# ePAG EpiCARE Newsletter
# July 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

### CDKL5 Deficiency Disorder Awareness Day

June 17th marks CDKL5 Awareness Day, a special occasion dedicated to shedding light on CDKL5 Deficiency Disorder (CDD). On this day, we have united to raise awareness about this rare genetic condition that affects thousands of children worldwide.
CDKL5 Deficiency Disorder is a neurodevelopmental disorder caused by mutations in the CDKL5 gene. It predominantly affects girls, causing severe developmental delays, epilepsy, motor impairment, and a range of other neurological challenges. Living with CDD can be incredibly challenging for both the affected individuals and their families.
By spreading awareness, we can encourage earlier diagnoses, promote understanding among healthcare professionals, and drive advancements in therapeutic interventions. Together, we can make a difference in the lives of those living with CDKL5 Deficiency Disorder.
Together, let's create a world where every individual with CDKL5 Deficiency Disorder receives the care, understanding, and opportunities they deserve. Together, we can bring hope to the CDKL5 community and make a lasting impact.

### SYNGAP1 Developmental and epileptic encephalopathy Awareness Day

On June 21st, we came together to raise awareness for SYNGAP1 Developmental and Epileptic Encephalopathy, a rare and debilitating genetic disorder that affects individuals around the world. SYNGAP1 is a gene responsible for producing a protein critical for brain development and function. When mutations occur in this gene, it can lead to SYNGAP1 Developmental and Epileptic Encephalopathy, a complex condition characterized by developmental delays, intellectual disability, epilepsy, and various neurological challenges. SYNGAP1 Awareness Day provides an opportunity to shine a light on this rare disorder and extend support to the affected individuals and their families. It's a day to educate others about the impact of SYNGAP1 and advocate for improved understanding, research, and resources.
By increasing awareness, we strive to accelerate research efforts, promote early diagnosis, and facilitate access to appropriate treatments and interventions. Together, we can make a difference in the lives of those living with SYNGAP1 Developmental and Epileptic Encephalopathy.
On this day many SYNGAP1 association gathered together in creating this amazing video

### Dravet syndrome Awareness Day


On June 23rd, we have united to raise awareness for Dravet Syndrome, a rare and severe form of epilepsy that profoundly impacts the lives of those affected and their families.
Dravet Syndrome, also known as Severe Myoclonic Epilepsy of Infancy (SMEI), is a genetic disorder characterized by early-onset seizures that are often prolonged and difficult to control. It typically emerges within the first year of life, causing developmental delays, cognitive impairment, and various neurological challenges.
Dravet Syndrome Awareness Day serves as a powerful reminder of the importance of understanding and supporting individuals living with this complex condition. It is a day dedicated to sharing knowledge, fostering compassion, and advocating for improved treatments and resources.
33 associations world wide joined forces together to raise awareness about Dravet syndrome!

### Sturge Weber syndrome Awareness Day


On June 27th, we have joined together to raise awareness for Sturge-Weber Syndrome (SWS), a rare and complex neurological disorder affecting individuals worldwide. Sturge-Weber Syndrome is characterized by a facial birthmark, known as a port-wine stain, along with neurological abnormalities and eye complications. It can lead to various challenges, including epilepsy, developmental delays, glaucoma, and cognitive impairments.
Sturge-Weber Syndrome Awareness Day on June 27th provides an opportunity to educate others about this condition and show support for individuals and families living with SWS. It's a day to share stories, promote understanding, and advocate for improved resources, research, and access to care.

### GLUT1 Deficiency Syndrome Awareness Day


GLUT1 Deficiency Syndrome is caused by a mutation in the SLC2A1 gene, leading to impaired glucose transport in the brain. This results in a range of neurological symptoms, including epilepsy, movement disorders, developmental delays, and cognitive impairments. By raising awareness, we can help ensure early diagnosis, appropriate management strategies, and enhanced quality of life for those affected by GLUT1 Deficiency Syndrome.
Show your support for GLUT1 Deficiency Syndrome Awareness Day. Together, let's shine a light on this condition, promote understanding, and offer hope to the GLUT1 community. By working together, we can make a difference and improve the lives of those living with GLUT1 Deficiency Syndrome.
Let's unite our voices, raise awareness, and strive for a future where individuals with GLUT1 Deficiency Syndrome receive the care, understanding, and opportunities they deserve. Together, we can make a positive impact in the lives of those affected by this rare neurological disorder.

