Thursday 7 november 2024

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ERN GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart

ERN GUARD-HEART BIMONTHLY NEWSLETTER YEAR 2024 NUMBER 5

ERN GUARD-Heart Webinars

Amsterdam, 06-11-2024

The webinars of ERN GUARD-Heart are planned at the last Wednesday of the month from 17:00 – 18:00 PM. All the webinars were recorded and are uploaded for review on the website: https://guardheart.ern-net.eu/experts/webinar-series/ . Besides the regular webinars, we have a collaboration with the AEPC for a couple of joint webinars each year. This will be continued in 2025. The next webinar will be held 27-11-2024, at 17:00: 'Cardiomyopathies and Pregnancy' . The flyer will be shared seperately per e-mail as well, please forward this to your colleagues in the departments. This is also interesting for nurses and counselors. Everyone is welcome to register. ♥

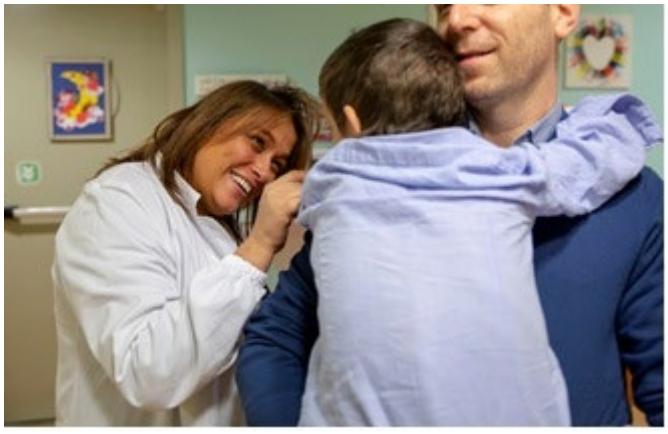
Who are our ePAG's?
Giovanna Campioni - AICCA

Milano, 31-10-2024

Giovanna is patient representative of AICCA (Associazione Italiana dei Cardiopatici Congeniti Adulti ONLUS), the Italian GUCH association.

'My name is Giovanna Martina Campioni and I was born in Frosinone, my father's hometown. My parents, living in Germany since they were little, wanted to give birth to their firstborn in Italy, and so it happened. They had, like all parents, a thousand plans for me, they already saw me as grown up, healthy, beautiful and strong. But their thousand desires were shattered right from my birth, I was strong nor healthy, and perhaps not even very beautiful, I was born black, cyanotic. They discovered after fifteen days that their daughter had a serious heart malformation. I was born with a complex congenital heart disease such as tricuspid atresia, dextrocardia in situs solitus, interruption of the inferior vena cava with azygos continuation, stenosis of the pulmonary valve, and malposition of the great vessels, in short, I was missing half a heart. I spent most of my first eleven years in hospital, sometimes even many months at a time and for this reason I never felt uncomfortable in hospital environments, the hospital was my home, the doctors and nurses were my family, my friends. At 5 years they decided to operate me for the first time. I remember the day I left intensive care, I remember my joy in believing that my rebellious little heart was finally healed. When I was 12 my parents decided to return to live in Italy, where the climate would be more favorable for me, according to German cardiologists. In Italy, unfortunately, only the heat was the positive note because for everything else it was very difficult to find a hospital that was able to treat me as I needed. My parents, like many parents, had to undertake journeys of hope throughout Italy and Europe with me. At 12 years oldI found myself catapulted into a small village in the province of Lecce, in south Italy.

Very beautiful, but very different from the place where I had grown up, despite multiple hospitalizations, my habits were completely different, I felt like a fish out of water. The middle school period ended and high school began, which gave me the opportunity to be myself and be considered equal to others, a place where I didn't have to defend myself. The hospitalizations continued, but my classmates and teachers never made me feel bad about my absence, instead giving me all the support I needed. Hospitalizations have always been frequent, and a second operation was necessary, but unfortunately my family and I had great difficulty finding a suitable cardiology center in Italy. After months of research I was operated on in Paris. I spent two very difficult months in hospital. Having surgery at 18 is different than having surgery at 5. You have a different awareness, you are aware that you could stop living. It was a challenging period, but I got through it, and this made me even more determined not to give up in the face of any obstacle.
At the age of 20 after long struggles, I received permission to go and study in Rome at the Faculty of Law.'



