EpiCARE

SEPTEMBER 2023 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

Our webinars on t rare and complex epilepsies take place every 2nd and 4th Thursday of the month, and the replays are always available on our website.

Thursday October 5th
17h CET

Use of EEG in clinical trials

Sandor Beniczky
&
Caroline Neuray

Register

Thursday October 19th
17h CET

Metabolic neonatal seizures and the diagnostic contribution of neonatal cerebral MRI

Maarten Lequin & K. Koop

Register

You can check the programme of the upcoming webinars and register to the webinar mailing on the EpiCARE webinars webpage:

Webinars webpage

LATEST NEWS

Look back on the IEC 2023

The congress was, as usual, a great opportunity to connect with colleagues, update each other on new guidelines and discuss hot topics, while highlighting the work done in the past 2 years. We were also glad to observe that the rare epilepsy sessions were very well attended, it seems the topic is raising interest!

Patient representatives were also included in this event, and we were glad to catch up with our ePAG members Isabella Brambilla and Gregori Cabanach. They presented a poster, and used this opportunity to connect with the IBE, and other guests!

ACT Workshop

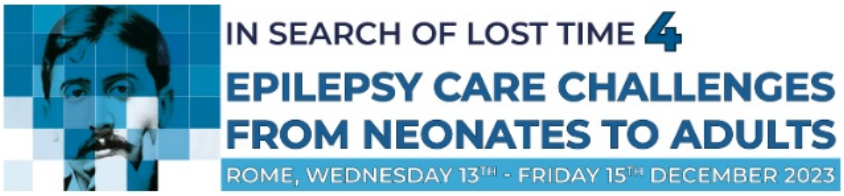
EJP RD Advisory Committee for Therapeutics (ACT) workshop (ERN-focused event)

Nicola openshaw-Lawrence (management team), Julia Pitsch (EpiCARE member Bonn, Germany) and Carol-Anne Partridge (EpAG) attended in person the EJP RD Networking Support Scheme: Advisory Committee for Therapeutics (ACT) workshop, which was held in Newcastle at the beginning of September to introduce the Act Model to ERNs.

A great deal of information was shared by the TREAT-NMD Advisory Committee for Therapeutics (TACT), who have been successfully operational for over 10 years, and this will be shared within ERN EpiCARE to discuss how this model can be adopted for use.

EVENTS

Register now for the next Rome workshop!



It is our pleasure to invite you to save the date of the 4th edition of the "In search of lost time" workshop, endorsed by the EpiCARE, taking place in Rome, Italy from Wednesday December 13th in the afternoon to Friday December 15th lunchtime.

This year the workshop will be on "Epilepsy care challenges from neonates to adults",

a contribution to the activities of the ERN EpiCARE to translate into actions in Europe the WHO Directive on "Epilepsy and other neurological disorders".

The workshop will discuss in detail two important areas of epilepsy care:

- Neonatal seizures and epilepsies
- Epileptic Seizures in brain disorders

Registration & more information

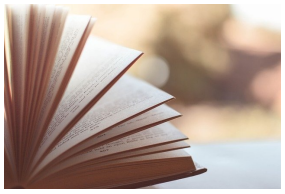
- Oct. 3-7: 5th Bologna EPIPED-EEG Course: EEG Interpretation in Pediatric Epilepsies. More info
- Oct. 7-8: 7th European Rett Syndrome Conference. More info / 5th European GRIN Conference. More info
- Oct. 9-10: 2023 Lafora Disease Science Symposium. More info
- Oct. 27-28: 11th Symposium ATP1A3 in Disease. More info

EpiCARE SCIENTIFIC PUBLICATIONS

- Patel, S. H., Panagiotakaki, E., Papadopoulou, M. T., Fons, C., De Grandis, E., Vezyroglou, A., Balestrini, S., Hong, H., Liu, B., Prange, L., Arzimanoglou, A., Vavassori, R., & Mikati, M. A. (2023).
  Methodology of a Natural History Study of a Rare Neurodevelopmental Disorder: Alternating Hemiplegia of Childhood as a Prototype Disease. Journal of child neurology, 8830738231197861. Advance online publication. Link.

