EpiCARE SUMMER 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!

Seizure burden and neurodevelopmental outcome in neonates with hypoxic-ischemic encephalopathy

September 8th at 17:00 CET

Geraldine Boylan
&
Maria Roberta Cilio

Register

Intracranial vascular malformations and epilepsy

September 22nd at 17:00 CET

Jana Dominguez Caral
&
Daniel Natera De Benito

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

Publication on ERN support to Ukraine

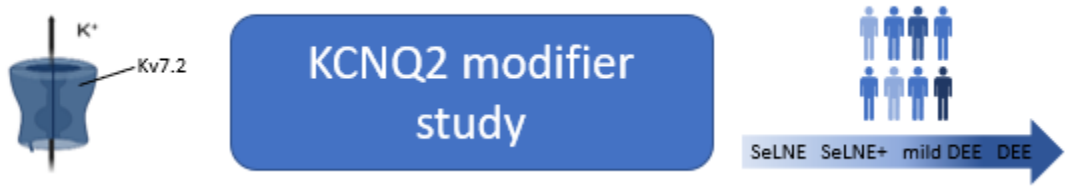
European Reference Networks

#ERNcare4Ua
Rare Diseases Doctors

A paper on the actions taken by the European Reference Networks to support the ukrainian rare disease community during the Ukraine crisis has just been published!

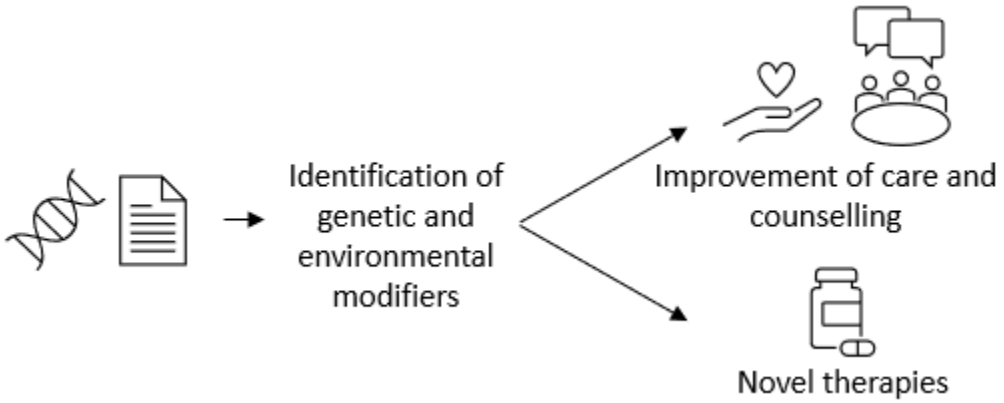
Read the publication

Identification of genetic and environmental modifiers in KCNQ2-related disorders.



Do you follow patients with KCNQ2-related disorders? Then consider including patients in our **KCNQ2 modifier study**. Despite relatively good genotype-phenotype correlations, phenotypic variability does exist and complicates counseling for individual patients. This study aims to **determine genetic and environmental (e.g., anti-seizure medication) modifiers to enhance the possibility of making an accurate prognosis for individual patients**. Insights in genetic modifiers could further help to identify additional disease pathways that can be targeted to improve outcome in the general KCNQ2 population.

Detailed clinical information and DNA of every patient is mandatory to be included in the study. Please contact Sarah Weckhuysen or Charissa Millevert for more information.



EVENTS

14th International Epilepsy Colloquium - Sept 14-16 in Lausanne

LATE CALL FOR ABSTRACTS

A late call for abstracts is scheduled for August 12-19, 2022. Do not miss the chance to be part of this cutting-edge program!

REGISTRATION

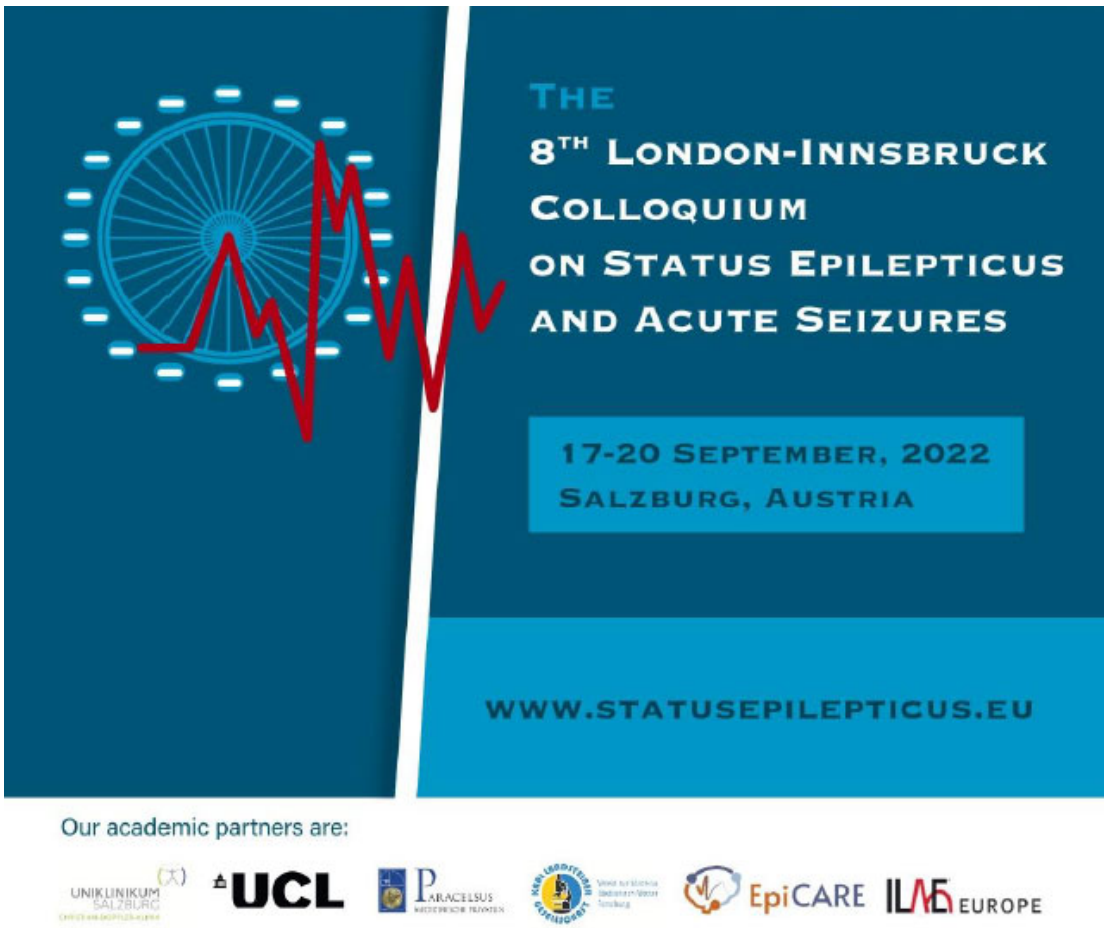
A **special reduction rate** will be applied to all those working at ERN EpiCARE centers (full, affiliated or collaborating partners). Registration should be concomitant to the Late-breaking abstract submission.

