EpiCARE JUNE 2022 NEWSLETTER

The goal of our newsletter is to keep you informed about actions related to the European Reference Network on rare and complex epilepsies, EpiCARE.

You will find information about our next events, educational and research activities and calls, but also news from the coordination team, our different members and our ePAG community.

NEXT WEBINARS

Learn more every 2nd and 4th Thursday of the month with EpiCARE!

Epilepsy after traumatic brain injury

July 7th at 17:00 CET

Asla Pitkanen
&
Jakob Christensen

Register

Sturge-Weber syndrome

July 21st at 17:00 CET

Martin Tisdall
&
Anne Comi

Register

On the webinar wepage, you can check out the agenda for the next webinars and subscribe to the webinars mailing to get a reminder a few days before each webinar.

Webinars webpage

LATEST NEWS

Glut1 leaflet now available on our website!

The EpiCARE ePAGs, Monica Lucente and Prof. Valentina De Giorgis redacted a new leaflet on Glut1 deficiency syndrome.

As for the other leaflets, it contains information on this rare epilepsy for both patients (or caregivers) and healthcare professionals.

You can find it on this webpage, along with the other leaflets, patient's journeys and resources for rare and complex epilepsies.

Supporting Ukraine : HUB for Rare Diseases

Rare Diseases
HUB Ukraine

To better support the rare disease ukrainian community, a new helpdesk was created by the Hospital Sant Joan de Déu in Barcelona, Spain : Rare Diseases HUB Ukraine.

The Hub concept was developed in response to the need to help Ukrainians affected by war, who need clinical and / or logistical support in the field of rare diseases. The Hub is designed to facilitate this by being fast moving; structurally flexible; reliable; visible; and able to leverage available resources.

The Hub entry team will assess the severity, urgency and need for logistical support of each request. Patient journeys and action protocols covering recurrent requests will be generated. The Hub core will include clinical professionals and admin support, while the Response Team will include organisation hunters.

The Hub contact details are provided:
+34 93 600 61 11
www.rarediseaseshub4ua.org
rarediseaseshub4ua@sid.es

EVENTS

European Conference on Rare Diseases and Orphan products

We will participate at the ECRD again this year, and proposed an abstract on the European Collaboration on Epilepsy Trials (ECET).

As per our patient representatives, on June 29th at 16:30 – 18:00 CET, Isabella Brambilla will be speaking about the inequalities in accessing health and social care for people living with rare diseases, and Allison Watson will give insight in inequalities in accessing education, work and independent living for people living with rare diseases!

Our ePAG members, Isabella, Rosaria and Monica together with Rare Epilepsy Italia, prepared poster for ECRD! Rosaria also submitted IAHCRC-CLOUD Platform: A data sharing infrastructure serving the multicentre studies of the International Research Consortium on Alternating Hemiplegia of Childhood.

More information about the ECRD here.

ILAE EUROPE epilepsy congress

14th European Epilepsy Congress
9-13 July 2022 | Geneva, Switzerland

As always, we are glad to collaborate with the ILAE and will actively participate in this 14th European Epilepsy Congress!

EpiCARE will hold a session, present a poster AND have a booth, so come meet us!

More information about the event here.

Upcoming events:

- 14-18 July: XVI Workshop on Neurobiology of Epilepsy – Early onset epilepsies : neurobiology and novel therapeutic strategies. Satellite event of the 14th ILAE EEC.
- 16-23 July: 5th Dianalund Summer school on EEG and Epilepsy. More info

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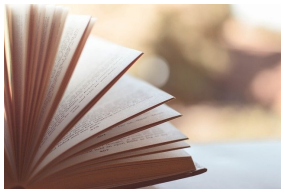
EpiCARE SCIENTIFIC PUBLICATIONS

