediatric Special Electrophysiology Conditions the thematic areas Congenital heart disease in adults (GUCH) and children and 'Other Rare Cardiac diseases' will be open for applicants as well. Existing members received a request to inform the project management whether they fulfill the criteria for inclusion in the thematic area Congenital Heart disease and if they are interested to be included in it. ♥



ERN monitoring: data collection

Amsterdam, 17 September 2019

The monitoring process of the ERNs continues with an updated document, which has been sent to all members on the 17th of September. The indicators and the number of indicators have not changed, but the definitions are clarified and with a more detailed description. The 18 indicators are listed below (2 of them have to be developed), subdivided into 8 objectives:

Objective 1: To ensure that ERNs are operational

1.1 Within an ERN, the number & percentage of Member States with full Health Care Providers as members.

1.2 Number of Health Care Providers represented in the ERN.

1.3 Number of affiliated partners represented in the ERN.

1.4 Number of patient organisations represented in the ERN.

Objective 2: To improve access to clinical advice, diagnosis, treatment and follow-up of patients within the ERNs.

2.1 Total number of new patients referred to the Health Care Providers participating in the ERN with the diagnosis of a disease/ condition that fall within the scope of the ERN.

2.2 Number of patients entered into CPMS.

Objective 3: To optimize patient outcomes by combining healthcare professionals' skills & resources used.

3.1 Number of patients entered into CPMS and reviewed by the ERN (a panel case review which leads to an outcome report).

3.2 Time taken to provide multidisciplinary clinical advice – non urgent cases: days (median) between referral to ERN and multidisciplinary clinical advice.

Objective 4: To increase capacity of professionals to recognize and manage cases of rare and complex conditions and disease within the scope of the ERN.

4.1 Number of educational webinars/videos aimed at healthcare professionals delivered by the coordination or HCPs members of the ERN.

4.2 Number of formal educational activities(i.e. those accruing higher educational credits) aimed at healthcare professionals organized by the ERN.

Objective 5: To reinforce clinical research in the field of rare and complex conditions and disease by collecting data and carrying out research activities.

5.1 Number of Clinical Trials or Observational prospective studies (with > 1 Member State and Health Care Provider within the ERN).

5.2 Number of accepted peer-reviewed publications in scientific journals regarding disease-groups within the ERN and which acknowledge the ERN.

Objective 6: To ensure that patients referred to ERNs have equal access to high and quality healthcare services.

6.1 Cumulative number of Clinical Practice Guidelines and other types of clinical Decision Making Tools and the number of new ones, adopted for disease within the scope of the ERN.

6.2a Cumulative number of Clinical Practice Guidelines and the number of the new ones **written** by the ERN.

6.2b Cumulative number of other types of Clinical Decision Making Tools (clinical consensus statements or consensus recommendations) and the number of new ones, written by the ERN for disease within the scope of the ERN.

Objective 7: To guarantee that knowledge is spread outside the ERN so that more people can benefit from the ERN activities.

7.1 Number of congresses/conferences/ meetings at which the ERN activities and results were presented.

7.2 Number of individual ERN website hits.

Objective 8: complex and long-term indicators which need further development

8.1 Level of patient satisfaction

8.2 Health Care Provider Compliance to Clinical Guidelines. ♥



ERN monitoring: how to proceed

There were specifically a lot of questions about the interpretation of indicator 2.1:

Total numbers 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.

->this is really about NEW patients (who were not in the hospital system before)

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

→ So; Persons who come for genetic testing, and have a genotype negative testresult should NOT be included. The genotype positives should be counted (also when they are phenotype negative). The EU-platform for data-entry will close at the end of October. The members received an action request (with digital PDF and Excel file) for sending their (revised) data of 2018, and the data of the first semester of 2019, before the 20th of October to the project management (n.hofman@amsterdamumc.nl). Please fill in both: the digital PDF and the Excel file.♥

HCP Name: _____

Country: _____

Name representative: _____

Date: _____

Continuous monitoring 2018 - REVIEW

2.1 Total number of new patients referred to the Health Care Provider, with the diagnosis of a disease/condition that fall within the scope of the ERN (please fill in Excel file)

4.1 Number of educational webinars/videos aimed at healthcare professionals delivered by the coordination or HCP members of the ERN

4.2 Number of formal educational activities

7.1 Number of congresses/ conferences/ meetings at which the ERN activities and results were presented

Continuous Monitoring – Requested data 2019 (first six months)

