European Patient Advocacy Group

ePAG EpiCARE Newsletter
September 2023

It is our pleasure to announce that this year we will start with a 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

STXBP1 Awareness month

In the month of September, we come together to shed light on a group of rare and often misunderstood disorders - STXBP1 related disorders. These conditions affect individuals from all walks of life, but awareness is still limited. It's time to change that!
STXBP1 related disorders are rare of genetic conditions caused by mutations in the STXBP1 gene. They can lead to a wide range of symptoms, including developmental delays, epilepsy, movement disorders, and intellectual disabilities.
Many affected individuals and their families face daily challenges, yet their strength and resilience are nothing short of inspiring.
By raising awareness, we can encourage research, support families, promote early diagnosis and advocate for inclusivity. Join us this September to spread awareness about STXBP1 related disorders. Whether you're sharing information or hosting fundraising events, your support can make a significant difference in the lives of those affected by these disorders. Together, we can create a world where STXBP1 related disorders are better understood, diagnosed, and ultimately, where individuals affected by them can thrive.

Lissencephaly awareness day

LISSENCEPHALY Awareness

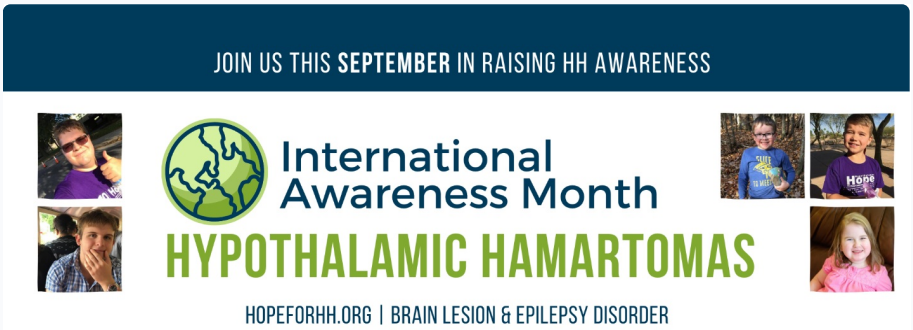
On September 8th, we came together to shine a light on Lissencephaly, a rare and challenging brain malformation. Lissencephaly, which means "smooth brain", is a condition where the brain's surface lacks the normal folds and grooves, leading to various developmental and health challenges for affected individuals and their families.
This Awareness Day is a reminder that even in the face of extraordinary challenges, there is strength, love, and hope.
By raising awareness, we aim to: educate as many people may not have heard of lissencephaly, support families who face daily hurdles, promote research and advocate for inclusion. This Lissencephaly Awareness Day, let's unite in solidarity. Whether you're wearing a ribbon, sharing information, or participating in events, your efforts make a difference. Together, we can provide hope, support, and a brighter future for those affected by Lissencephaly.

Hypothalamic Hamartoma syndrome Awareness Day

On September 15th, we shine a light on Hypothalamic Hamartoma Syndrome (HH), a rare neurological condition that affects individuals and their families in profound ways. HH may be uncommon, but its impact on those affected is significant.
This Awareness Day is an opportunity to:

1. Raise Awareness: Many people have never heard of HH, and its symptoms can often be misdiagnosed. By spreading awareness, we can help individuals get the proper diagnosis and support they need.
2. Support Families: Families dealing with HH face unique challenges. Let's show our support by offering a helping hand, sharing resources, and lending an empathetic ear.
3. Advocate for Research: Awareness drives research and innovation. Encouraging more research into HH can lead to better treatments and improved quality of life for those affected.
4. Promote Understanding: Individuals with HH have unique abilities and potential. Let's advocate for understanding and inclusion, celebrating their strengths and accomplishments.

Join us on September 15th to stand in solidarity with the HH community. Whether you're wearing purple, sharing information, or participating in awareness events, your efforts can make a real difference. Together, we can provide hope, support, and a brighter future for those affected by Hypothalamic Hamartoma Syndrome.
To learn more about HH visit the EpiCARE website where you can find the leaflets for healthcare professionals, patients and caregivers as well as the Patient Journey!



ePAGs EpiCARE news

ePAGs at IEC 2023 in Dublin

ePAG members, chair Isabella Brambilla and Gregori Cabanach, have attended the IEC in Dublin!
With a lot of networking, and following sessions, they have presented an online poster about Patient Journey, titled "Patient Journeys: A Tool to Raise Awareness on the Evolution, Common Needs and Critical Issues of Patients with Rare and Complex Epilepsies".

Among different activities, they have also attended the IBE breakfast session, a round table where the focus was on rare and complex epilepsies! .

New leaflets and Patient Journeys

With the ongoing Awareness month, wo couldn't have picked a better time for the leaflets and Patient Journey for STXBP1 disorders to be published! We thank the whole medical team with Dr. Ganna Balagura who took the initiative for devoting their time in preparing these documents as well as to the patient organisations from France, Germany, Israel, Italy, Spain and USA!

You can find the leaflet here and the Patient Journey here.

Translations are progressing and we are happy to announce that the first translation in Greek is available for Ring 20 chromosome syndrome!
If you wish to disseminate information about your disease, share the knowledge and raise awareness in your own national language, contact us at: epag.epicare@gmail.com

Upcoming events

Gentle reminder for the 5th GRIN Europe conference

5th GRIN Europe conference, 7-8 October 2023, organized by our GRIN Europe association, formerly known as GRIN2B Europe is starting soon!
Barcelona is the chosen location for the 2023 conference given its easy access from many parts of Europe. The conference will be held both in person and online, and online participation is free of charge!

