

Saturday 01 October 2022

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GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart



ERN GUARD-HEART BIMONTHLY NEWSLETTER

YEAR 2022 NUMBER 6

ERN GUARD-Heart Webinars

Amsterdam, 29 September 2022

Last summer, ERN GUARD-Heart started a series of Webinars for healthcare professionals. All interested (multidisciplinary) colleagues from full ERN member and Affiliated Partner hospitals are invited to register for general information and announcements about these webinars.*

The first two Webinars were held and the number of attendees increases, although the number of participants lacks behind the number of subscriptions. Evaluation will take place to try to improve the direct connected number of participants. The webinars are recorded and will be available via the ERN website. During each Webinar 1 or 2 patients will be discussed by anyone who wants to discuss a case and will be evaluated by 1 or 2 ERN expert(s). Thereafter all participants will be encouraged to ask questions and to discuss the cases. Each webinar will cover one specific thematic area, and will take no more than 45 minutes. Currently, about 225 participants registered already. For each webinar, a flyer will be send to the webinar-email-list with the topic, title, and name of the speaker and a registration link. A separate registration per webinar is needed. Thereafter the link to the meeting will be shared.

* Registration to the general Webinar email-list is possible via this link:

<https://forms.gle/6KZjrnsECvJncbwK9> . ♥

Extra packages for Exchange Programme available

Amsterdam, 21 July 2022

The ERN Exchange Programme is running until the end of the year and at this moment our available packages are reserved or already used. However, we are allowed to request for extra packages which are still unused by other ERNs. It is a very nice opportunity to visit one of the expert centres in the ERN for a 5 days (or somewhat longer, up to 30 days). It works on a 'first come, first served' basis. Please contact Nynke Hofman (n.hofman@amsterdamumc.nl) for the procedures and forms. Since the exchanges have to take place before the 31 January 2023 (and the paperwork should be finished before the end of this year), it is important not to wait too long anymore. ♥



European
Reference
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challenges
today



ERN-dinner and Board Meeting in Barcelona

Amsterdam, 26 September 2022

On Monday-evening 29 August 2022, the ERN Board members had a dinner together in Barcelona at restaurant Dos Torres. With great food and, thanks to the concept, a lot of opportunities to talk to each other, it was a very nice evening. Thanks to Georgia Sarquella-Brugada who organized this very well!. The Board Meeting was held 30 August in Sant Joan de Déu. This was after a long period with only online meetings, our first hybrid meeting and with 39 attendees in the room physically and 25 online, it was good represented. Thanks to Georgia and her team again for the nice and well-equipped meeting room, great technical assistance and even 'ERN-cookies' on the coffee-table! ❤





15-16-17 March 2023: ERN GUARD-Heart Board Meeting and EJPRD Networking Symposium

Amsterdam, 26 September 2022

On Wednesday 15 March 2023, the ERN project management organizes a hybrid ERN GUARD-Heart board meeting in Amsterdam and dinner in the evening. This meeting will be combined with a two-day symposium and networking event on the genetics of inherited arrhythmia syndromes, probably of interest to all the centres who join the ERN in thematic area 1 (familial electrical diseases in adults and children).

The symposium is organized by the Amsterdam UMC and ERN GUARD-Heart and is funded by the EJP RD Networking Support Scheme. It will include talks on cutting edge research from some of the world's leading investigators in this field. It will also include discussions on developing the new and existing networks required to further progress research in this field and translate recent advances into clinical applicability - the development of multicentre patient registries, national and international databases and future collaborative projects and grant applications. The complete programme is available at page 4 and 5 of this newsletter.

How to register for the symposium?

<https://www.eventbrite.com/e/ejprd-networking-symposium-complex-genetics-of-inherited-arrhythmias-tickets-399392793597>

Please note that the numbers of tickets for in-person attendance is limited, so you should book early if interested. There is also an option for online only attendance (for Day 1 only).