2.1 Total number of new patients referred to the Health Care Provider, with the diagnosis of a disease/condition that fall within the scope of the ERN (please fill in Excel file)

4.1 Number of educational webinars/videos aimed at healthcare professionals delivered by the coordination or HCP members of the ERN

4.2 Number of formal educational activities

7.1 Number of congresses/ conferences/ meetings at which the ERN activities and results were presented

Educational videos by ERN GUARD-Heart experts

Amsterdam, 23 September 2019

ERN GUARD-Heart has started to produce online educational videos for healthcare professionals who wish to gain more detailed knowledge on rare cardiac diseases & different aspects of multidisciplinary management of these diseases, e.g., next-generation sequencing, sports, pregnancy. The first two educational videos are available on the official ERN website (see [link](#)). During the boardmeeting in Paris the list of topics and speakers was completed. Some presenters (those with a deadline of 1 November) received last week instructions on how to record and send the Powerpoint presentation with voice-over. Others will be contacted soon as well. ♥





6th ERN GUARD-Heart Board Meeting



Paris, 02 and 03 September 2019



**Meeting with
Representatives of
Associated National
Centres from Austria,
Estonia, Lithuania,
Slovakia, and Slovenia.**



**Board-
meeting
of ERN
GUARD-
Heart in
Paris**

03.09.2019



7th Board Meeting of ERN GUARD-Heart



The 7th board meeting of the ERN GUARD-Heart will be held in **Rome, Italy, on Friday 14 February 2020**. On the evening before, Thursday 13 February a dinner will be organized for the participants. Please save the date. A request to register will be sent by e-mail in October.

Latest ERN GUARD-Heart Publications

1. Lahrouchi N, Raju H, Lodder EM, Papatheodorou S, Miles C, Ware JS, Papadakis M, Tadros R, Cole D, Skinner JR, Crawford J, Love DR, Pua CJ, Soh BY, Bhalshankar JD, Govind R, Tfelt-Hansen J, Winkel BG, van der Werf C, Wijeyeratne YD, Mellor G, Till J, Cohen M, Tome-Esteban M, Sharma S, Wilde AAM, Cook SA, Sheppard MN, Bezzina CR, Behr ER. The yield of postmortem genetic testing in sudden death cases with structural findings at autopsy. *Eur J Hum Genet.* 2019 Sep 18. doi: 10.1038/s41431-019-0500-8.
2. Raju H, Ware JS, Skinner JR, Hedley PL, Arno G, Love DR, van der Werf C, Tfelt-Hansen J, Winkel BG, Cohen MC, Li X, John S, Sharma S, Jeffery S, Wilde AAM, Christiansen M, Sheppard MN, Behr ER. Next-generation sequencing using microfluidic PCR enrichment for molecular autopsy. *BMC Cardiovasc Disord.* 2019 Jul 23;19(1):174. doi: 10.1186/s12872-019-1154-8.
3. Fellmann F, van El CG, Charron P, Michaud K, Howard HC, Boers SN, Clarke AJ, Duguet AM, Forzano F, Kauferstein S, Kayserili H, Lucassen A, Mendes Á, Patch C, Radojkovic D, Rial-Sebbag E, Sheppard MN, Tassé AM, Temel SG, Sajantila A, Basso C, Wilde AAM, Cornel MC; on behalf of European Society of Human Genetics, European Council of Legal Medicine, European Society of Cardiology working group on myocardial and pericardial diseases, European Reference Network for rare, low prevalence and complex diseases of the heart (ERN GUARD-Heart), Association for European Cardiovascular Pathology. European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. *Eur J Hum Genet.* 2019 Jun 24. doi: 10.1038/s41431-019-0445-y.

Upcoming Cardiology Events

Update in Clinical Cardiology	16 – 18 October 2019	Boston – Massachusetts
Advanced Course on prevention of Sudden Cardiac Death	18 and 19 October 2019	Milan – Italy
Patient Meeting CMP	26 October 2019	Bucharest - Romania
Patient Meeting CPVT	9 November 2019	Amsterdam UMC – The Netherlands
Patient and Experts PLN Meeting	28 November 2019	Murcia – Spain
Boardmeeting ERN GUARD-Heart	14 February 2020	Rome - Italy

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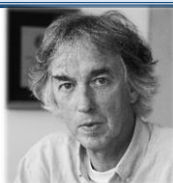
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