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The story of Giovanna Campioni

...Continued from previous page

''My parents I had found a center that dealt with congenital heart disease that I could turn to in case of need. As luck would have it, I would have used it more than I wanted, in fact I took most of my university exams get ready in hospital. Also during this period the hospital became my home and the staff my family.
At 23, a third operation was necessary, again in Paris. The script was the same as the previous time. My life was and is a continuous recovery, restarting, starting again. I had to learn it. Enjoy every moment as if it were your last, but keeping in mind that you have to do everything to ensure it isn't your last.
In those years the hospitalizations continued, but I could never imagine having to undergo even a fourth operation. Never more than in those days did I feel that I might not make it.
I woke up in intensive care.
I convinced myself that nothing could disturb me anymore, not even the strongest pain. I realized, and today I am certain, every intervention for me was a rebirth, every rebirth provided me with the tools to start again. It was precisely during these months of hospitalization that I met two wonderful doctors, Dr. Giamberti, the heart surgeon who operated on me, and Dr. Chessa, my cardiologist.
They were the ones who taught me that my illness was not a limitation, as some wanted me to believe, but a precious resource that I could use to do good for others.
Today I'm the general Director in the Aicca association - Italian Association of congenital heart patients in children and adults.
As a child I could never have imagined that my half heart and my diversity could one day allow me to do a job that I love above all else, but above all I could never have imagined that I could do so much, that I could, perhaps, live and not simply survive.
I'm different, I continue with hospitalizations, my pathology has worsened. I had to implant a defibrillator and have been on the heart transplant list for a year.
Despite this diversity and adherence to treatments, albeit few given the rarity of the pathology, I allow myself to be free to live as I like'.

LMNA-cardiac collaboration with Citizen Health

Amsterdam, 07-08-2024

LMNA Cardiac is now connected to Citizen (https://www.citizen.health/). Citizen builds technology solutions which enable patients with rare and complex diseases to access the best therapies and support, while continuing to help get much-needed drugs and new therapies to market faster. The field of the genetic cardiomyopathies was new. The shared goals are to build a natural history dataset that can be utilized by researchers working on life-altering therapies, while sharing insights to patients to help with day-to-day care. ♥

16th ERN GUARD-Heart board meeting

Amsterdam, 30-10-2024

The 16th board meeting of ERN GUARD-Heart was held online. Usually this meeting is combined with the ESC-meeting, but this year this was unfortunately not feasible due to the very expensive rates in London. The slides of the meeting and a short report have been shared recently. Next board meeting will be held **Friday 14 March 2025** in the Netherlands (in Amsterdam or close to Amsterdam). We will keep you informed. ♥

European Reference Networks

Joint Transnational Call by ERDERA

Amsterdam, 07-11-2024

We would like to inform you about the opportunity to apply to the upcoming 2025 Joint Transnational Call by ERDERA for proposals of research collaborative projects. It is focused on "**Pre-clinical therapy studies for rare diseases using small molecules and biologicals – development and validation**". The call is pre-announced (in draft -not legally binding) and expected to officially open on 10th Dec 2024: https://erdera.org/news/erdera-pre-announces-2025-joint-transnational-call-for-proposals-on-rare-disease-therapies/ .♥

Call for Proposals on Rare Disease Therapies

The European Rare Diseases Research Alliance (ERDERA) is excited to announce the upcoming launch of its Joint Transnational Call (JTC) for Proposals 2025. Set to officially open on December 10, 2024, this call will invite research teams from across Europe and beyond to submit collaborative projects focused on **"Pre-clinical therapy studies for rare diseases using small molecules and biologicals – development and validation"**.

This JTC will build upon the significant progress made by previous European Joint Programme on Rare Diseases (EJP RD) calls, with the aim of fostering international partnerships to develop innovative therapies for rare diseases. National and regional funding bodies from over 30 countries, including France, Belgium, Germany, Italy, and all Baltic states, have expressed interest in participating.

Aim of the Call

The primary objective is to support interdisciplinary, transnational collaborations that leverage complementary expertise to develop new therapies for rare diseases. Research projects should focus on pre-clinical studies involving small molecules or biologicals. This could include the development of novel therapies, biomarkers for therapy efficiency, or replication of pre-clinical studies to validate findings.

Please note that this is a draft announcement and the list of funders is not final.

Topics of Interest

Eligible proposals may include research on:

- Development of pre-clinical therapies using cell, organoid, or animal models.
- Biomarker identification that correlates with therapeutic efficacy.
- Pre-clinical proof-of-concept studies showing pharmacological activity and safety data.

Notably, gene therapies, rare neurodegenerative diseases like Alzheimer's, and rare cancers are excluded from this call. Projects should have a special focus on therapies that can translate into clinical applications.

Application Timeline

The JTC 2025 will follow a two-stage submission process:

- **December 10, 2024**: Call opens.
- **February 13, 2025**: Pre-proposal submission deadline.
- **July 9, 2025**: Full proposal submission deadline. Funding decisions are expected by December 2025.

Get Involved

Interested applicants can join an informational webinar on December 17, 2024, for guidance on preparing submissions. Additionally, partnerships with patient advocacy organizations (PAOs) and the inclusion of Early Career Researchers (ECRs) are highly encouraged to foster a collaborative and inclusive research environment.

For further details, please contact the Joint Call Secretariat at SelteneErkrankungen@dlr.de or the relevant national contact point.

Latest ERN GUARD-Heart Publication (s)

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