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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 3

## ERN exchange programme 2021-2022

**Amsterdam, 26 May 2021**

The ERN Exchange Programme 2021-2022, funded by the European Commission, aims to promote the sharing of knowledge and collaboration between the healthcare professionals in the ERNs. In the coming two years, three editions of exchanges will take place. The first edition has started in March 2021. The project ends in August 2022.

For ERN GUARD-Heart (as a relatively small ERN compared to other ERN), the programme includes 34 so-called exchange packages. A single package provides compensation for:

- ✔ 5 working days for 1 person
- ✔ Travel arrangements are included
- ✔ 200,- Euros per working day (for the cost of accommodation, travel insurance, food and local transport).
- ✘ It will not include insurance for professional liability for visitors and any costs uncured by the hosting healthcare providers (HCPs).

How to apply for an exchange visit:
- See page 2 -

***Content of the visits***

The exchange programme is meant to meet the goals and strengthen the capacity on the ERN level. It is not meant for research nor for individual development (though this may be a secondary effect of the exchange programme). The thematic scope includes medical practice and skills but also organisational aspects of a network. Each ERN defines the strategic goals and priorities of the exchange programme, according to the specific situation of the network. These goals and priorities include:

- geographical scope (priority countries/HCPs , equal distribution over countries);
- professional groups (medical specialists, nursing staff, lab staff, others);
- thematic coverage (clinical aspects, psychosocial support, organisational strengthening).

***Who is eligible***

Health professionals working in the ERN centres are eligible for the programme – this includes all disciplines related to the relevant expertise area of the ERN. Formally affiliated member HCPs are also eligible for exchange visits. Professionals coming from a third country and professionals that are not affiliated to the ERN are not eligible for exchange visits. HaDEA/SANTE accept limited participation of patients in travels if these are related to projects linked to ERNs activities. Equally, hosting centres should be approved ERN members. Exchange visits to (members of) other ERNs are allowed. Visits to centres outside the EU27+Norway are not foreseen. ♥

# How to apply for an exchange visit

***- See information on page 1 –***

Please contact the project management team if you have a suggestion for an ERN exchange visit (either as a hosting centre or to be a visitor). Describe your suggestions in a few lines and include the thematic area (i.e., arrhythmias, cardiomyopathies, special electrophysiology conditions, or one of the new thematic areas congenital heart disease or other rare cardiac diseases). Your request will be discussed in the coordinating team and with the Ecorys team (which supports the EU with the mobility agreements). Once the activity is approved (by the EU) and the mobility agreement is signed, there is a minimum of 6 weeks before the visit can be started in order to be able to make proper arrangements. ♥

# Your preferences for the next Board meeting in September

Amsterdam, 26 May 2021

In the hope that the COVID-19 situation will further improve with the ongoing vaccinations across Europe, and remains under control, we would like to ask your preference for the next board meeting.

Please provide your opinion: should we organize a virtual meeting or a face-to-face meeting in the first week of September? Please follow the link to fill in the poll (just one question and a suggestion): https://docs.google.com/forms/d/e/1FAIpQLSdRDd6_jSdjQeadDweBf2vl88J7AJj5uIPHyLpIU76wMYm7iw/viewform?usp=sf_link ♥


# Coordinators Group Meeting

Brussels, 30 April 2021

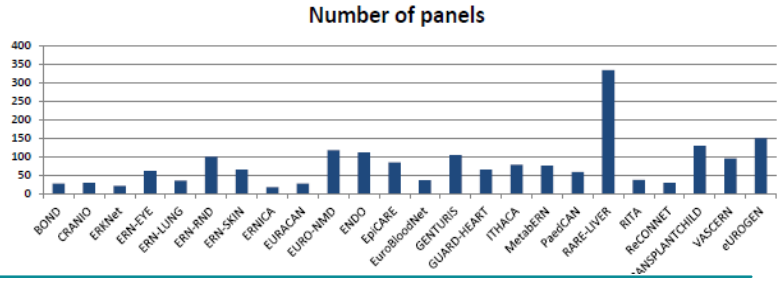
The latest ERN Coordinators Group meeting took place on 30 April. An important topic was the future of the ERNs and their funding. The European Commission presented the good news that the ERNs will be financed by the EU and the co-financing component of the future grants will be removed. Thus, the costs for the coordinating healthcare providers will be reimbursed for 100% (in stead of 60%). ♥

European Commission

# CPMS activity per ERN since November 2017

Amsterdam, 31 May 2021

Each two months the numbers of the CPMS activity (users and panels) are published by the ERN IT-support group. One of our goals is to increase the number of panels. The total number of registred users is not so important: because the costs are related to the number of users: only register if you really want to be a user. ♥



# Latest ERN GUARD-Heart Publications


1. Garcia-Pavia P, Bengel F, Brito D, Damy T, Duca F, Dorbala S, Nativi-Nicolau J, Obici L, Rapezzi C, Sekijima Y, Elliott PM. Expert consensus on the monitoring of transthyretin amyloid cardiomyopathy. Eur J Heart Fail. 2021 Apr 29. doi: 10.1002/ejhf.2198.

