

Monday 10 June 2024



GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart



ERN GUARD-HEART BIMONTHLY NEWSLETTER

YEAR 2024 NUMBER 3



Think
Cardiomyopathy!
Understand your
family's heart
health.

Cardiomyopathy Awareness 2024

'Think Cardiomyopathy'

Amsterdam, 06-06-2024

June is the Cardiomyopathy Awareness month in 2024. On the website of the Global Heart Hub ([Think Cardiomyopathy - Global Heart Hub](#)), there are videos available, infographics, and key messages which are translated into 10 different languages. Please, spread the content. Many of our ePAGs are involved in this initiative. ♥

Next board meeting ERN GUARD-Heart

Amsterdam, 06-06-2024

The next ERN GUARD-Heart board meeting will be held online at Friday morning 27 September 2024. ♥



Joint action on Integration of ERNs into National Healthcare systems - JARDIN



Amsterdam, 06-06-2024

To improve the accessibility of the ERNs for patients in member states, the EU is funding a pioneering 3-year project involving all member states plus Norway and Ukraine, the Joint Action on Integration of ERNs into National Healthcare Systems ('JARDIN'). JARDIN will be coordinated by Professor Till Voigtländer from the Medical University of Vienna in Austria and will produce recommendations, and implementation pilots in the main fields of action, such as patient pathways, national reference networks, and data management for rare diseases. These pilots constitute a promising way to provide EU member states with directly implementable solutions for their healthcare systems. JARDIN's kick-off meeting was held in Brussels from 6th-8th March 2024.

ERN GUARD-Heart is not actively involved as participant in JARDIN, but we will be requested to answer different surveys. Those surveys will be sent by JARDIN. The initial survey was to identify contacts who are best suited to support the Joint Action in subsequent surveys. It is still possible to respond to it: a unique opportunity for all ERN members to be represented in this project and be recognized at the national level and ultimately receive better support and funding opportunities in the future.

If you are interested, please, complete the survey here: <https://ec.europa.eu/eusurvey/runner/Survey1JARDIN>

The deadline to complete the survey is June 14th, 2024. ♥



Who are our ePAG's? Katja Laine - Finland

Helsinki, 10-06-2024

One of the European patient advocates ERN GUARD-Heart in the spotlight: Katja Laine, from Finland, tells her story and her active way to cope with it.

My journey with congenital heart defects started almost 28 years ago, when my daughters complex heart defect was diagnosed when she was three days old. There was no indication of the CHD during the pregnancy, so the diagnose was a huge shock for us. She was diagnosed with pulmonal atresia (PA), double outlet right ventricle (DORV), large ventricular septal defect (VSD) and so-called criss-cross heart, which is very rare defect. All these diagnoses meant, that her heart defect could not be fixed and the only option that would allow her to survive, would be changing her blood circulation to a univentricular heart (UVH). That meant multiple operations and only time would tell how things would go on. Before we got out of the hospital, she was operated on twice already. During the years she has had four operations all together and multiple cardiac catheterizations.

At the time she was born (1996) there wasn't much if any peer support available, especially when it came to UVH patients. There weren't any self-help groups available in Finland as there are now-a-days on the internet or on Facebook. Basically, the only option was to contact the patient organisation and thankfully we were able to find a few families and patients with similar heart defects. That gave us lot of hope and faith for the future – when the doctors were not able to do so. We were attending a lot of different kinds of events and camps, that the Finnish Association for Heart Children and Adults were organising, and we found those very helpful. After a little while, I did find myself working as a volunteer and peer-support person organising events for the heart families in the area we lived in. My driving force has been and still is the gratitude for the help our family got during those years – without the help we would have not managed! And if I can help even one family living in a similar situation as we had in the beginning, it makes it totally worth it!



