Monday 2 August 2021

ERN GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES

Share. Care. Cure.

ERN GUARD-HEART BIMONTHLY NEWSLETTER YEAR 2021 NUMBER 4

10th board meeting online on 15 September 2021

Amsterdam, 27 July 2021

The 10th board meeting of the ERN GUARD-Heart will be held online, on Wednesday 15 September 2021 (from 14.00 till 17.00).
Due to the unstable COVID situation, unfortunately, it is not reliable to organize a physical or hybrid meeting. The provisional agenda will be send as a separate attachment by e-mail. ♥

EJP RD funded ERN Training Workshop in Lyon

Lyon, 17 June 2021

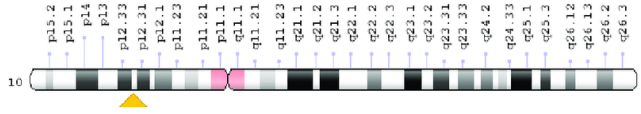
The 'Research Training Workshop' Call of the EJP aims at identifying workshop topics to train ERN researchers and/or clinicians in relevant innovative themes with a cross-ERN added value. Selected applicants will receive financial support for the organization of a 2-days workshops for approximately 20 participants. The team of Philippe Chevalier (Lyon, France) has received a positive response on their application for a workshop on 'functional studies of genetics variants found in patients with cardiac and neuromuscular diseases'. The date for the workshop is not yet known. ♥

EUROPEAN JOINT PROGRAMME RARE DISEASES

CACNA1C Registry is open

Rome, 15 July 2021

The team of Fabrizio Drago and Anwar Baban from Bambino's Gesu Hospital in Rome initiated and created the CRF for the online registry for *CACNA1C* mutation carriers. The REDCap Registry has been built in Amsterdam and is in production now.
It would be valuable if all the centres who take care of individuals or families with CACNA1C mutations would include their data in this registry, to put together the results and be able to improve the characterization of this population. The registry is user-friendly. To add you patients to this registry, we have to add the registry to your REDCap account, or, if you don't have an account already, we have to create one. This can be done very easily.
Please send a message to Fabrizio and Anwar (fabrizio.drago@opbg.net and anwar.baban@opbg.net), as registry leaders, in which you explain how many patients you would like to add to the registry, and put Nynke (n.hofman@amsterdamumc.nl) in the c.c., then the technical issues related to the accounts will be organized as well. ♥



REDCap
Research Electronic Data Capture

ERN News

Reminder: ERN exchange programme 2021-2022

Amsterdam, 27 July 2021

ERN Exchange Programme 2021-2022, funded by the European Commission, aims to promote the sharing of knowledge and collaboration between healthcare professionals in the ERNs. In the next two years, three editions of exchanges will take place. The first edition has started in March 2021. The project ends in August 2022. The program is open to clinicians and other healthcare professionals and invites both junior and experienced professionals. Both visitors and hosts must work in ERN GUARD-Heart member centres or Affiliated Partners.

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

- ✔ 5 working days for 1 person
- ✔ Travel arrangements
- ✔ 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 HCP. ♥

First exchanges on rare kidney disease: The ERKNet experience

The European Rare Kidney disease Network (**ERKNet**) took the honour of organizing the first exchange visit in the context of the ERN Exchange Programme. In the last week of June, two junior doctors, a nephrologist and a resident, from the Azienda Ospedaliera Papa Giovanni XXIII hospital in Bergamo, travelled to the Necker-Enfants Malades University Hospital in Paris for a two-week visit. The nephrologist attended activities of the ward and the outpatient clinics in order to improve experience in the diagnosis and treatment of nephrotic syndrome and complement disorders. The resident doctor engaged in knowledge sharing on genetic counselling and molecular pre- and post-natal testing as well as paediatric kidney transplantation.

In July, a senior and a junior paediatric nephrologist – from Necker-Enfants Malades University Hospital in Paris – visited Heidelberg University Hospital in Germany. They worked with their German colleagues to compare clinical practice, personalized immunosuppressive therapy before and after transplantation, and shared experiences about peritoneal dialysis in children. Please read their experiences below for inspiration for an exchange proposal. Exchanges are available for up to 3 weeks (several packages can be used for one visit if requested).