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4. Conte G, Wilde A, Behr ER, Scherr D, Lenarczyk R, Gandjbachkh E, Crotti L, Brugada-Sarquella G, Potpara T. Importance of Dedicated Units for the Management of Patients With Inherited Arrhythmia Syndromes. Circ Genom Precis Med. 2021 Apr;14(2):e003313. doi: 10.1161/CIRCGEN.120.003313. This publication is attached.


## New Educational Video Released

**Amsterdam, 20 May 2021**

Last week the 11th educational video (for professionals) was published online on the website and the YouTube channel. Anwar Baban (Rome. Italy) presents 'Genetic Counseling'. Hopefully some other new videos will follow soon. The ERN members who received a reminder last week will be contacted soon again to arrange an appointment for direct-recording in Zoom. Probably this will make it a little easier and speed things up. ♥

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## New Translations Patient Information Available

**Amsterdam, 20 May 2021**

Recently, the German translations of the patient folders have been published, and made available on the ERN website. Thanks to Eric Schülze-Bahr for this. Lithuanian and Hungarian translations are on their way as well. ♥

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Circulation: Genomic and Precision Medicine

RESEARCH LETTER

# Importance of Dedicated Units for the Management of Patients With Inherited Arrhythmia Syndromes

Giulio Conte, MD, PhD; Arthur Wilde, MD, PhD; Elijah R. Behr, MD; Daniel Scherr, MD; Radoslaw Lenarczyk, MD; Estelle Gandjbachkh, MD; Lia Crotti, MD; Georgia Brugada-Sarquella, MD, PhD; Tatjana Potpara, MD, PhD

The inherited arrhythmia syndromes (IAS) are a group of genetic heart diseases predisposing to sudden cardiac arrest.[1] Patients with IAS and their family members receive diagnostic and therapeutic management, which is heterogeneous across centers and suboptimal with regard to adherence to current guidelines. In particular, genetic testing, which is of utmost importance for its implications in the treatment of some IAS (ie, long QT syndrome, LQTS), is not always performed.[2,3]

The data that support the findings of this study are available from the European Heart Rhythm Association (EHRA) upon reasonable request. Additionally, Institutional Review Board approval was obtained by EHRA.

The aim of this EHRA survey analysis was to evaluate the relationship between the presence of dedicated IAS units, center volume, and management of patients with IAS. The EHRA Scientific Initiatives Committee conducted the present survey in collaboration with the European Cardiac Arrhythmia Genetics' Focus Group and European Reference Network (ERN )Gateway to Uncommon And Rare Diseases of the HEART (GUARD-HEART). A center-based online questionnaire was constructed to collect information about presence of dedicated IAS units, center volume, and diagnostic and therapeutic management of patients with the following diseases: Brugada syndrome, LQTS, early repolarization syndrome, catecholaminergic polymorphic ventricular tachycardia, and idiopathic ventricular fibrillation. Dedicated IAS unit was defined by the presence at a given Institution of a structured multidisciplinary service, including electrophysiologists specialized in IAS, device specialists, genetic counselors, and psychiatric support, for the management of patients and their family members who have a confirmed diagnosis or who are seeking an opinion regarding a possible diagnosis of IAS. The link was sent out to the EHRA Research Network Centers and ECGen members. Forty-four European centers were included in the analysis: 27 (61%) had a dedicated unit for the management of IAS patients, whereas there was no dedicated unit in the remaining 17 (39%; Table). Out of 27 centers with dedicated units, 10 (37%) managed >100 patients in the previous 12 months, whereas all centers without a dedicated unit had lower volumes. Moreover, centers without a dedicated unit were more likely to have very low volumes (<20 patients/y) of adults (47% versus 7%, $P<0.01$) and pediatric patients (87% versus 41%, $P=0.03$). There were no significant differences between centers on the use of pharmacological challenges in the diagnostic assessment of IAS. However, centers without a dedicated unit performed less genetic testing for all the different types of IAS, including those where a genetic diagnosis can influence therapeutic choices. Specifically, genetic testing for LQTS was performed in 92% and 59% of centers with and without dedicated units, respectively ($P=0.01$). Centers with a dedicated unit were more likely to perform an electrophysiology study with programmed ventricular stimulation for risk stratification (71% versus 41%) and substrate ablation procedures (82% versus 53%) for patients with Brugada syndrome.