Invitation for Abstracts

Abstracts are invited for this symposium, especially from early career researchers. A number of submitted abstracts will be selected for talks during Session 6. There will also be poster viewing for all submitted abstracts. Please send abstracts to genetics_abstracts@amsterdamumc.nl (abstract deadline: 01/02/2023).

How to register for the ERN Board-meeting and dinner on Wednesday 15 March 2021?

The ERN GUARD-Heart management office will send an e-mail with a link to a form on which you can fill in your preferences. For ERN members who come to Amsterdam there will be travel costs reimbursed up to 300,- EURO, and a one night stay in a hotel-room will be covered as well (for one person per HCP). For speakers the travel tickets and a two night stay will be arranged. They will also be contacted personally about this.

For more information, please contact the meeting organizers:

Connie Bezzina (c.r.bezzina@amsterdamumc.nl)

Nynke Hofman (n.hofman@amsterdamumc.nl)

Roddy Walsh (r.t.walsh@amsterdamumc.nl). ❤

*Complex genetics of inherited arrhythmias:
moving from the research lab to the clinic*



**PROGRAMME SYMPOSIUM 'COMPLEX GENETICS OF INHERITED ARRHYTHMIAS'****DAY 1 – Thursday, March 16th 2023****0830 – 0900: Registration****0900 – 0915: Welcome and Introduction - Connie Bezzina (Netherlands)****0915 – 1030: Session 1 - Current state of the art for clinical genetic testing for inherited arrhythmias**

1. Genetic architecture of inherited arrhythmia conditions - Arthur Wilde (Netherlands)
2. How do results from genetic testing inform clinical management? – Tomas Robyns (Belgium)
3. Challenges of genetic testing outside of major research centres - Ruxandra Jurcut (Romania)

*Session moderators: Mark Abela (Malta), Annika Rydberg (Sweden)***1030– 1100: Coffee Break and Poster Viewing****1100 – 1230: Session 2 - Recent advances in arrhythmia genetics**

1. Variant interpretation - challenges and innovative approaches - Roddy Walsh (Netherlands)
2. High throughput functional studies to assess variant pathogenicity at scale - Andrew Glazer (USA)
3. Genome-wide association studies (GWAS) in Brugada syndrome and LQTS – Connie Bezzina (Netherlands)
4. Genome-wide association studies (GWAS) in ARVC – Peter van Tintelen (Netherlands)

*Session moderators: Stefen Kääh (Germany), Joseph Galvin (Ireland)***1230 – 1300: Session 3 - The patient perspective – how genetic testing impacts patients and families - Ruth Biller (PAO - ARVC-Selbsthilfe - Germany)****1300 – 1400: Lunch and Poster Viewing****1400 – 1515: Session 4 - The genetic dark side of under characterised arrhythmia conditions**

1. Idiopathic ventricular fibrillation and unexplained cardiac arrest - Elijah Behr (UK)
2. Early repolarisation syndrome - Julien Barc (France)
3. Clinical overlap between Brugada syndrome and Atrioventricular nodal re-entry tachycardia - Can Hasdemir (Turkey)

*Session moderators: Johan Saenen (Belgium), Jacob Tfelt-Hansen (Denmark)***1515 – 1545: Coffee Break and Poster Viewing****1545 – 1700: Session 5 - Challenges for deriving insights from GWAS**

1. The use of endophenotype analysis in biobank datasets to gain new insights into genetics of rare cardiac diseases - Rafik Tadros (Canada)
2. Insights into phenotype penetrance from PRS in biobank studies - James Ware (UK)
3. PRS: ready for prime time? - Dan Roden (USA)

Session Moderators: Philippe Charron (France), Juan Gimeno (Spain)

**DAY 2 – Friday, March 17th 2023****0900 – 1015: Session 6 – Talks by early career researchers chosen from submitted abstracts**

Session moderators: Elena Arbelo (Spain), Eric Schulze Bahr (Germany)

1015 – 1045: Coffee Break and Poster Viewing**1045 – 12.45: Session 7 (working lunch) – Discussions for future networking, multi-centre registries, grant proposals**