- Trivisano, M., Dominicis, A., Stregapede, F., Quintavalle, C., Micalizzi, A., Cappelletti, S., Lisa Dentici, M., Sinibaldi, L., Calabrese, C., Terracciano, A., Vigevano, F., Novelli, A., & Specchio, N. (2023).
  Refining of the electroclinical phenotype in familial and sporadic cases of CSNK2B-related Neurodevelopmental Syndrome. Epilepsy & behavior : E&B, 147, 109436. Advance online publication. Link.

- Specchio, N., Nabbout, R., Aronica, E., Auvin, S., Benvenuto, A., de Palma, L., Feucht, M., Jansen, F., Kotulska, K., Sarnat, H., Lagae, L., Jozwiak, S., & Curatolo, P. (2023).
  Updated clinical recommendations for the management of tuberous sclerosis complex associated epilepsy. European journal of paediatric neurology : EJPN : official journal of the European Paediatric Neurology Society, 47, 25–34.Link

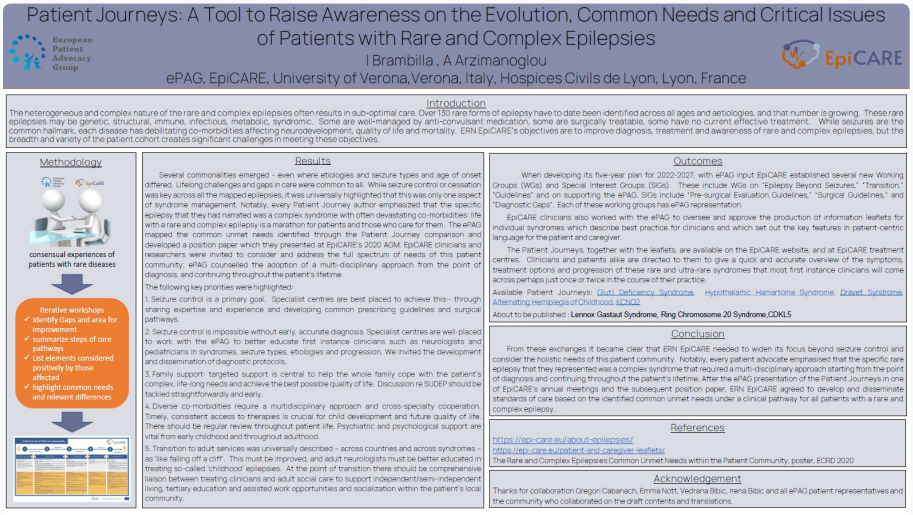


You can consult recent publications on rare and complex epilepsies, epilepsy surgery and other topics on our scientific publications webpage.

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

A word from our ePAGs

Awareness days



Patient Journeys: A Tool to Raise Awareness on the Evolution, Common Needs and Critical Issues of Patients with Rare and Complex Epilepsies

I Brambilla , A Arzimanoglou

ePAG, EpiCARE, University of Verona,Verona, Italy, Hospices Civils de Lyon, Lyon, France

European Patient Advocacy Group

EpiCARE

Introduction

The heterogeneous and complex nature of the rare and complex epilepsies often results in sub-optimal care. Over 130 rare forms of epilepsy have to date been identified across all ages and aetiologies, and that number is growing. These rare epilepsies may be genetic, structural, immune, infectious, metabolic, syndromic. Some are well-managed by anti-convulsant medication, some are surgically treatable, some have no current effective treatment. While seizures are the common hallmark, each disease has debilitating co-morbidities affecting neurodevelopment, quality of life and mortality. ERN EpiCARE's objectives are to improve diagnosis, treatment and awareness of rare and complex epilepsies, but the breadth and variety of the patient cohort creates significant challenges in meeting these objectives.

Methodology

Results

Several commonalities emerged - even where etiologies and seizure types and age of onset differed. Lifelong challenges and gaps in care were common to all. While seizure control or cessation was key across all the mapped epilepsies, it was universally highlighted that this was only one aspect of syndrome management. Notably, every Patient Journey author emphasized that the specific epilepsy that they had narrated was a complex syndrome with often devastating co-morbidities: life with a rare and complex epilepsy is a marathon for patients and those who care for them. The ePAG mapped the common unmet needs identified through the Patient Journey comparison and developed a position paper which they presented at EpiCARE's 2020 AGM. EpiCARE clinicians and researchers were invited to consider and address the full spectrum of needs of this patient community; ePAG counselled the adoption of a multi-disciplinary approach from the point of diagnosis, and continuing throughout the patient's lifetime.

