



ePAG EpiCARE Newsletter May 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

Tuberous sclerosis complex awareness day

Tuberous sclerosis complex (TSC) is a genetic disorder that affects approximately 1 in 6,000 individuals. TSC causes the growth of non-cancerous tumors in various organs, including the brain, heart, kidneys, and skin.

One of the most common and challenging symptoms of TSC is seizures. Approximately 85% of individuals with TSC experience seizures at some point in their life, and for many, seizures are the first sign of the disorder. Seizures in TSC can range from mild to severe, and they can be difficult to control with medication.

In addition to seizures, TSC can cause a wide range of other symptoms, including developmental delays, intellectual disabilities, behavioral problems, skin abnormalities, and kidney disease. The severity of symptoms can vary widely between individuals with TSC, even within the same family.

Despite its prevalence, many people have never heard of TSC. May 15th is TSC Awareness Day, a day to raise awareness about this rare disorder and the challenges faced by individuals and families affected by it.

On TSC Awareness Day, many organizations and physicians came together to raise awareness about this rare disorder and support those who are affected by it!



Meet our ePAGs

Carol-Anne Partridge is co-founder of [CDKL5 UK](https://www.cdkl5uk.org/) and former director of the International Foundation for CDKL5 Research. Aside from CDKL5 UK she represents the CDKL5 associations in Europe by being the member of ePAG EpiCARE– a group which is working to bring awareness and standardised care, among other things, for people living with rare and complex epilepsy in Europe. She is a qualified children's social worker and works full time in South West England.

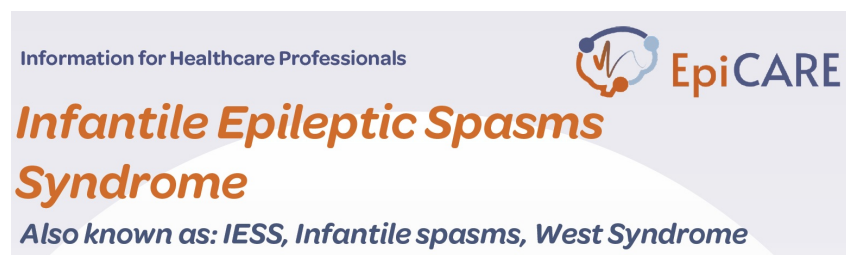


Carol-Anne is passionate about supporting families, raising awareness and challenging assumptions. Have a look on a video to meet our ePAG, her view on the roles of ePAGs and benefits this work brings to the patient community!

[Carol-Anne Partridge presentation video](#)

News

New leaflet



New leaflet for healthcare professionals and patients and caregivers is dedicated to Infantile Epileptic Spasms Syndrome. [View and download the leaflet.](#)

We thank UK Infantile Spasms Trust for devoting their time in the preparation of these documents!

New translations

New translations are also ready and published on our webpage. Newly added are the translations in Italian and Croatian!

If you are interested in translating the leaflet for your condition, please contact epag.epicare@gmail.com

Upcoming events



2nd SCN2A and SCN8A Conference

The 2nd SCN2A and SCN8A Conference will be held on 26th to 27th of May in Køge, Denmark.

[Full program and registration details](#)

Inaugural family and scientific conference on CHD2-related disorders

The conference, organized by Coalition to Cure CHD2, will be held on June 2nd - 4th, 2023 at Disney's Coronado Springs Resort in Lake Buena Vista, Florida!

[More information about the conference and registration.](#)



GNAO1 European conference

GNAO1 European conference, organized by the association Famiglie GNAO1, will be held on June 16th-17th in Rome, Italy!

[Program and registration](#)



Meeting of SYNGAP1 families by Famiglie Syngap1 Italia

The first meeting of the Syngap1 families will take place in Rome on 20th and 21st of June 2023.

The APS FAMIGLIE SYNGAP1 ITALIA association is building an event for all Italian families to create a moment of discussion and sharing, to get to know each other and to support and advise us.

June 21st is the date chosen internationally to raise awareness for this rare genetic anomaly.

[More information](#)

5th GRIN Europe conference

It is our great pleasure to inform you about the upcoming 5th GRIN Europe conference, 7-8 October 2023, organized by our GRIN Europe association, formerly known as GRIN2B Europe. We have picked once again Barcelona as the location for the 2023 conference given its easy access from many parts of Europe.

For this conference we have 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 we encourage collaborative work and future research discussions. Our 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.

[Meet the patients' associations](#)

Famiglie GNAO



Main aims/objectives:

- Spread the knowledge of the disease and organizing international conferences entirely dedicated to the GNAO1 gene mutation.
- Support the newly diagnosed families.
-
- Facilitate the creation of an international network of clinicians and researchers involved in GNAO1, in order to foster knowledge sharing and potential collaborations.
- Promote fundraising to economically support research projects as well as projects that aim to improve the daily life of the GNAO1 patients.
- Implement inclusion projects to improve the quality of life of the patients and their families.

Contacts and social media:

Website: www.gnao1.it

Email: info@gnao1.it

Facebook: <https://www.facebook.com/FamiglieGNAO1>

GLUT1 Deficiency UK

Glut1 Deficiency UK is a non-profit family led charity dedicated to improving the lives of those in the Glut1 Deficiency community through its mission of:

- Increasing awareness of Glut1 Deficiency
- Improved education of families and health professional in relation to Glut1 Deficiency
- Advocacy for families and patients impacted by Glut1 Deficiency
- Support and funding for Glut1 Deficiency research
- Most important or highlighted project:
- Find someone with Glut1
- Pushing for early diagnosis



Contacts and social media

Email: Hello@glut1deficiency.org.uk

Website: www.glut1deficiency.org.uk

Facebook: @Glut1DeficiencyUK

Linkedin: glut1deficiencyuk

Instagram: glut1deficiencyuk

Twitter: @Glut1UK

Apoyo Drave



Apoyo Dravet is a community of people affected by rare epilepsies and Dravet syndrome. It acts as a collaborative platform that connects patients and family members with volunteers and other contributors (including scientists, doctors, psychologists, educators, technicians and others). It promotes scientific research, medicine, technology and innovation as a part of social intervention seeking to improve the quality of life of patients, their families and their social circles.

Most important or highlighted project :

- Scientific Research
- Social Innovation
- Patient-centric Technology Department

Apoyo Dravet es una comunidad de afectados por enfermedades raras con epilepsia y síndrome de Dravet. Se constituye en una plataforma colaborativa que aglutina pacientes y familiares, voluntarios y colaboradores (científicos, médicos, psicólogos, educadores, tecnólogos y otros intervinientes). La entidad promueve la investigación científica, la medicina, la tecnología y la innovación en la intervención social en aras de mejorar la calidad de vida de pacientes y de sus entornos familiares y sociales.

Proyectos más importantes:

- Investigación Científica

- Innovación social
- Tecnología orientada al afectado

Contacts and social media
E-mail: contacto@apoyodravet.eu
Telephone: + 34 687 27 38 74
Web: <https://www.apoyodravet.eu/>
Facebook: ApoyoDravet
Instagram: @apoyodravet
Twitter: @apoyodravet
LinkedIn: ApoyoDravet

Follow us on social media!



[Unsubscribe](#)

© 2021 EpiCARE All rights reserved.
You are receiving this email as you may work at one of the EpiCARE centres or you are taking part in one or more of EpiCARE expert groups or you may have attended one of our webinars.

[See in browser](#)