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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 2023 NUMBER 6

October ERN- Webinar: guidelines

Amsterdam, 02-10-2023

In the next ERN Webinar, on Wednesday 25 October at 13:00 till 13:45 there will be a presentation of the new guidelines for the management of cardiomyopathies. Please join this webinar and encourage you colleagues to do so. Registration is possible by the following link: https://forms.gle/5fE2CSHy73as8WM19. You can also find an introduction to these guidelines on page 5 of this Newsletter. ♥

Translation Patient Folders

Amsterdam, 02-10-202

The number of translations of the ERN patient folders has increased in the last year. Currently, ERN members are working on translations in the Portuguese and Ukrainian language. Our aim is to complete the list with translations with at least the languages of all countries of all our members and affiliated partners. Therefore, we invite centres from Zagreb (Croatian), Katowice (Polish), Dublin (Gaelic), Bratislava (Slovak), Tartu (Estonian), and Malta (Maltese) to help us with the completion of these translations. Please contact us for the details through the following email address: n.hofman@amsterdamumc.nl. ♥

Update grant proposal

Amsterdam, 19-09-2023

The grant proposal for the direct grant to the ERNs, which includes support for the coordination, management and operational activities of the ERNs has been re-submitted, after it was cancelled due to legal problems with Ukraine-related activities. In the new application, these activities have been, as requested, gathered into one separate work package. There was also an online meeting between ERN GUARD-Heart and one (of the four) cardiology centres in Kyiv (the Kyiv Heart Centre) who are looking forward to collaborate with the ERN and join our activities. The applications will be evaluated by the European Commission this month. The period for this 4-years grant started already on 1st of October 2023. ♥

Board-meeting in Amsterdam

Amsterdam, 28 August 2023

At Monday 28th of August, we had an ERN Board meeting in the Rosarium, a nice place conference centre in the Amstel Park in Amsterdam. With 38 participants on site and 22 online, almost all HCPs were represented. Thank you for that! It was a busy period, with the ESC congress during the days before, but, just as the previous years, it was practical to combine the Board meeting with the ESC congress, when many participants are already at place. ♥

Who are our ePAGs?

Amsterdam, 02-10-2023

One of the European patient advocates of ERN GUARD-Heart in the spotlight: ***Simone Louisse*** tells about herself and the activities of her patient organisation.

Since 2018, I combine my professional career with activities as a patient representative for a small foundation in the Netherlands and for ERNs in Europe as European Patient Advocate. Connecting patient representatives with each other and other stakeholders to increase their impact is one of my main personal goals.

In 1969 I was born in Amsterdam (The Netherlands) with a congenital heart disease called Tetralogy of Fallot. My 3rd "open heart" surgery I had when I was 12 and my last was in 2011 to replace my pulmonary valve. Despite the surgeries and a few medical events, I live a mostly normal life in a small town south of Amsterdam.

After obtaining a Master of Science in Neurobiology from the University of Amsterdam, I pursued a career in the market research company NielsenIQ. I currently work as validation team leader on the global data platform. NielsenIQ is one of the largest international market research companies).

In 2018 I joined a small Dutch foundation called Hart4Onderzoek (Heart4Research) to support its goal of raising funds for research for adults with congenital heart disease. Since 2021 we have shifted our focus to include also research and support for mental health and quality of life of the patients in our community. I am the treasurer of the organization since 2021.

With the money that we raise we have funded several small research projects of starting researchers. The last research project we supported was "experience of patients with congenital heart diseases with physical exercise and sports". We also had a successful patient day in March of 2023 linked to the same topic and supported by the ERN.

One of the fundraising events we participate in every year is the "Dam to Dam run". We have between 40 and 50 people running 5 or 10 miles and raising funds. The Netherlands UMC Hart Center is sponsoring us to enable our participation in the event.

In 2018, I became a member of the European Patient Advocacy Group (ePAG) for ERN GUARD-Heart, and soon after I joined the ePAG steering committee for all ERNs. I co-chair the committee since January 2023. With representatives from all 24 ERNs, we exchange best practices and work together to increase the impact of the ePAG and patient advocacy in general. EURORDIS supports us by training on leadership and other competencies.