The second half of the morning on day 2 will be devoted to a series of discussions and breakout sessions to address the following issues:

- Challenges and new studies required to move towards complex genetic testing for arrhythmia patients and families in the clinic?
- What can we learn from advances in other cardiac diseases?
- Developing new and existing networks, collaborations and multi-centre registries.
- Future grant applications – which topics to address, what are the expertise and contributions from different centres.
- Building national and international databases

Session moderators: Connie Bezzina (Netherlands), Lia Crotti (Italy)

12.45-13.00: Closing remarks

Teaching Webinars for Congenital Heart Disease professionals

Amsterdam, 28 September 2022

The Association for European Paediatric and Congenital Cardiology (AEPC) together with the ERN GUARD-Heart centers of thematic area 4 (Congenital Heart Disease) are planning weekly interactive teaching webinars on congenital heart disease. These webinars will start before the end of this year and will be hosted by the AEPC office. The webinars will cover all topics of congenital heart disease required for the basic training in paediatric and congenital cardiology. Members of the AEPC Working Groups as well as professionals from the GUARD-Heart centers will actively participate in this joint educational program. ♥

Welcome to new ePAGs from Italy

Amsterdam, 28 September 2022

The list of the European Patient Advocacy Groups in ERN GUARD Heart increases. Recently, the applications of two associated partners from Italy were approved: Campioni Associazione Italiana dei Cardiopatici Congeniti bambini e Adulti (AICCA Onlus), represented by Giovanna Campioni and Italy AICARM: Associazione Italiana per lo studio e la ricerca delle CARDioMiopatie (Italian Association for the study and research of CARDioMyopathies), represented by Matteo Pincioli, are new ePAG-members. Now there are 14 associated ePAG-members and 5 supporting partners included in ERN GUARD-Heart. New patient organizations, particularly from EU Member States which are not represented yet with a patient organization in the ERN (i.e. Belgium, Czech Republic, Hungary, Croatia, Ireland, Poland and Portugal) are very welcome. Also patient organizations from Member States with an affiliated partner can introduce patient representatives. For more information please contact the project management team (n.hofman@amsterdamumc.nl) or Ruth Biller (familie.biller@gmx.de) chair of the ePAGs of ERN GUARD Heart. ♥



Latest ERN GUARD-Heart Publication(s)

