European Patient Advocacy Group

ePAG EpiCARE Newsletter
March 2024

It is our pleasure to announce that this year we will continue with our **newsletter** dedicated to patients' associations. It is our wish to **connect** and **engage more** with the wide community of rare and complex epilepsies and **provide better support** by disseminating relevant information.

If you wish to disseminate the information about your association, projects, activities and events you are planning, please contact: epag.epicare@gmail.com

AWARENESS DAYS

SCN2A Awareness Day

On February 24th we joined together to raise awareness for SCN2A-related disorders, a group of rare genetic conditions that impact individuals worldwide. Let's come together to shine a light on SCN2A-related disorders, support those affected, and advocate for greater understanding and resources.

Understanding SCN2A-Related Disorders

SCN2A-related disorders are caused by mutations in the SCN2A gene, which encodes a sodium channel critical for proper brain function. These disorders can present with a range of neurological and developmental challenges, including epilepsy, intellectual disability, autism spectrum disorder, and movement disorders.

Challenges Faced by Individuals and Families

Living with an SCN2A-related disorder presents numerous challenges for affected individuals and their families. Managing seizures, developmental delays, and navigating the healthcare system can be overwhelming. Families often seek support, information, and resources to help them on their journey.

The Importance of Awareness and Support

Raising awareness about SCN2A-related disorders is crucial for early diagnosis, access to appropriate care, and improved outcomes. By increasing understanding within our communities, we can ensure that those living with these conditions receive the support, resources, and empathy they need.

Advocating for Research and Resources

Advancements in research are essential for deepening our understanding of SCN2A-related disorders and developing targeted treatments. Advocacy plays a vital role in driving research forward, advocating for increased funding, and promoting collaboration among researchers, clinicians, and affected families.

Rare disease day

Living with epilepsy is challenging, but facing a rare form can present even more hurdles. Today we distinguish more that 130 rare and complex epilepsies!
Let's raise awareness, spread understanding, and support those who fight these lesser-known battles every day. Together, we can make a difference in their lives.
Have a look at our Awareness video here.

KCNQ2 Awareness week

KCNQ2 DEE is a severe form of epilepsy caused by mutations in the KCNQ2 gene. It manifests in early infancy with frequent seizures, developmental delays, and various neurological challenges.
Here are some key aspects of KCNQ2 DEE and the needs of patients and their families:

- ***Understanding the Condition***: KCNQ2 DEE presents a complex clinical picture, characterized by multiple seizure types. These seizures can be challenging to control with medications, leading to significant impacts on the quality of life for both patients and their families.
- ***Early Diagnosis and Intervention***: Early diagnosis is crucial for KCNQ2 DEE to ensure timely intervention and support. However, due to its rarity and the variability of symptoms, diagnosis can be delayed or missed. Increased awareness among healthcare professionals can lead to earlier identification and access to appropriate treatments and services.
- ***Holistic Care and Support***: Managing KCNQ2 DEE requires a multidisciplinary approach involving neurologists, geneticists, therapists, and other healthcare providers. Patients and families benefit from comprehensive care plans addressing not only seizure management but also developmental, cognitive, and behavioural needs. Support services such as counselling, respite care, and community resources play a vital role in enhancing the well-being of individuals and families affected by KCNQ2 DEE.
- ***Advocacy and Awareness***: Raising awareness about KCNQ2 DEE is crucial for fostering understanding, support, and research efforts. By amplifying the voices of patients, families, and advocacy organizations, we can advocate for improved access to care, research funding, and policies that prioritize the needs of rare disease communities.

GRI related disorders Awareness month

Welcome to GRI Awareness Month! Throughout March, we're dedicated to illuminating the importance of GRI genes, delving into their key aspects, and addressing the needs of families impacted by these genetic variations.
Understanding GRI Genes:
GRI genes, including GRIA, GRIN, and GRIK, play pivotal roles in cognitive function and neurological development. These genes encode for receptors and ion channels that are integral to synaptic transmission and plasticity, influencing learning, memory, and behaviour.
Key Aspects:
Neurological Phenotypes: Variations in GRI genes have been associated with a diverse array of neurological phenotypes, ranging from cognitive impairments and developmental delays to epilepsy and neuropsychiatric disorders such as autism spectrum disorder (ASD) and schizophrenia.
Impact on Neurodevelopment: Proper functioning of GRI genes is essential for normal brain development and function. Disruptions in GRI signalling pathways can perturb synaptic plasticity, neural connectivity, and ultimately, cognitive abilities.
Therapeutic Implications: Understanding the genetic basis of neurological conditions associated with GRI genes is crucial for the development of targeted therapies and interventions. Advances in pharmacogenomics and precision medicine hold promise for personalized treatment approaches tailored to individuals' genetic profiles.

Family Needs:
Genetic Counselling and Testing: Families benefit from access to genetic counselling services to understand the implications of GRI gene variations for themselves and their children.
Early Intervention and Support Services: Early identification of GRI-related neurological conditions enables prompt intervention and access to specialized support services. Early childhood intervention programs, behavioural therapies, and educational accommodations can optimize developmental outcomes for affected individuals.
Community Resources and Advocacy: Connecting with support networks and advocacy organizations empowers families to navigate the challenges associated with GRI gene variations. These resources provide a platform for sharing experiences, accessing information, and advocating for research funding and policy changes.
Research Participation: Engaging in research initiatives focused on GRI genes is instrumental in advancing scientific understanding and developing novel therapies. Participation in research studies, clinical trials, and registries contributes to the collective effort to unravel the complexities of genetic neurodevelopmental disorders.
As we observe GRI Awareness Month, let us unite in raising awareness, fostering support, and advocating for the needs of individuals and families impacted by GRI gene variations. Together, we can illuminate the path towards greater genetic intelligence and neurological well-being.

Publications

- Amin S, Møller RS, Aledo-Serrano A, et al. Providing quality care for people with CDKL5 deficiency disorder: A European expert panel opinion on the patient journey. Epilepsia Open. Published online March 7, 2024. doi:10.1002/epi4.12914
- Ribeiro-Constante R, Tristán-Noguero A, Martínez Calvo FF, et al. Developmental outcome of electroencephalographic findings in SYNGAP1 encephalopathy. Front. Cell Dev. Biol. Published online March 5, 2024. Doi:10.3389/fcell.2024.1321282

Events

First online meeting with patient organisation for rare and complex epilepsies in Europe

ePAG EpiCARE is happy to announce the 1st online meeting with patient associations for rare and complex epilepsies in Europe!

Online meeting will be held on May 31st from 6-8 pm CET.

Stay tuned as agenda and meeting links will be shred soon!

For more information, please epag.epicare@gmail.com

Syngap1 Family Race, Madrid, Spain

The first SYNGAP-1 solidarity race is a 5km and 10km race through the Montse del Pilar in Madrid. The donations colleted from the race will go entirely to the research of this syndrome, which is a rare disease caused by a mutation in the Syngap1 gen.

For more information, here you have the https://www.syngap1.es/

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