The following key priorities were highlighted:

1. Seizure control is a primary goal. Specialist centres are best placed to achieve this:- through sharing expertise and experience and developing common prescribing guidelines and surgical pathways.

2. Seizure control is impossible without early, accurate diagnosis. Specialist centres are well-placed to work with the ePAG to better educate first instance clinicians such as neurologists and pediatricians in syndromes, seizure types, etiologies and progression. We invited the development and dissemination of diagnostic protocols.

3. Family support- targeted support is central to help the whole family cope with the patient's complex, life-long needs and achieve the best possible quality of life. Discussion re SUDEP should be tackled straightforwardly and early.

4. Diverse co-morbidities require a multidisciplinary approach and cross-specialty cooperation. Timely, consistent access to therapies is crucial for child development and future quality of life. There should be regular review throughout patient life. Psychiatric and psychological support are vital from early childhood and throughout adulthood.

5. Transition to adult services was universally described – across countries and across syndromes – as 'like falling off a cliff'. This must be improved, and adult neurologists must be better educated in treating so-called 'childhood' epilepsies. At the point of transition there should be comprehensive liaison between treating clinicians and adult social care to support independent/semi-independent living, tertiary education and assisted work opportunities and socialization within the patient's local community.

Outcomes

When developing its five-year plan for 2022-2027, with ePAG input EpiCARE established several new Working Groups (WGs) and Special Interest Groups (SIGs). These include WGs on "Epilepsy Beyond Seizures," "Transition," "Guidelines" and on supporting the ePAG. SIGs include "Pre-surgical Evaluation Guidelines," "Surgical Guidelines," and "Diagnostic Gaps". Each of these working groups has ePAG representation.

EpiCARE clinicians also worked with the ePAG to oversee and approve the production of information leaflets for individual syndromes which describe best practice for clinicians and which set out the key features in patient-centric language for the patient and caregiver.

The Patient Journeys, together with the leaflets, are available on the EpiCARE website, and at EpiCARE treatment centres. Clinicians and patients alike are directed to them to give a quick and accurate overview of the symptoms, treatment options and progression of these rare and ultra-rare syndromes that most first instance clinicians will come across perhaps just once or twice in the course of their practice.

Available Patient Journeys: Glut1 Deficiency Syndrome, Hypothalamic Hamartoma Syndrome, Dravet Syndrome, Alternating Hemiplegia of Childhood, KCNQ2

About to be published : **Lennox Gastaut Syndrome, Ring Chromosome 20 Syndrome,CDKL5**

Conclusion

From these exchanges it became clear that ERN EpiCARE needed to widen its focus beyond seizure control and consider the holistic needs of this patient community. Notably, every patient advocate emphasised that the specific rare epilepsy that they represented was a complex syndrome that required a multi-disciplinary approach starting from the point of diagnosis and continuing throughout the patient's lifetime. After the ePAG presentation of the Patient Journeys in one of EpiCARE's annual meetings and the subsequent position paper, ERN EpiCARE agreed to develop and disseminate standards of care based on the identified common unmet needs under a clinical pathway for all patients with a rare and complex epilepsy.

References

https://epi-care.eu/about-epilepsies/
https://epi-care.eu/patient-and-caregiver-leaflets/
The Rare and Complex Epilepsies Common Unmet Needs within the Patient Community, poster, ECRD 2020

Acknowledgement

Thanks for collaboration Gregori Cabanach, Emma Nott, Vedrana Bilic, Irena Bilic and all ePAG patient representatives and the community who collaborated on the draft contents and translations.

ePAG EpiCARE has presented a poster during IEC in Dublin titled Patient Journeys: A Tool to Raise Awareness on the Evolution, Common Needs and Critical Issues of Patients with Rare and Complex Epilepsies.

For those who haven't had the opportunity to view it, the poster is available here.

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!

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