- Clinical Risk Score to Predict Pathogenic Genotypes in Patients With Dilated Cardiomyopathy.** Escobar-Lopez L, Ochoa JP, Royuela A, Verdonschot JAJ, Dal Ferro M, Espinosa MA, Sabater-Molina M, Gallego-Delgado M, Larrañaga-Moreira JM, Garcia-Pinilla JM, Basurte-Elorz MT, Rodríguez-Palomares JF, Climent V, Bermudez-Jimenez FJ, Mogollón-Jiménez MV, Lopez J, Peña-Peña ML, Garcia-Alvarez A, López-Abel B, Ripoll-Vera T, Palomino-Doza J, Bayes-Genis A, Brugada R, Idiazabal U, Mirelis JG, Dominguez F, Henkens MTHM, Krapels IPC, Brunner HG, Paldino A, Zaffalon D, Mestroni L, Sinagra G, Heymans SRB, Merlo M, Garcia-Pavia P. *J Am Coll Cardiol.* 2022 Sep 20;80(12):1115-1126. doi: 10.1016/j.jacc.2022.06.040. PMID: 36109106.
- Indications and utility of cardiac genetic testing in athletes.** Castelletti S, Gray B, Basso C, Behr ER, Crotti L, Elliott PM, Gonzalez Corcia CM, D'Ascenzi F, Ingles J, Loeys B, Pantazis A, Pieleles GE, Saenen J, Sarquella Brugada G, Sanz de la Garza M, Sharma S, Van Craenebroek EM, Wilde A, Papadakis M. *Eur J Prev Cardiol.* 2022 Sep 7;29(12):1582-1591. doi: 10.1093/eurjpc/zwac080. PMID: 36070487.
- Device-related complications in the subcutaneous and transvenous ICD: a secondary analysis of the PRAETORIAN trial.** Knops RE, Pepplinkhuizen S, Delnoy PPHM, Boersma LVA, Kuschyk J, El-Chami MF, Bonnemeier H, Behr ER, Brouwer TF, Kaab S, Mittal S, Quast AFBE, van der Stuijt W, Smeding L, de Veld JA, Tijssen JGP, Bijsterveld NR, Richter S, Brouwer MA, de Groot JR, Kooiman KM, Lambiase PD, Neuzil P, Vernooy K, Alings M, Betts TR, Bracke FALE, Burke MC, de Jong JSSG, Wright DJ, Jansen WPJ, Whinnett ZI, Nordbeck P, Knaut M, Philbert BT, van Opstal JM, Chicos AB, Allaart CP, Borger van der Burg AE, Dizon JM, Miller MA, Nemirovsky D, Surber R, Upadhyay GA, Weiss R, de Weger A, Wilde AAM, Olde Nordkamp LRA. *Eur Heart J.* 2022 Aug 28;ehac496. doi: 10.1093/eurheartj/ehac496. Online ahead of print. PMID: 36030464.
- Should variants of unknown significance (VUS) be disclosed to patients in cardiogenetics or not; only in case of high suspicion of pathogenicity?** van der Crabben SN, Mörner S, Lundström AC, Jonasson J, Bikker H, Amin AS, Rydberg A, Wilde AAM. *Eur J Hum Genet.* 2022 Aug 26. doi: 10.1038/s41431-022-01173-z. Online ahead of print. PMID: 36008533.
- Natural History of MYH7-related Dilated Cardiomyopathy.** de Frutos F, Ochoa JP, Navarro-Peñalver M, Baas A, Bjerre JV, Zorio E, Méndez I, Lorca R, Verdonschot JA, García-Granja PE, Bilinska Z, Fatkin D, Fuentes-Cañamero ME, García-Pinilla JM, García-Álvarez MI, Girolami F, Barriales-Villa R, Díez-López C, Lopes LR, Wahbi K, García-Álvarez A, Rodríguez-Sánchez I, Rekondo-Olaetxea J, Rodríguez-Palomares JF, Gallego-Delgado M, Meder B, Kubanek M, Hansen FG, Restrepo-Córdoba MA, Palomino-Doza J, Ruiz-Guerrero L, Sarquella-Brugada G, Perez-Perez AJ, Bermúdez-Jiménez FJ, Ripoll-Vera T, Rasmussen TB, Jansen M, Sabater-Molina M, Elliot PM, Garcia-Pavia P; European Genetic Cardiomyopathies Initiative Investigators. *J Am Coll Cardiol.* 2022 Aug 11:S0735-1097(22)05713-8. doi: 10.1016/j.jacc.2022.07.023. PMID: 36007715.
- COVID-19 vaccination in patients with long QT syndrome.** Wu CI, Schwartz PJ, Ackerman MJ, Wilde AAM. *Heart Rhythm* 02. 2022 Aug 3. doi: 10.1016/j.hroo.2022.07.011. Online ahead of print. PMID: 35937046 Free PMC article. Review.
- 2022 ESC Guidelines for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death.** Zeppenfeld K, Tfelt-Hansen J, de Riva M, Gregers Winkel, Behr ER, Blom NA, Charron P, Corrado D, Dagres N, de Chillou C, Eckardt L, Friede T, Haugaa KH, Hocini M, Lambiase PD, Marijon E, Merino JL, Peichl P, Priori SG, Reichlin T, Schulz Menger J, Sticherling C, Tzeis S, Verstraël A, Volterrani, M, and ESC Scientific Document Group. *Eur. Heart Journal* (2022) 00; 1-130. <https://doi.org/10.1093/eurheartj/ehac262>

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