## ePAGs EpiCARE news

### Cross-ERN ePAG collaboration

Our ePAG is joining forces with ePAGs from ERN ITHACA in a new transversal group Epi-ID!
Understanding the common difficulties, we share as a patient community has opened the opportunity to collaborate on identifying fundamental needs of patients affected with epilepsy and associated neurological disorders and disabilities! Looking forward to working with ITHACA ePAGs!

## Meet our ePAGs

Rosaria Vavassori is the founder of the italian association for Alternating Hemiplegia of chidlhood currently involved in the International consortium for the research on alternating hemiplegia of childhood and AHC18+ e.V. as she has an adult son affected by this rare and complex condition. She is also an ePAG member and Data Manager of the IAHCRC Consortium, coordinating the IAHCRC-CLOUD Project for the development and management of the Platform. 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!

Rosaria Vavassori presentation video

## Upcoming events

### International Epilepsy Congress

During the IEC in Dublin, International Bureau for Epilepsy is organising a Breakfast round table: Rare and Complex Epilepsies with the participation of members of ePAG EpiCARE! Save 5th of September at 8 am on your calendar and join this session in Dublin!
While at IEC, don't forget to join us at the poster session, where ePAGs will be presenting Patient Journeys!

### 2nd German-speaking SCN2A conference and family reunion

We are happy to announce our 2nd German-speaking SCN2A Conference. From the 1st to the 3rd of September 2023, SCN2A Germany e.V. and Dr. Walid Fazeli (UKB) cordially invite all German-speaking families to Bonn for the second SCN2A Family Conference.
Interesting presentations on SCN2A-related epilepsy and autism spectrum disorders, a short report of the European SCN2A/SCN8A conference, insights into the topic of cortical visual impairment, an update on the Natural History Study, and more will await the visitors. On Saturday, family activities are on the program. This day is intended to give those affected by SCN2A the opportunity to get to know each other and to exchange experiences and ideas.
SCN2A Germany e.V. is looking forward to welcoming all affected and interested people to the conference. For further information about this event please visit: https://www.scn2a.de/scn2a-konferenz-im-september-2023-in-bonn or write an Email to info@scn2a.de.

### 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. Barcelona is the chosen location for the 2023 conference given its easy access from many parts of Europe.
For this conference there are 13 confirmed speakers from different parts of the world

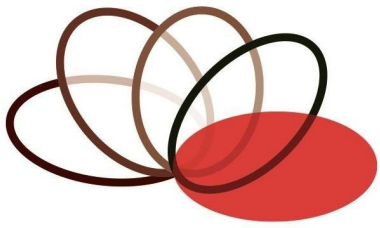
and from different areas of interest in GRIN disorders. Their presentations will be followed by a round-table event where collaborative work and future research discussions will be encouraged. This conference is without doubt the most important scientific event in Europe for GRIN-related research and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

## Meet the patients' associations

Nederlandse vereniging voor mensen met een Wijnvlek of Sturge-Weber syndroom (Dutch association for people with a Port-wine stain or Sturge-Weber syndrome)

Main Aims & objectives of the NvWSWs:

- Member/patient contact
- Supply of information
- Advocacy
- To support each other with awareness, dealing with and accepting the consequences of the syndrome.
- The association may provide members with information about developments/treatments and gives members a platform to exchange experiences.
- Giving information/education about the SW syndrome.
- Encouraging development of knowledge about the SW-syndrome.
- Promoting mutual solidarity.
- Co-operating with other associations.
- Directing members to other institutes.
- Giving telephone advice.



Projects:

- Encouraging the development of a medical specialist/expert centre for adult SWS patients (at present, a medical expert centre is available at the ERASMUS MC/SOPHIA at Rotterdam/The Netherlands for children/youngsters up to the age of 18).
- Buddies project (Maatjes project) whereby, for example parents of SWS patients give advice and support to other parents.