Myocarditis after vaccination with mRNA vaccines

Munich, 28 July 2021

EMA's safety committee (PRAC) is recommending to list myocarditis and pericarditis as new side effects in the product information for mRNA vaccines, together with a warning to raise awareness among healthcare professionals and people taking these vaccines. So does the German Paul-Ehrlich-Institute (PEI).

The German patient organisation ARVC Selbsthilfe e.V. is raising a question for the experts of genetic heart diseases. Well known to experts, ARVC/ACM is a condition:
- which occurs more frequently in men and often becomes symptomatic age 20-40 years,
- is sometimes initially diagnosed as myocarditis (although this is more common in young women),
- with hot phases, probably triggered by inflammation, in particular in very young patients <18-years-old
- for which autoimmune mechanisms are being increasingly discussed.

Therefore, from a theoretical point of view, the question is whether it might be possible that those vaccinated who developed myocarditis after administration of an mRNA vaccine might be carriers of a pathogenic mutation of ARVC/ACM, and that ACM has been triggered by the vaccination. In this case, it could be reasonable to
- specifically look for major and minor criteria of ARVC/ACM in ECGs and MRIs in these patients,
- obtain a family history for arrhythmias and cases of sudden cardiac death in the family of these patients,
- offer genetic testing for ARVC/ACM if there are suspicious findings.

If you think that this is an approach worthy of further investigation and if you have any ideas how to proceed, please let us know. Would you advise ACM patients against vaccination with a mRNA vaccine in the future, since vector vaccines are now available in sufficient numbers and the EMA sees no causal relationship with myocarditis with COVID-19 Vaccine Janssen and Vaxzevria?

Comments and ideas are very welcome!

We would also like to probe the possibility of doing a study in this domain with support of the ERN. So, the question is: have you seen any cases in the recent months? Please let us know.

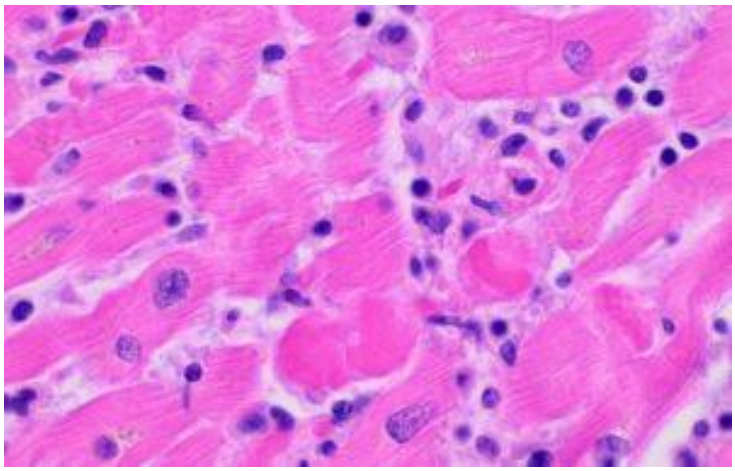
Ruth Biller, ePAG chair (info@arvc-selbsthilfe.org) and Nynke Hofman (n.hofman@amsterdamumc.nl)

References

Arrhythmogenic Right Ventricular Cardiomyopathy Presenting as Clinical Myocarditis in Women
https://doi.org/10.1016/j.amjcard.2020.12.090

'Hot phase' clinical presentation in arrhythmogenic cardiomyopathy
https://doi.org/10.1093/europace/euaa343

Evidence From Family Studies for Autoimmunity in Arrhythmogenic Right Ventricular Cardiomyopathy: Associations of Circulating Anti-Heart and Anti-Intercalated Disk Autoantibodies With Disease Severity and Family History
https://doi.org/10.1161/circulationaha.119.043931



EURORDIS Black Pearl Awards

The 11th edition of the Awards will take place online on TUESDAY, 8th FEBRUARY 2022 from 18:00 until 19:30 CET and will bring together persons living with a rare disease, patient advocates, policy makers, scientists, healthcare professionals, industry representatives, and more. The February event is an annual awards ceremony launching the month of Rare Disease Day. Since 2012, EURORDIS-Rare Diseases Europe has organised this event to recognise the major achievements and outstanding commitment of patient advocates, patient organisations, policymakers, scientists, companies, and media who strive to make a difference for the rare disease community.
It's nominations time again!
Don't miss your chance to nominate your star of the rare disease community, or enter yourself, for one of the EURORDIS Black Pearl Awards 2022.

