EpiCARE MAY 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!

Stroke and epilepsy

June 9th at 17:00 CET

Matthias Koepp
&
Johan Zelano

Register

Epilepsy surgery for polymicrogyria

June 23rd at 17:00 CET

Georgia Ramantani
&
Louis Maillard

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

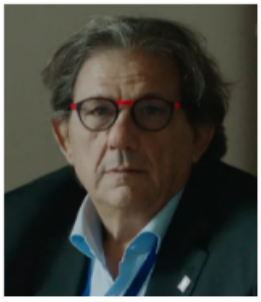
LATEST NEWS

European Epilepsy Education Award 2022 goes to Prof. Arzimanoglou

European Epilepsy Education Award

The award is given in recognition of outstanding contributions to the advancement of epilepsy education in Europe. The award consists of a diploma and carries no monetary value. It is announced in conjunction with the European Congress on Epileptology to which the awardee will have the registration fee waived. Awards are made to one individual every two years.

2022



Alexis Arzimanoglou
France

Public launch of the VARIA-ATP1A3 Study

IAHCRC International Consortium
research and care for the ATP1A3 diseases

IAHCRC STUDY VARIA-ATP1A3

PRESENTATION OF THE STUDY VARIA-ATP1A3

LOOKING INTO THE RARE VARIANTS OF THE MOST COMMON GENETIC CAUSE OF ALTERNATING HEMIPLEGIA IN CHILDHOOD

Study Coordinator
Katharina Vezyroglou, University College of London – Great Ormond Street Children Hospital, UK

Are you looking after a patient with a variant in the gene ATP1A3? Please consider joining the VARIA-ATP1A3 study!

ATP1A3 is associated with a broad neurological disorder spectrum. Out of 168 ATP1A3 variants published so far, 2/3 of published patients carry only 8 (c.2401G>A, c.2443G>A, c.2839G>A/C, c.2452G>A, c.1838C>T, c.2273T>G, c.2267G>A and c.2267G>T), for which there is clear genotype/phenotype correlation, as they are each associated with a specific phenotype (AHC/ RDP/ CAPOS/ RECA). We call these common variants. It remains unknown whether genotype/phenotype correlation exists for the other (rare) variants, as for most only 1-2 patients have been described. This might be due to rarity or patients are not recognized as having an ATP1A3-related condition.
Within the International AHC Research Consortium, an online survey of the phenotypic characteristics of patients carrying rare ATP1A3 variants has started to:

- establish the width of the ATP1A3-related disease spectrum;
- establish if genotype/phenotype correlation exists for rare ATP1A3
- establish a registry of patients with rare variants.

We would love to invite collaborators across the EPICARE network to join us in this effort and I am very happy to be contacted with any questions.
Coordinating clinician: Dr Aikaterini Vezyroglou
Institution (dept, hospital, city, country): Neurology, Great Ormond Street Hospital, London, UK
Contact email: k.vezyroglou@ucl.ac.uk

A **study on the VARIA-ATP1A3** has publicly launched last month, about the phenotypes correlated to rare variants in ATP1A3, directed by Dr. Aikaterini Vezyroglou (University College of London, UK). The description of the study is available here.

All research centers interested to participate are kindly invitated to contact Dr. Vezyroglou. Please, spread the news and the invitation to participate in this new great study on the ATP1A3-related diseases, for a better diagnosis and prognosis for all affected patients! Thank you!!

Supporting Doctors, Patients and Families from Ukraine.

The webpage on the EpiCARE website is regularly updated to provide useful informations and resources for people affected by epilepsies in Ukraine.
Have a look and share this link as needed:

https://epi-care.eu/epicare-support-to-ukraine/

A word from our ePAGs

EPNS Congress
ePAG Chair Isabella Brambilla attended the EPNS congress held in Glasgow, and presented the following topic: **Importance and expectations of early diagnosis** for the patients and their relatives, in which she emphasised **EpiCARE's commitment to early diagnosis** not only for Dravet Syndrome, but for all rare and complex epilepsies. **Epilepsy can be more than just seizures** and its **effects on cognition, behaviour and emotion can be profound**. Autistic features among other disorders can severly influence the quality of life of patients affected!