For this conference there are 13 confirmed speakers from different parts of the world and from different areas of interest in GRIN disorders. Their presentations will be followed by a round-table event where collaborative work and future research discussions will be encouraged.
This conference is without doubt the most important scientific event in Europe for GRIN-related research and is essential to build a support network for families living with GRIN children.
For more information visit GRIN Europe brand new website.

Dravet Syndrome UK Parent/Carer Conference

Friday 17th November
In-person at the Leonardo Royal Hotel, London City OR live-stream online
Dravet Syndrome is a complex epilepsy. As well as seizures, those affected have learning disability (severe to profound in over 75%), autism, challenging behaviour and difficulties with speech, mobility, feeding and sleeping.
The DSUK Parent/Carer Conference is a unique opportunity for parent/carers, who require practical support and information for all aspects of living with this devastating condition to meet with others who are going through the exact same experiences. It also offers a unique opportunity to meet and to hear directly from expert guest speakers from the UK and around the world. From learning about family rights, to discovering insights about the latest research and treatments; from asking Dravet experts your burning questions to sharing experiences with other Dravet families - our conference provides a friendly environment for furthering knowledge and understanding about living with Dravet Syndrome.
You can view the full agenda and register to attend by visiting the Dravet website.

Dravet Syndrome UK Professionals Conference

Saturday 18th November
In-person at the Leonardo Royal Hotel, London City OR live-stream online
The medical and scientific understanding of Dravet Syndrome has increased significantly during recent years. We've entered an incredibly exciting and hopeful time for the future of Dravet Syndrome, with developments in gene therapy and other treatments that seek to address not only the symptoms of the condition but also its underlying causes.
We're pleased to invite you to the DSUK Conference Professionals Day - the only scientific meeting in the UK that is dedicated to furthering knowledge about Dravet Syndrome. The conference, chaired by Professor Helen Cross OBE, delivers a packed agenda of leading voices in Dravet Syndrome to enable discussion of the latest updates, news and medical research.
The DSUK Conference Professional Day is CPD accredited by the RCP and the RCPCH.
Further information and registrations.

Save the date for European Dravet Syndrome Conference 2024

Fundación Síndrome de Dravet is organising a Dravet syndrome conference in 2024.
Save 14th and 15th of March 2024 in your calendars and join the conference in Madrid.
For preliminary program and registration, click here.

Meet the patients' associations

Dup15q France

Dup15q France is a nonprofit French association that gathers families affected with this condition. "15q duplication syndrome" or "Dup15q" for short, is a rare genetic disorder caused by a duplication on chromosome 15 in the region q11.2 to 13.1.
When this duplication is mentioned, it can be found by various names: isodicentric chromosome 15 (idic 15), pseudo isodicentric 15 (psu idic 15), partial trisomy 15, tetrasomy 15, supernumerary marker chromosome 15 (CMS15), inverted duplication 15 (inv dup 15) and interstitial triplication 15 or interstitial duplication 15 (int dup 15). The dup15q syndrome is a rare genetic disorder, with an estimated prevalence of around 1/15,000. In most cases, duplication of chromosome 15q11.2-13.1 is not inherited, but occurs spontaneously (de novo) during cell division at the start of embryonic development.

The association work on various projects to achieve their missions:
- Bring families together in a spirit of solidarity and mutual support
- Support parents at the time of diagnosis and guide them towards appropriate medical and paramedical care for their child
- Explain Dup15q syndrome, which is not well known in France at present
- Provide documentation in French, translating the most important documents if necessary
- Bring together families affected by Dup15q syndrome for the annual "Family Walk".
- Lead the Dup15q France association's various discussion forums
- Support genetic research on "15q", in liaison with the French associations "Prader-Willi France" and the "Association française du Syndrome d'Angelman" (AFSA)
- Cooperate with the various international associations representing Dup15q syndrome.

Website: https://www.dup15qfrance.fr/page/502718-bienvenue
Email: info@dup15qfrance.fr
Facebook

Dravet syndrome UK

Dravet Syndrome UK (DSUK) is an independent charity dedicated to improving the lives of those affected by Dravet Syndrome through support, education and medical research by:

supporting families affected by Dravet Syndrome emotionally, practically and financially, raising awareness and understanding of Dravet Syndrome among medical professionals and funding medical research to increase understanding of Dravet Syndrome, improve its management, and work towards better outcomes.

For more information about the charity and their activities:
website: https://www.dravet.org.uk/
Email: info@dravet.org.uk
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Hope for Hypothalamic Hamartoma

Hope for Hypothalamic Hamartomas (Hope for HH) is a volunteer-based nonprofit organization founded by parents of children with hypothalamic hamartomas (HH) with a goal to create a single, credible source for information about the diagnosis, treatment, and support of individuals with HH.

Every family touched by this rare disorder has a unique and often heart-breaking story of how they attained a correct diagnosis. Obtaining a correct diagnosis can take months and even years and often involves incorrect diagnoses. Once a diagnosis is confirmed, many families struggle with choosing an appropriate course of treatment, if one is even available. Regardless of treatment, managing the daily lives of HH patients and dealing with the long term and frequently devastating effects of HH requires ongoing information and support.
Hope for Hypothalamic Hamartomas drives education, support and research in this complex syndrome. They focus their efforts and funding on early detection, improved treatments, and managing HH as a complex medical syndrome with the goal of one day finding a cure.

This month of September is HH awareness month, and September 15th is International Hypothalamic Hamartomas Awareness Day and we are encouraging people to donate and to share photos of themselves in HH shirts on our social media! HH shirts can be found at: Hope for Hypothalamic Hamartomas | Official Merchandise | Bonfire

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