The 12 award categories (follow the link below) recognise the outstanding efforts of individuals, organisations, companies, researchers, scientists, media, and policy makers in bringing about change to improve the lives of the 30 million people in Europe and 300 million worldwide living with a rare disease. Nominations can be submitted from anywhere in the world!

→ **CALL FOR NOMINATIONS**

Nominations deadline: 10th September 2021.
Any questions regarding the nomination process? Do not hesitate to contact Martina Bergna, EURORDIS Events Manager, at martina.bergna@eurordis.org. ♥

Latest ERN GUARD-Heart Publications

1.Asselbergs FW, Sammani A, Elliott P, Gimeno JR, Tavazzi L, Tendera M, Kaski JP, Maggioni AP, Rubis PP, Jurcut R, Heliö T, Calò L, Sinagra G, Zdravkovic M, Olivotto I, Kavoliūnienė A, Laroche C, Caforio ALP, Charron P; Cardiomyopathy & Myocarditis Registry Investigators Group. Differences between familial and sporadic dilated cardiomyopathy: ESC EORP Cardiomyopathy & Myocarditis registry. ESC Heart Fail. 2021;8:95-105. doi:10.1002/ehf2.13100.

2. Kaufman ES, Eckhardt LL, Ackerman MJ, Aziz PF, Behr ER, Cerrone M, Chung MK, Cutler MJ, Etheridge SP, Krahn AD, Lubitz SA, Perez MV, Priori SG, Roberts JD, Roden DM, Schulze-Bahr E, Schwartz PJ, Shimizu W, Shoemaker MB, Sy RW, Towbin JA, Viskin S, A M Wilde A, Zareba W. Management of Congenital Long-QT Syndrome: Commentary From the Experts. Circ Arrhythm Electrophysiol. 2021 Jul;14(7):e009726. doi: 10.1161

3. Garnier S, Harakalova M, Weiss S, Mokry M, Regitz-Zagrosek V, Hengstenberg C, Cappola TP, Isnard R, Arbustini E, Cook SA, van Setten J, Calis JJA, Hakonarson H, Morley MP, Stark K, Prasad SK, Li J, O'Regan DP, Grasso M, Müller-Nurasyid M, Meitinger T, Empana JP, Strauch K, Waldenberger M, Marguiles KB, Seidman CE, Kararigas G, Meder B, Haas J, Boutouyrie P, Lacolley P, Jouven X, Erdmann J, Blankenberg S, Wichter T, Ruppert V, Tavazzi L, Dubourg O, Roizes G, Dorent R, de Groote P, Fauchier L, Trochu JN, Aupetit JF, Bilinska ZT, Germain M, Völker U, Hemerich D, Raji I, Bacq-Daian D, Proust C, Remior P, Gomez-Bueno M, Lehnert K, Maas R, Olaso R, Saripella GV, Felix SB, McGinn S, Duboscq-Bidot L, van Mil A, Besse C, Fontaine V, Blanché H, Ader F, Keating B, Curjol A, Boland A, Komajda M, Cambien F, Deleuze JF, Dörr M, Asselbergs FW, Villard E, Trégouët DA, Charron P. Genome-wide association analysis in dilated cardiomyopathy reveals two new players in systolic heart failure on chromosome 3p25.1 and 22q11.23. Eur Heart J.2021;42:2000-2011. doi: 10.1093/eurheartj/ehab030.

4. Mizia-Stec K, Charron P, Gimeno Blanes JR, Elliott P, Kaski JP, Maggioni AP, Tavazzi L, Tendera M, Felix SB, Dominguez F, Ojrzynska N, Losi MA, Limongelli G, Barriales-Villa R, Seferovic PM, Biagini E, Wybraniec M, Laroche C, Caforio ALP; EORP Cardiomyopathy Registry Investigators. Current use of cardiac magnetic resonance in tertiary referral centres for the diagnosis of cardiomyopathy: the ESC EORP Cardiomyopathy/Myocarditis Registry. Eur Heart J Cardiovasc Imaging. 2021 Jun 22;22(7):781-789. doi: 10.1093/ehjci/jeaa329.

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