Monday 26 august 2024

ERN GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES
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ERN GUARD-HEART BIMONTHLY NEWSLETTER YEAR 2024 NUMBER 4

The launch of the ATS Registry!

Copenhagen, May 2024

The international ATS registry is now open. Andersen-Tawil Syndrome (ATS) is an extremely rare disease where the clinical presentation ranging from relatively benign morphologic abnormalities of the head, face, and extremities to periodic paralysis and life-threatening cardiac arrhythmia. ATS is transmitted in an autosomal dominant manner and associated with loss-of-function and dominant negative variants in the gene *KCNJ2* and has previously been classified as LQTS7. Due to the rarity of ATS and the heterogeneity of the clinical presentation, most studies are performed using small study population. This may result in significant delay of diagnosis as well as challenges in determining proper treatment options for ATS patients. At present, no definitive recommendations for ATS management are available.
Registry coordinator: Jacob-Tfelt-Hansen.
Registry managers: Priya Bhardwaj, MD, & Stine Bøttcher Jacobsen, PhD.
For more information, please contact the registry-managers by e-mail:
priya.kumar.bhardwaj@regionh.dk ,
stine.b.jacobsen@sund.ku.dk ♥

Agenda online board-meeting

Amsterdam, 26-08-2024

Friday 27 September the next ERN GUARD-Heart board meeting will be held online (9:00 – 13:00). The agenda will be send by e-mail next week. ♥

Conect4children.org

Amsterdam, 07-08-2024

Conect4children (c4c) Stichting (Foundation) is looking forward to celebrating the success of the C4C project at the 4c International Symposium, a dissemination and collaboration event organised by c4c (https://conect4children.org/). The goal of the event is to share the achievements and results of the project, including the start-up of the not for profit, c4c Stichting, that continues outside the c4c consortium. During the event, C4c will share their successes and ways in which future partners will be able to utilize the results of the project, either through future grant projects, scientific societies or pharmaceutical companies.

Please register your attendance to the event here.
Event details:
Dates: 13-14 November 2024
Venue: Wicked GroundsHQ, Generaal Vetterstraat 55, 1059 BT Amsterdam.

Over the past six years, academic and industry partners have worked tirelessly towards the c4c vision: better medicines for babies, children and young people through a pan-European clinical trial network. This event aims to share the project results, showcase its achievements and discuss future plans. It offers an excellent platform for collaboration and networking with key stakeholders in paediatric research and drug development. For questions, please contact: Barbara Reynolds (Richards), conect4children Foundation (Stichting). Email: barbara.reynolds@conect4children.eu ♥

Who are our ePAG's?
Inês Pinto Gonçalves - Portugal

Lisbon, 22-08-2024

Inês is the patient representative of "Associação Coragem" (Courage Association) – a patients' organization with a strong liaison with Hospital de Santa Cruz Pediatric Cardiology Department, whose head office is based at the hospital, and for more than a decade supports patients and families with congenital heart disease and contributes to raise awareness about this situation.

'I was in 2011, in the eighth month of pregnancy, when I received my son's first diagnosis and the only thing I knew, until then, about the subject, was that hearts beat on the left side of the chest.
The baby I thought would be perfect turned out to be full of holes. A atrioventricular septal defect, a single valve, a ventricular disproportion, a patent arterial duct, an aortic coarctation and even a pulmonary banding almost stopped my heart too.
In those weeks before my son was born, I only had enough time to ask questions, get the answers I needed, with the help of many doctors and exams, and without realizing how quickly time had gone by, he was born.
He was born in a private hospital where he stayed for a week to stabilize his pulmonary part. After that, he was transferred to the incredible Santa Cruz Hospital where he underwent surgery to close patent arterial duct and put a banding in the pulmonary artery. It was more than a month and a half before he went home.
Finding out my first child had congenital heart disease was a shock. Absolutely everything was new. The appointments, the medical terms, the exams, the medications, the wires, the machines, the sounds. No one prepares us for living in a hospital and having a child fighting for his life. Sometimes I had the feeling that if they gave me any more bad news they would need to give me a bed too.
Two difficult years followed. He needed second medical opinions, a catheterization, many appointments and exams so that, at 8kg and two years old, he underwent surgery again to solve the initial problem: atrioventricular septal defect, a single valve and a ventricular disproportion.
The way to solve it was not clear, so I took my son to the operating room that February 5, 2013 without knowing very well what would be possible to do with that little heart. It was the hardest 8 hours of my life but the difficult part really happened: the correction was completely done, the atrioventricular septal walls were reconstructed perfectly, two valves were built and the left ventricle regained its size.
The possibility of living with half a heart, undergoing more surgeries and having a short life expectancy had disappeared from the horizon.
It was great to see him able to walk and talk over the next few months. He gained a new life.
But, a year later, and realizing that he was not gaining height and growing, he had to undergo new tests and exams. This no longer had anything to do with the heart (I supposed), but new fears arrived. It was discovered that he has panhypopituitarism, a clinical syndrome of deficiency in the production of all pituitary hormones. He is currently medicated for growth hormone - with daily injection since he was 4 years old -, for hypothyroidism and for very low cortisol levels, the treatment for this last condition has a great influence on the functioning of the heart, as it increasing blood pressure.