EpiCARE Medical Doctors (onsite participation and access to all activities): 380€
EpiCARE Medical Doctors (online participation): 150€
EpiCARE Students and Fellows (onsite participation and access to all activities): 260€
EpiCARE Students and Fellows (online participation): 110€
EpiCARE Neuropsychologists and Nurses (onsite participation and access to all

activities): 140€
EpiCARE Neuropsychologists and Nurses (online participation): 60€

Register here

Congress website



European Epilepsy Congress - 9-13 July, Geneva, Switzerland

We are glad to inform you about the 8th London-Innsbruck Colloquium on Status Epilepticus and Acute Seizures. These meetings have been held every two years since 2007, and the series has become a landmark meeting in the international epilepsy calendar.

Great news! The organizers have decided to grant every EpiCARE member a **reduced rate** for the colloquium. I.e. early registration rate until the congress. To benefit from this rate, you can register by sending a mail to status@cmi.at.

See the programme here

Congress website

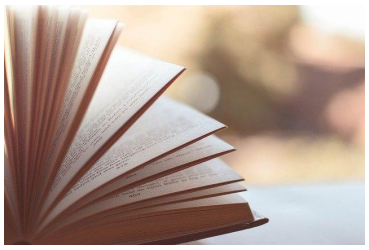
Upcoming events:

- 8-11 Sept: International Summer school for neuropathology and epilepsy surgery @

Erlangen University Hospital. More info
- 23-25 Sept: **Ring20** Families Conference in Cambridge. More info
- 19-21 Oct: conference on **Alternating Hemiplegia of Childhood and ATP1A3** diseases in Edinburgh. More info and registrations.

EpiCARE SCIENTIFIC PUBLICATIONS

- Lesca, G., Baumgartner, T., Monin, P., De Dominicis, A., Kunz, W. S., & Specchio, N. (2022). Genetic causes of rare and common epilepsies: What should the epileptologist know?. European journal of medical genetics, 65(9), 104570. Advance online publication. Find the publication here.



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

European Epilepsy Congress

It was great to meet again in person at the European Epilepsy Congress in Geneva.
Great topics were covered in the different sessions, including priorities on transition, new drugs and the SUDEP topic. It was very important to also be able to integrate the patient's point of view in the different sessions. Our chair, Isabella Brambilla had an opportunity to provide a presentation within EpiCARE's session! Thanks to the whole EpiCARE team for the fantastic collaboration.

Pictured here: Isabella Brambilla, ePAG Chair and Alexis Arzimanoglou, EpiCARE Coordinator.

GLUT1 Awareness day

This past July 10th, the second annual International Glut1 Awareness Day was celebrated. Patients, clinicians, families, caregivers, and friends came together to promote awareness of the syndrome on social media and in their communities, in hopes that knowledge can lead to support and inclusion for all those affected by Glut1DS. This year's special focus was on awareness within the medical community, in hopes of promoting early diagnosis and early treatment, which have an immense impact on outcomes particularly in Glut1DS. Glut1 Deficiency is a rare genetic disorder that impairs brain metabolism, caused by mutations in the SLC2A1 gene.The most common symptoms are seizures, cognitive issues, developmental delays, speech and language disorders, low muscle tone, eye/head movements and unusual body movements. Other symptoms that may occur: low stamina and energy, hemiplegia, ADD, memory problems, and sleep disturbances. Currently, there is no cure for Glut1 Deficiency, but the standard of care treatment is ketogenic dietary therapy. Educate yourself, because what counts most is to be aware. For more information, reach out to your local patient organization.

Our ePAGs joined ECRD on June 29th at 16:30 – 18:00 CET, with Isabella Brambilla and Allison Watson speaking about the inequalities in accessing health and social care for people living with rare diseases, and 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! Check out the poster from Rare Epilepsy Alliance Italy and IAHCRC.

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

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Follow us on social media !

Our mailing address is:

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