As newly appointed chair for the Congenital group of the GUARD-Heart ePAGs, my most important aim is to connect the patient organizations (and their patients) with the thematic area leads and members in the ERN (clinicians/hospitals), and to improve the collaboration between these groups. Additionally, an important focus is the formal recognition of our role in the ERNs and stimulating a stronger involvement of ePAGs in defining ERN priorities and ERN Budget proposals. The patient-clinician feedback sessions and the informal meetings with the thematic area leads were a good starting point for a great collaboration in the future. ♥

More information can be found on the following website: Hart4Onderzoek.nl (In Dutch only at the moment)

Contact: slouisse62@gmail.com or penningmeester@hart4ondderzoek.nl

HCP Performance Indicators

Amsterdam, 28 August 2023

As presented in the ERN GUARD-Heart board meeting, the ERNs have to measure the activity of the separate ERN members as a part of the progress of the Network. This will be done besides the yearly 'monitoring exercise' with help of so-called 'performance indicators'. Within different areas points can be collected for specific ERN-activities (see table below). ***The evaluation will be conducted by 6-monthly status reviews***. Data can be used for benchmark studies between HCPs. Annual feedback will be shared with all HCPs in the Network. Improvement plans will be needed for centres with a total score <20%.

The activities can be counted from the beginning of the new grant period (01-10-2023), so the first evaluation will be done in Spring 2024. ***We strongly recommend each HCP to administrate their activities clearly from the beginning (from this week onward)!*** As the Network had a very positive 5-years evaluation by the European Commission, we don't expect any problems with the performance of our members. It is a way to collect and report clearly. Please don't hesitate to contact the Network management if you have any questions.

This evaluation method is mandatory for full members and affiliated partners, although the affiliated partners don't have to reach the 20% -limit.

Area	HCP Activity	Points	% of total
Training	Webinars per HCP (50 points per webinar) Clinical exchange: per host & visitor	100 50	15%
Patient education	Patient meetings (national) per HCP Laymen abstracts ERN publications per HCP (25 points each) Publications in laymen press (25 points each)	50 50 50	15%
ERN publications	Scientific ERN joint papers, registry based (25 points each) Scientific ERN joint papers, not-registry based (10 points each)	50 50	10%
Guidelines & Consensus Documents	Per guideline group leadership (per guideline 60 points) Per guideline group membership (per guideline 20 points)	60 40	10%
Registries (at least 50% of patients seen per HCP included)	Number of registries in which patients are enrolled: 1-2 3-4 ≥ 5	 75 150 250	25%
CPMS	Per case reviewed (per case 10 points) Per case submitted (per case 25 points)	50 50	10%
Monitoring	Timely delivery of complete data	100	10%
Contribution to ERN organization	Thematic area/ coordination per chair Attendance per board meeting per HCP	35 15	5%

Publication of 2023 guidelines for the management of cardiomyopathies

Barcelona, 04-10-2023
Author: Elena Arbelo

During the 2023 Congress of the European Society of Cardiology (ESC) in Amsterdam, (25–28 August) a new guideline on for the management of cardiomyopathies was published with the participation of several members of our ERN among the authors and reviewers.

This is the first guideline internationally to cover all cardiomyopathy subtypes except for the section on hypertrophic cardiomyopathy (HCM), for which we provide a focused update to the 2014 *ESC Guidelines on diagnosis and management of hypertrophic cardiomyopathy.*

There are several aspects to be highlighted:

First, the **focus is placed on the patient pathway**, from presentation, through initial assessment and diagnosis, to management, highlighting the importance of considering cardiomyopathy as a cause of common clinical presentations (e.g. heart failure, arrhythmia) and the importance of utilizing a multiparametric approach following the identification of the presenting phenotype to arrive at an aetiological diagnosis. The starting point for the diagnostic work-up of cardiomyopathies is a detailed description of the morphological and functional traits and these should include the identification of ventricular scar and other myocardial tissue identification on CMR (5 different cardiomyopathy phenotypes: HCM, DCM, NDLVC, ARVC and RCM). From there, additional traits need to be evaluated (including arrhythmias/conduction disturbances, extracardiac involvement, genetic testing, other laboratory parameters, pathology, etc.) to reach a more specific aetiology-based diagnosis, which will allow better management and risk stratification. Central to this approach is **not only the individual patient, but also the family as a whole**; clinical findings in relatives are essential for understanding what happens to the patient, and vice versa.