In conclusion, dedicated IAS units frequently combine specialized care for adult and pediatric patients, genetic testing, and specific diagnostic and therapeutic procedures more frequently compared to centers with a low volume. However, treatment/outcome superiority of IAS units was

**Key Words:** arrhythmia, cardiac ■ Brugada syndrome ■ genetic testing ■ long QT syndrome ■ tachycardia

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For Sources of Funding and Disclosures, see page 253.



*Circulation: Genomic and Precision Medicine* is available at www.ahajournals.org/journal/circgen

## Nonstandard Abbreviations and Acronyms

| | |
|---|---|
| **IAS** | inherited arrhythmia syndromes |
| **LQTS** | long QT syndrome |
| **VF** | ventricular fibrillation |
| **VT** | ventricular tachycardia |

not examined in this survey. In the 2011 Hear Rhythm Society (HRS)/EHRA consensus statement on the state of art of genetic testing and 2013 HRS/EHRA/Asian Pacific Heart Rhyhtm Society (APHRS) expert consensus on the diagnosis and management of IAS, genetic testing was recommended for probands with a clinical diagnosis and for all family members of a successfully genotyped proband (class I recommendation).[1,2] In LQTS, the risk of life-threatening arrhythmic events, which is modulated by the duration of QTc interval and the genetic substrate, is not equal for all patients.[4] Specific gene mutations are associated with different arrhythmic risk and potential therapeutic benefits. Therefore, genetic testing in these patients has important prognostic implications due to the interplay between genetic substrate, QTc duration, and arrhythmia risk and impact on the response to pharmacotherapy.[4] Patients with LQTS not undergoing genetic testing may therefore not receive an appropriate therapeutic approach. Moreover, genetic testing, including pre- and post- genetic testing counseling, is valuable for identifying variants within genes known to be associated with increased risk for disease features and allows for predictive testing of at-risk family members.[2,3,5] According to this survey's results, underuse of genetic testing is more likely to occur in centers without dedicated IAS units. Therefore, we make strong plea for institutions to commit the creation and implementation of dedicated IAS units or, otherwise refer these patients to dedicated centers where they and their families can be seen in a multidisciplinary setting.[3] Further efforts to improve patient care in this setting are strongly warranted.

**Table. Patient Volume of Centers With and Without IAS Units and the Related Patients' Management.**

| | Centers with IAS units (N=27) | Centers without IAS units (N=17) | *P* value |
|---|---|---|---|
| No. of adult patients seen in the last 12 mo | | | |
| <20 | 2 (7%) | 8 (47%) | <0.01 |
| 20–50 | 7 (26%) | 7 (41%) | 0.33 |
| 50–100 | 8 (30%) | 2 (12%) | 0.27 |
| >100 | 10 (37%) | 0 | <0.01 |
| No. of centers managing pediatric patients | 22 (81%) | 8 (47%) | 0.02 |
| No. of pediatric patients seen in the last 12 mo | | | |
| <20 | 9/22 (41%) | 7/8 (87.5%) | 0.03 |
| 20–50 | 6/22 (27%) | 1/8 (12.5%) | 0.63 |
| 50–100 | 5/22 (23%) | 0 | 0.28 |
| >100 | 2/22 (9%) | 0 | 1.00 |
| Brugada syndrome | | | |
| Pharmacological challenge | 26/27 (96%) | 15/17 (88%) | 0.54 |
| Genetic testing | 24/27 (89%) | 9/17 (53%) | 0.05 |
| Electrophysiology study | 20/27 (74%) | 7/17 (41%) | 0.05 |
| Ventricular arrhythmogenic substrate ablation | 22/27 (82%) | 9/17 (53%) | 0.09 |
| AF ablation | 18/27 (67%) | 13/17 (76%) | 0.73 |
| Long QT syndrome | | | |
| Pharmacological challenge | 4/27 (15%) | 1/17 (6%) | 0.63 |
| Genetic testing | 25/27 (92%) | 10/17 (59%) | 0.01 |
| Early repolarization syndrome | | | |
| Pharmacological challenge | 5/27 (18%) | 0/17 | 0.13 |
| Genetic testing | 12/27 (44%) | 1/17 (6%) | <0.01 |
| Catecholaminergic polymorphic VT | | | |
| Pharmacological challenge | 6/27 (22%) | 2/17 (12%) | 0.45 |
| Genetic testing | 23/27 (85%) | 5/17 (29%) | <0.01 |
| Idiopathic VF | | | |
| Pharmacological challenge | 14/27 (52%) | 7/17 (41%) | 0.54 |
| Genetic testing | 21/27 (78%) | 3/17 (18%) | <0.01 |

AF indicates atrial fibrillation; IAS, inherited arrhythmia syndromes; VT, ventricular tachycardia; and VF ventricular fibrillation.

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