IMPORTANCE AND EXPECTATIONS OF EARLY DIAGNOSIS FOR THE PATIENTS AND THEIR RELATIVES

Epilepsy and **Developmental Epileptic Encephalopathies** are a **health priority** both because of the **complexity of managing the disease** and because of the **associated social burden** and **indirect costs** mainly due to social and work restrictions of patients and caregivers.

Complex and rare epilepsies represent a real health emergency and require special consideration for several reasons. Many of these forms
- have an unknown pathogenesis
- are responsible for disabling comorbidities,
- require a high medical standard for appropriate diagnosis and treatment.

EpiCARE

For all these reasons, several national and European projects have been developed to strengthen and promote interactions and expertise in this field and **to improve the standard of care and treatment of people with epilepsy.**
The European Community has included epilepsy among the diseases benefiting from a **European Reference Network - EpiCARE** to provide high quality and cost-effective care evenly distributed and available throughout Europe.

The diagnosis of epilepsy must be early and accurate, as well as drug or surgical treatment. The choice of therapy should also be adapted to the age and gender of the patient, and should also take into account the frequent presence of possible comorbidities.

15th of May was the Tuberous Sclerosis Complex (TSC) awareness day!

TSC Global Awareness Day
May 15

TSC is a genetic disorder caused by a mutation on TSC1 or TSC2 gene, affecting over 1 million people worldwide.
It causes tumor formation in all the vital organs, and epilepsy is present in almost 90% of patients with infantile spasms being a common seizure type in the infancy. Besides epilepsy, many develop autism spectre disorder including intellectual difficulties. As a systematic disease it affects many different organ systems such as akin, kidney and heart along with previously mentioned epilepsy. Quality of life of patients and their family is severely affected due to the high condition burden. Hence there is an emerging need in multidisciplinary approach to individuals with TSC in order to achieve the best possible outcome!

EVENTS

Lots of interesting events coming up!

European Academy of Neurology - 25-28 June, Vienna, Austria

Following the signature of a Memorandum of Understanding with the European Academy of Neurology, we are glad to participate in their great annual congress in Vienna !
Come meet us at booth number X35, the whole duration of the congress!
More information about the event here.

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

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:

- 27 June - 1st July: 11th European Conference on Rare Diseases & Orphan Products (ECRD 2022). More info
- 14-18 July: XVI Workshop on Neurobiology of Epilepsy – Early onset epilepsies : neurobiology and novel therapeutic strategies. Satellite event of the 14th ILAE EEC.

EpiCARE SCIENTIFIC PUBLICATIONS

Heers, M., Böttcher, S., Kalina, A., Katletz, S., Altenmüller, D. M., Baroumand, A. G.,

Strobbe, G., van Mierlo, P., von Oertzen, T. J., Marusic, P., Schulze-Bonhage, A., Beniczky, S., & Dümpelmann, M. (2022).

Detection of interictal epileptiform discharges in an extended scalp EEG array and high-density EEG-A prospective multicenter study. Epilepsia, 10.1111/epi.17246. 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.

CASE DISCUSSIONS

EpiCARE Case Discussions are scheduled every 2nd, 3rd and 4th Wednesday of the month from 4:00pm to 5:30pm CET.

Both **surgical and non-surgical cases** are discussed by panels of experts, full or affiliated members of EpiCARE. See the agenda on our website.

To receive the link to join the case discussions, you must register. Registration is only required once to be placed on the mailing list. Follow this link to register.

Please note that Pr. Ingmar Blümcke is also organizing monthly online **neuropathology review** meeting of interesting and/or challenging epilepsy surgery cases.
This Zoom link will work for every meeting on **every second Thursday per month** in 2022.

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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