Also, **cardiomyopathies should be considered across the life course**, from paediatric to adult age (including transition), and considering the different clinical phases (e.g. concealed, overt, end stage).
A novel aspect of this GL is the importance of CMR not only for diagnosis but also to monitor disease progression and aid risk stratification and management. In fact, CMR is recommended in all patients with cardiomyopathy at **initial evaluation** and should be considered in **patients with cardiomyopathy** during **follow-up** to monitor disease progression and aid risk stratification and management. In families in which a **disease-causing variant** has been **identified** CMR should be considered in genotype-positive/phenotype-negative relatives. CMR may also be considered in case of no genetic diagnosis.

Another key aspect of the CM GLs is genetic testing, which is recommended in ALL patients fulfilling diagnostic criteria for cardiomyopathy where it enables diagnosis, prognostication, therapeutic stratification, or reproductive management of the patient, or where it enables cascade genetic evaluation of their relatives who would otherwise be enrolled into long-term surveillance. In adult family members, cascade genetic testing should be offered in case of a genetic diagnosis. In children, genetic testing should also be considered. Importantly, genetic counselling plays a key role in the management of these patients and families and in the shared-decision process around genetic testing.

The guidelines emphasize the need for a **co-ordinated, systematic, and individualized pathway** that delivers optimized care by a **multidisciplinary and expert team**. The composition of the multidisciplinary team will depend on the patient's and family's needs and the local availability of services. While referral cardiomyopathy units are essential for complex cases with diagnostic and/or treatment difficulties that require expertise that may only be available in high-volume centres, general adult and paediatric cardiologists have a key role to play in the diagnosis, management, and follow-up of patients with cardiomyopathy. Therefore, a shared-care approach between cardiomyopathy specialists and general adult and paediatric cardiology centres is strongly recommended.

In fact, the guideline underscores the role of expert networks section 5.2 Co-ordination between different levels of care:

The creation of/regional/national/ international networks, such as the European Reference Network for Rare and Low Prevalence Complex Diseases of the Heart (ERN GUARD-Heart) (https://guardheart.ern-net.eu) allows clinicians and health professionals to share information about these pathologies, for the benefit of cardiomyopathy patients." ♥

Latest ERN GUARD-Heart Publications

1. Brida M, De Rosa S, Legendre A, Ladouceur M, Dos Subira L, Scognamiglio G, Di Mario C, Roos-Hesselink J, Goossens E, Diller G, Gatzoulis MA. Acquired cardiovascular disease in adults with congenital heart disease. Eur Heart J. 2023 Sep 27:ehad570. doi: 10.1093/eurheartj/ehad570. Epub ahead of print. PMID: 37758198.

2. Larrañaga-Moreira JM, Rodriguez-Serrano AI, Domínguez F, Lalario A, Zorio E, Barriales-Villa R; Dilemma International Cardiomyopathy, Heart Failure Registry Investigators Group. Impact of SARS-CoV-2 infection in patients with cardiac amyloidosis: Results of a multicentre registry. Med Clin (Barc). 2023 Sep 6:S0025-7753(23)00399-8. English, Spanish. doi: 10.1016/j.medcli.2023.06.025. Epub ahead of print. PMID: 37684159.

3. van Dissel AC, Opotowsky AR, Burchill LJ, Aboulhosn J, Grewal J, Lubert AM, Antonova P, Shah S, Cotts T, John AS, Kay WA, DeZorzi C, Magalski A, Han F, Baker D, Kay J, Yeung E, Vonder Muhll I, Pylypchuk S, Kuo MC, Nicolarsen J, Sarubbi B, Fusco F, Jameson SM, Cramer J, Gupta T, Gallego P, O'Donnell C, Hannah J, Dellborg M, Kauling RM, Ginde S, Krieger EV, Rodriguez F, Dehghani P, Kutty S, Wong J, Wilson WM, Rodriguez-Monserrate CP, Roos-Hesselink J, Celermajer DS, Khairy P, Broberg CS. End-stage heart failure in congenitally corrected transposition of the great arteries: a multicentre study. Eur Heart J. 2023 Sep 7;44(34):3278-3291. doi: 10.1093/eurheartj/ehad511. PMID: 37592821; PMCID: PMC10482567.

4. Neumann B, Vink AS, Hermans BJM, Lieve KVV, Cömert D, Beckmann BM, Clur SB, Blom NA, Delhaas T, Wilde AAM, Kääb S, Postema PG, Sinner MF. Manual vs. automatic assessment of the QT-interval and corrected QT. Europace. 2023 Aug 2;25(9):euad213. doi: 10.1093/europace/euad213. PMID: 37470430; PMCID: PMC10469369.

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