Activities:

- May 2023 NvWSWs day when the NvWSWs association reports to its members and guest speakers are invited.
- September 2023 Childrens day where young patients can meet.
- Quarterly newsletter
- Webinars

Website: http://www.wijnvlek-sturgeweber.nl/
Email: info@wijnvlek-sturgeweber.nl
Phone number: +31 332471466
Facebook

Associazione Italiana Glut1

Main aims/objectives:

- Offering support to families with GLUT1 deficiency syndrome.
- Our strategic plan is focused on 4 main areas:
- Support and incentive for scientific research (from diagnosis to possible treatment)
- Advocacy activities
- Awareness activities
- Support for families to improve the quality of life

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Highlighted projects:

- Continued implementation of the National Disease Registry
- Management and continuous development of "Ketonet", the app for managing ketogenic diet
- Support for newly diagnosed and non-diagnosed families (psychological, bureaucratic/operational, training, material,…)
- Providing families (patients, parents, siblings) with a path of psychological support free of charge
- Family, researchers' and clinicians' meetings
- Financing 3 research projects through a grant, managed applying the peer review methodology
- Dissemination on the disease (through institutional channels, social networks and relations with national and international patient networks and institutions)
- Advocacy activities with institutions, especially the Ministry of Health
- National and international collaborations with Patients' and Scientists' Networks.

For more information visit their website and have a look on a beautiful family video

Contacts and social media
Website: http://www.glut1.it/
Email: famiglie@glut1.it
Facebook

SYNGAP Elternhilfe e.V

The patient organization SYNGAP Elternhilfe e.V. from Krefeld in Germany with currently sixty members was founded in 2016 and is fully registered as non-profit organization since 2021. The association supports families with children suffering from Syngap1 syndrome. In Germany currently 108 Syngap patients are known.

The main work of Syngap Elternhilfe covers the following three topics: awareness, patient support (Whatsapp group) and research. Thereby the engagement goes beyond the Syngap1-syndrome and where necessary, joint projects are made with other associations for rare diseases to achieve a better outreach.

Highlighted projects:

- PATRE - PATient based phenotyping and evaluation of Therapy for Rare Epilepsies
  The starting point of our research activities was the observation of a mother that the epileptic convulsion in Syngap1 can be triggered by chewing food.This observation was firstly described in detail in the following publication.
  This observation led to the idea to collect information from parents of patients affected by Syngap1 Syndrome. Therefore, the PATRE project (PATient based phenotyping and evaluation of therapy for Rare Epilepsies) was started in collaboration with our research team from Paracelsus Medizinische Privatuniversität Salzburg (Austria). The most important questions and observations of our Syngap families were collected in a data base. The team wants to answer these questions by collecting the observations and making them available to researchers for scientific work on Syngap1. One of the results is this recently published presentation, answering the question: What are the best medications?
- HORIZON Europe – EURAS - EUropean network for neurodevelopmental RASopathies
  Initiated by Syngap Elternhilfe e. V., the EURAS project aims to gain a deeper understanding of the underlying disease mechanisms, create a framework for improved diagnosis, and develop effective treatments for neurodevelopmental RASopathies. Focus is specially on Cardio-facio-cutaneous syndrome (CFC), Costello syndrome (CS), and SYNGAP1-related encephalopathy (SYNGAP1. Noonan syndrome is additionally included for the creation of a European patient register. The patient register will be based on the work and experience made with PATRE. For this there will be important patient involvement of European associations under the lead of the Syngap Elternhilfe e.V. More information.
- RARE DISEASES RUN – RUN FOR RARE
  The Rare Diseases Run was initiated by Syngap Elternhilfe e.V for the first time

in 2022 and shall raise awareness for people affected by rare diseases. The run takes place in the week of the rare diseases day and will be organized as a virtual run. The run is organized together with nineteen other associations with the goal to increase the outreach of this project. Important contribution in articles, radio interviews, and social media posts have been generated, drawing attention to rare diseases. In 2023 about three thousand runners participated in the run and more than 32.000 Euro in donations were collected. This made the event the biggest run for rare diseases in Europe. The preparations for the Rare Diseases Run 2024 already started.

- Inclusion Box
  For educational purposes Syngap Elternhilfe e.V. together with other associations for rare diseases have created the Inclusion Box for kindergartens and schools in Germany, Austria and Switzerland. The Inclusion Boxes are equipped by the participating associations with illustrative and information materials (books, assistive equipment, inclusive toys, etc.) and can be borrowed by interested schools. The aim is to teach children about disabilities and rare diseases and to provide an opportunity to put themselves in the shoes of affected people. This should help to create more empathy and tolerance to disabled people.

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