It's true that his health has to be well managed, I don't know what could come next, he will forever be a cardiac patient and he will have medication for the rest of his life to suppress all hormonal failures and to prevent his heart from getting worse but, what I know today, despite everything he already had and still has, he lives a normal life like any teenager. This is the most important thing and I am grateful for this every day: he is alive and happy.' ♥

LMNA PRIORITY project awarded prestigious Leducq grant

Boston, July 2024

The PRIORITY project has received a grant of $8,000,000 from the Leducq Foundation. The grant was awarded to the consortium for research into dilated cardiomyopathy (LMNA-DCM). The project is called PRIORITY: Cardio-LaminoPathy: fRom pathomechanIsms tO peRsonalIzed TherapY.

LMNA-related dilated cardiomyopathy (LMNA-DCM) is a severe and inherited disease caused by mutations in the LMNA gene. This gene provides instructions for making proteins called lamins (Lamins A/C), which help maintain the structure of the cell nucleus and play an important role in regulating chromatin organization, gene expression, and cytoskeletal organization by interacting with numerous other proteins. There is no effective treatment for LMNA-DCM because we do not fully understand the disease's underlying mechanisms. Therefore, the proposed PRIORITY (cardio-laminoPathy: fRom pathomechanIsms tO peRsonalIzed TherapY) network aims to fill this knowledge gap and develop personalized, disease-modifying therapies.

Aims of PRIORITY

PRIORITY has three main Aims. Aim 1 investigates the clinical and genetic signs associated with LMNA-DCM, identifying factors to understand what factors affect the severity and progression of the disease. Aim 2 pursues the underlying disease mechanism(s) contributing to LMNA-related DCM, uncovering the complex cellular and molecular processes underlying the pathogenesis and disease progression using state-of-the-art technologies. Aim 3 develops mechanism-based therapies for LMNA-related DCM by leveraging existing concepts and novel findings from Aim 2, potentially offering personalized therapeutic options for LMNA-DCM patients.

Network

The team is composed of internationally renowned experts from prestigious European institutions (Sorbonne Université-Inserm, Amsterdam University, Maastricht University, University of Zurich, Paris-Cité University) and the USA (Duke, Cornell, and Stanford), along with early career investigators. We will partner with LMNAcardiac.org, a patient-led foundation, to ensure a patient-centered approach and to address the needs of those affected by LMNA-DCM. International collaborations and exchanges will offer unique interdisciplinary training opportunities for early career scientists in the network. Dr. Gisèle Bonne, Research Director Sorbonne Université-Inserm and Dr. Andrew Landstrom, Associate Professor Duke University School of Medicine, are the coordinators with the support of the Netherlands Heart Institute as coordinating office.

Do you want more information? https://www.fondationleducq.org/network/priority-cardio-laminopathy-from-pathomechanisms-to-peonalized-therapy/ or contact https://www.lmnacardiac.org/ ♥

LMNA Cardiac Diseases Network

Fondation Leducq

The launch of the FA registry!

Madrid, June 2024

We are contacting you from the Inherited Cardiac Diseases Unit at Hospital Universitario Puerta de Hierro (Madrid, Spain) to formally invite your center to participate in an international retrospective registry focused on the natural history of cardiac involvement in Friedreich's ataxia (FA). Although cardiac involvement is common and one of the leading causes of mortality in FA, the cardiac phenotype and prognosis of cardiac involvement in these patients are still not well defined. Our study aims to determine the natural history of cardiac involvement in patients with FA through the collaboration of European centers within ERN initiatives. Therefore, the member centres of ERN-RND are invited to participate as well in this registry. We would be honored and delighted if your center could participate in this registry.

If you agree to collaborate, we would like to collect anonymized data of patients with FA as soon as possible. All participating centers in the registry will have at least one author on the resulting publications. Additional authors will be assigned to centers contributing with a higher number of cases.

We have already received favorable approval from ERN_GUARD_HEAART and Hospital Universitario Puerta de Hierro ethics committee. Data will be collected through a REDCap database.

If you are interested in participating, please let us know, and we will send you the access credentials. We aim to collect data by the end of October, but please feel free to inform us if you need additional time.

Fernando Dominguez & Pablo Garcia Pavia
(fdominguezr@salud.madrid.org) ♥

Upcoming Webinar

Amsterdam, 26-08-2024

The monthly Webinar series will re-start, after the summer break on Wednesday 25 September, 17:00.
The flyer and link for subscription will be send by e-mail soon. ♥

Latest ERN GUARD-Heart Publication (s)

1. Svensson A, Jensen HK, Boonstra MJ, Tétreault-Langlois M, Dahlberg P, Bundgaard H, Christensen AH, Rylance RT, Svendsen JH, Cadrin-Tourigny J, Te Riele ASJM, Platonov PG. Natural Course of Electrocardiographic Features in Arrhythmogenic Right Ventricular Cardiomyopathy and Their Relation to Ventricular Arrhythmic Events. J Am Heart Assoc. 2024 Aug 20;13(16):e031893. doi: 10.1161/JAHA.123.031893. Epub 2024 Aug 19. PMID: 39158567.

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