- McTague, A., Brunklaus, A., Barcia, G., Varadkar, S., Zuberi, S. M., Chatron, N., Parrini, E., Mei, D., Nabbout, R., & Lesca, G. (2022).
  Defining causal variants in rare epilepsies: An essential team effort between biomedical scientists, geneticists and epileptologists.
  European journal of medical genetics, 104531. Advance online publication.
  Find the publication here

- Cardenal-Muñoz, E., Auvin, S., Villanueva, V., Cross, J. H., Zuberi, S. M., Lagae, L., & Aibar, J. Á. (2022).
  Guidance on Dravet syndrome from infant to adult care: Road map for treatment planning in Europe.
  Epilepsia open, 7(1), 11–26.
  Free PMC article

You can consult recent publications on rare and complex epilepsies, epilepsy surgery and other topics



on our scientific publications webpage.

EpiCARE members: please remember to mention EpiCARE in your affiliations or in the acknowledgements / references when appropriate.

A word from our ePAGs

CDKL5 Deficiency Disorder awareness day

CDKL5 deficiency disorder (CDD) is a rare genetic condition which is characterised by early onset epilepsy and children experience a range of difficulties including profound and multiple learning disabilities. The gene responsible for CDD was discovered in 2004. The CDKL5 gene is located on the X chromosome therefore it affects girls four times more than boys. With the onset of epilepsy gene panel testing,

more children are being diagnosed at an early age to the point where CDD is thought to be one of the most common forms of Genetic epilepsy, believed to affect 1 - 42,000 – 1 - 60,000. June is CDKL5 Awareness month, with June 17th being World CDKL5 Day. The community comes together every year, to fundraise and raise awareness, and there is also a yearly international family conference bringing together the medical, scientific and family communities together to share research updates and tips on living with the disorder. If you want to learn more about CDKL5 then check out CDKL5 Alliance website

SYNGAP1 awareness day

June is also dedicated to SYNGAP1 developmental and epileptic encephalopathy, with June 21st marked as an awareness day!

SYNGAP1-related non-syndromic intellectual disability is a rare genetic disorder caused by a variant on the SYNGAP1 gene. It is considered to be a spectrum disorder as patients are not affected in the same way or with the same severity. Unfortunately, it is not known what impacts the symptoms or their severity. Many of the most seen symptoms like intellectual disability, hypotonia, development delay, epilepsy (often refractory), sensory processing disorder, gross and fine motor skills delays, dyspraxia, speech delay, autism spectrum disorder, sleep and behaviour disorder, and visual abnormalities, might not be present in all of the patients.

Because of the complex symptoms and its presence, it is extremely important to accelerate the process of diagnosis, establish multidisciplinary health care team, and improve the quality of life of patients and their family!

Dravet syndrome awareness day
23rd of June people all over the word raised awareness about Dravet syndrome! Dravet Syndrome is a rare genetic encephalopathy characterized by a drug resistant epilepsy which appears in first year of life in previously healthy children. As first symptoms develop, they can quickly exacerbate, so patients can have multiple seizure types multiple times per day! Epilepsy is a severe issue for patients and their family, but unfortunately it is not the only one. Burden of the disease goes way further, as it causes cognitive and motor impairment, language disorder, behaviour and autistic traits, sleep and eating disorders, and skeletal deformity.
This seriously affects the quality of life of patients, but also of a whole family. Due to that it is highly important that the professional and non-professional community understand the needs of patients and do their best to provide them the best possible care with a multidisciplinary team, and involve them more in community's day-to-day activities. Although Dravet syndrome can be horrific, especially knowing the high risk of SUDEP, there are still moments, days, months that are overwhelmed with pure love and joy. Those moments fill our hearts and give us strength to push forward, until we finally reach the ultimate goal – find cure for Dravet syndrome!

ANYTHING TO SHARE?

Any news you would like to share, events planned, publications in the works?

Share with us any information you would like us to include in the next newsletter at a.robert@epi-care.eu

Follow us on social media !

Our mailing address is:

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