European Patient Advocacy Group

ePAG EpiCARE Newsletter
September 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

Lissencephaly Awareness Day - September 8th

On September 8th, we observed Lissencephaly Awareness Day, a day dedicated to raising awareness about this rare neurological condition. Lissencephaly, which translates to "smooth brain," is a disorder that affects brain development, leading to the absence of normal folds and grooves on the brain's surface.

Children and individuals diagnosed with Lissencephaly often face significant challenges, including developmental delays, seizures, difficulties with muscle tone and movement, as well as problems with feeding and breathing. While this condition is rare, it profoundly impacts the lives of those affected and their families.

Increased awareness and understanding can lead to earlier diagnoses, more comprehensive medical care, and the advancement of research that could improve the quality of life for those affected. While Lissencephaly presents unique challenges, it also serves as a reminder of the resilience and courage of the children, individuals, and families who live with it every day.

By joining together, we can help make a difference. Let's continue to advocate for research, show our support for those affected, and spread the word to raise awareness.

Hypothalamic Hamartoma Awareness Day - September 15th

On September 15th, we recognized Hypothalamic Hamartoma (HH) Awareness Day, a day dedicated to raising awareness about this rare brain condition that can cause significant neurological and developmental challenges.

A hypothalamic hamartoma is a non-cancerous growth located in the hypothalamus, a region deep within the brain that plays a key role in regulating

critical functions such as body temperature, hunger, emotions, and sleep. While benign, HH can lead to a variety of serious symptoms, including:

- Gelastic seizures (uncontrollable laughing seizures)
- Cognitive and developmental delays
- Behavioural problems
- Hormonal imbalances

Many individuals with HH experience epilepsy that is resistant to standard treatments, making diagnosis and management especially challenging. As it is a rare condition, awareness is often limited, leading to delays in diagnosis and treatment.

Greater awareness can help improve early detection, lead to better treatment options, and reduce the social and emotional impact on families affected by this rare disorder.

We encourage you to learn more about Hypothalamic Hamartoma, support ongoing research, and spread the word about this important cause. Your support can make a lasting difference in the lives of those affected.

For additional information, visit https://epi-care.eu/patient-and-caregiver-leaflets/

STXBP1-Related Disorders Awareness Month - September

September marks STXBP1-Related Disorders Awareness Month, a time dedicated to raising awareness about this rare genetic condition that can cause a range of neurological challenges. STXBP1-related disorders are caused by mutations in the STXBP1 gene, which plays a crucial role in brain function, specifically in the communication between neurons.

Children and individuals affected by STXBP1-related disorders often experience a variety of symptoms, including:

- Developmental delays and intellectual disabilities
- Epilepsy, with many experiencing drug-resistant seizures
- Movement disorders such as dystonia or ataxia
- Behavioural challenges including autism spectrum disorder

Because STXBP1-related disorders are rare, many people face delayed diagnoses or misdiagnoses, which can make managing symptoms even more difficult. Currently, there is no cure, but ongoing research is vital in the search for effective treatments and support.

Increasing awareness can lead to earlier diagnoses, better treatment plans, and improved quality of life for those living with STXBP1-related disorders. Advocacy and community support are essential in driving research forward and creating a future where individuals with STXBP1 mutations can thrive.

For more information on how to support STXBP1 research, spread awareness, or connect with others in the community, visit https://epi-care.eu/patient-and-caregiver-leaflets/

EVENTS

Together for rare and complex epilepsies Follow up meeting

Thank you for joining the last meeting Together for rare and complex epilepsies that was held on August 30th!
We are excited to announce the **hybrid meeting** for patient representatives of different rare and complex epilepsies! Workshops will be held **in Barcelona, Spain from May 9th to 10th 2025**.
We are organising an additional online meeting (in October/November) to provide updates for the ongoing activities including creation of a Complex and Rare Epilepsies Alliance (CREA)!
If you wish to participate or need more information, please contact: epag.epicare@gmail.com

Different faces of rare and complex epilepsies

It is our pleasure to announce the second conference titled DIFFERENT FACES OF RARE AND COMPLEX EPILEPSIES, organised by **Dravet syndrome Croatia**, that will be held in **Split**, Croatia, at hotel Amphora, from T**hursday 17th to Saturday 19th of October 2024**. The conference aims to encourage knowledge sharing and productive discussions on rare and complex epilepsies, and it will be held in person and online. On 18th of October, panel discussions will be held for healthcare professionals in English, and on 19th of October panel discussion will be held in Croatian.
For full program and registration, visit: https://dravetconference2024.conventuscredo.hr/en/.

6th European GRIN Conference

It is with great pleasure that we inform you that the **on-line Registration for our 6th European GRIN conference is open**. To facilitate registration, we have created a website with all the information you will need to stay informed about the event. We will be updating the website with more information as we complete the program over the next few weeks.

We sincerely hope you can join us from **Friday November 8th until Sunday November 10th in Barcelona**. Please share this information with other family members and/or your local associations/family groups. We hope that many of you will be able to participate in the conference again this year in person; but for those who cannot travel to Barcelona, we are offering, once again, online registration for the Saturday and Sunday events.

The conference will start on Friday November 8th at 18h at the **Abba Garden Hotel** with the “Meet and Greet” social event and poster session, from 19-21:30h, in the *Cervantes* Room. During this social event we hope families, researchers and specialist will interact, grab a bite to eat and learn more about GRIN/GRIA research projects and the fantastic job some associations and family groups are doing in order to advance the knowledge about GRI disorders.

On Saturday November 9th we will have a full day of talks by 11 distinguished speakers working on GRIN and GRIA disorders. They will share with us their latest work and great advances in the field. This event will be held in the PCCB auditorium at the Sant Joan de Deu Hospital, from 9h-18h. This event will be followed by “Dinner with the Speaker” event at the Abba Garden Hotel.

On Sunday November 10th we will have a moderated Discussion about complex diagnosis and treatment of GRIN and GRIA symptoms. In this discussion round, we aim at presenting some of the challenges families encounter when trying to have their GRI- kids diagnosed and treated. We hope with this discussion to create awareness and engage the medical and research communities to help us find better treatment options for some of these complex problems.

We have an exciting program ahead and are sure you will enjoy it. Please join us in-person in Barcelona for the 6thedition of the European GRIN conference.

We are very much looking forward to seeing you in November.

The Board of GRIN Europe

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