After a few years, I ended up working for the association. So, in many ways my daughters heart defect also affected my professional life. I have worked as an executive director of the Finnish Association for Heart Children and Adults for more than 20 years now, and I still find it very awarding. The association aims to improve the comprehensive care for individuals with CHD and in-born heart diseases through its activities, as well as to promote and support the treatment, rehabilitation, and well-being of children, adolescents, and adults in the target group. In addition to advocacy and lobbying, important activities include courses and camps combined with peer support and recreational activities, where participants can gain information and meet peers. Giving information about CHD for families and professionals is also a significant part of the association's work. It is important to us that no child, adolescent, or adult with a heart condition, nor their family members, feel alone in their situation. Next year, we will celebrate our 50-year anniversary of working towards these goals.

Today my daughter is doing relatively well despite of the heart defect, and she is living an active life with her family. She works as a nurse helping people with mental health issues. She is also very active in the association and a peer-support person herself. ♥





CPMS 2.0 Desktop version now available

Luxembourg, 31 May 2024



The CPMS 2.0 desktop version was launched successfully at Friday, 31st of May 2024. This secure, simplified and flexible platform empowers healthcare professionals across the EU and Ukraine to collaborate more effectively on rare medical cases, marking a significant milestone for the ERN community.

The data transfer of user and patient data from the old platform is ongoing. This process involves careful checks and requires individual ERN involvement. Please refrain from creating user profiles in the new platform – access for ERN clinicians will be automatically granted once their credentials are migrated. New ERN users can be created after switch-over. Once the data transfer for our ERN has been finished, we will provide you with detailed instructions for a smooth transition. Thereafter we will start with the thematic area based meetings for the case-discussions. CPMS 2.0 mobile is on the horizon! Pending approval from Google and Apple, the app will be available on their respective App Stores shortly. ♥

Latest ERN GUARD-Heart Publication (s)

1. Ishikawa T, Masuda T, Hachiya T, Dina C, Simonet F, Nagata Y, Tanck MWT, Sonehara K, Glinge C, Tadros R, Khongphatthanayothin A, Lu TP, Higuchi C, Nakajima T, Hayashi K, Aizawa Y, Nakano Y, Nogami A, Morita H, Ohno S, Aiba T, Juárez CK, Mauleekoonphairoj J, Poovorawan Y, Gourraud JB, Shimizu W, Probst V, Horie M, Wilde AAM, Redon R, Juang JJ, Nademanee K, Bezzina CR, Barc J, Tanaka T, Okada Y, Schott JJ, Makita N. Brugada syndrome in Japan and Europe: a genome-wide association study reveals shared genetic architecture and new risk loci. *Eur Heart J*. 2024 May 15;ehae251. doi: 10.1093/eurheartj/ehae251. Epub ahead of print. PMID: 38747976.
2. Albertini M, Santens B, Fusco F, Sarubbi B, Gallego P, Rodriguez-Puras MJ, Prokselj K, Kauling RM, Roos-Hesselink J, Labombarda F, Van De Bruaene A, Budts W, Waldmann V, Iserin L, Woudstra O, Bouma B, Ladouceur M. External Validation of a Risk Score Model for Predicting Major Clinical Events in Adults After Atrial Switch. *J Am Heart Assoc*. 2024 May 7;13(9):e032174. doi: 10.1161/JAHA.123.032174. Epub 2024 Apr 30. PMID: 38686874.
3. Lenarczyk R, Zeppenfeld K, Tfelt-Hansen J, Heinzel FR, Deneke T, Ene E, Meyer C, Wilde A, Arbelo E, Jędrzejczyk-Patej E, Sabbag A, Stühlinger M, di Biase L, Vaseghi M, Ziv O, Bautista-Vargas WF, Kumar S, Namboodiri N, Henz BD, Montero-Cabezas J, Dagres N, Peichl P, Frontera A, Tzeis S, Merino JL, Soejima K, de Chillou C, Tung R, Eckardt L, Maury P, Hlivak P, Tereshchenko LG, Kojodjojo P, Atié J. Management of patients with an electrical storm or clustered ventricular arrhythmias: a clinical consensus statement of the European Heart Rhythm Association of the ESC-endorsed by the Asia-Pacific Heart Rhythm Society, Heart Rhythm Society, and Latin-American Heart Rhythm Society. *Europace*. 2024 Mar 30;26(4):euae049. doi: 10.1093/europace/euae049. PMID: 38584423; PMCID: PMC10999775.

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