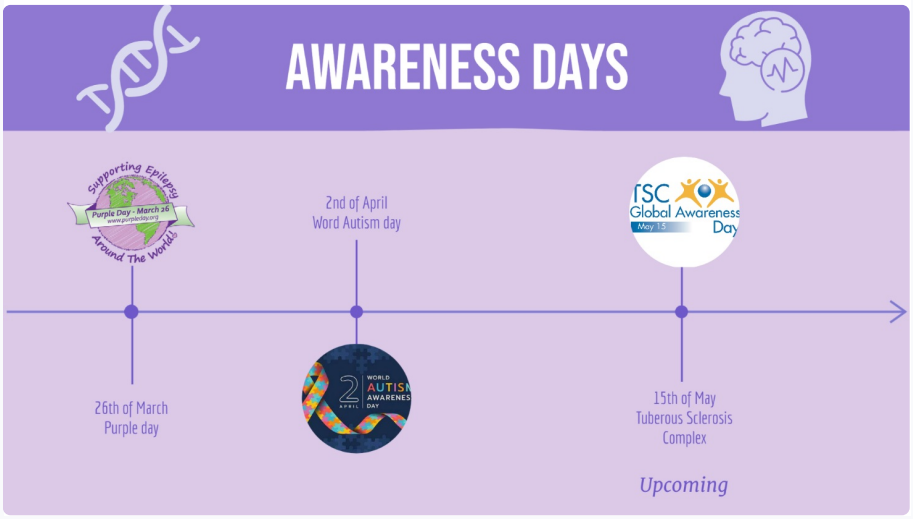
European Patient Advocacy Group

ePAG EpiCARE Newsletter
April 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



Purple day

On March 26th, Purple Day, a global initiative dedicated to raising awareness about epilepsy, a neurological disorder that affects millions of people worldwide was celebrated thanks to Cassidy Morgan. On this day, people are encouraged to wear purple and spread awareness about epilepsy by sharing their experiences, stories, and knowledge about this condition.

Purple Day is an opportunity to show support for people living with epilepsy, to educate others about the condition, and to promote understanding and acceptance. By coming together and raising awareness, we can help to break down the stigma that surrounds epilepsy and support those who are affected by this condition.

Word Autism Awareness Day

World Autism Day is a global initiative held on April 2nd and dedicated to raising awareness about autism, a complex neurological and developmental disorder that affects people in different ways. One aspect of autism that is not widely known is the high rate of comorbidity with epilepsy, particularly with rare and complex epilepsies. Research shows that individuals with rare and complex epilepsies have a significantly

increased risk of developing autism spectrum disorder (ASD) compared to the general population. In fact, up to 30% of individuals with epilepsy also have ASD. The link between epilepsy and autism is not yet fully understood, but it is believed that there may be shared genetic and neurobiological factors that contribute to the development of both conditions. As a result, it is important for individuals with epilepsy, particularly those with rare and complex epilepsies, to be closely monitored for signs of ASD and receive appropriate screening and support.
On World Autism Day, it is important to raise awareness not only about autism itself but also about the link between autism and epilepsy. By increasing understanding and promoting research, we can work towards better supporting individuals and families affected by these complex neurological conditions.

Meet our ePAGs

Emma Nott is based in the UK and in 2015, together with UK families affected by HH, she set up Hope for HH-UK and is a trustee of this British arm of the Hope for Hypothalamic Hamartoma organization. Since 2018 Hope for HH-UK has been a member of EpiCARE, the European network for the rare and complex epilepsies. 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!

Emma Nott presentation video

News

New articles

New article about KCNQ2-realted encephalopathy is published in Epilepsy & Behaviour!

The survey was developed in collaboration with caregivers from different European family associations, and was aimed at investigating the onset and frequency of epileptic seizures, anti-seizure medications (ASM), hospitalizations, stages of development, and comorbidities.
Huge gratitude to the organisations involved: Alessandra Costa, Elisa Remonato and Sara Gramaglia (European KCNQ2 Association Odv, Italy), T. De Benito (Asociación KCNQ2 España, Spain), Sebastian Bethge (KCNQ2 e.V. Germany, Germany), S. Cardot (KCNQ2 France Développement, France), Iga Montwill (KCNQ2 Poland Group, Poland) and Isabella Brambilla and Elisa Giarola (Dravet Italia Onlus, Alleanza Epilessie Rare e Complesse, Italy) for their activity and great contribution!

Article is available here (open access)


Epilepsia Open™ The Open Access Journal of the International League Against Epilepsy

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ORIGINAL ARTICLE | Open Access

A registry for Dravet syndrome: The Italian experience

Simona Balestrini, Viola Doccini, Sabrina Giometto, Ersilia Lucenteforte, Salvatore De Masi, Elisa Giarola, Isabella Brambilla, Federica Pieroni, Marco Perulli, Domenica Battaglia, Nicola Specchio, Francesca Ragona, Tiziana Granata, Simona Pellacani, Annarita Ferrari, Carla Marini, Sara Matricardi, Elisabetta Cesaroni, Lucio Giordano, Patrizia Accorsi, Vittorio Sciruicchio, Paolo Tinuper, Tullio Messana, Angelo Russo, Dario Pruna, Margherita Nosadini, Valentina De Giorgis, Davide Caputo, Residras Collaboration Group, Serena Pellegrin, Tommaso Lo Barco, Francesca Darra, Bernardo Dalla Bernardina, Renzo Guerrini ... See fewer authors

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Article about the Italian experience with Residras – registry for Dravet syndrome and other syndromes correlated with SCN1A and PCDH19 gene mutation was published in Epilepsia Open

The Residras registry represents a large collection of standardized national data for the DS population. It relies on a shareable and interoperable framework, which promotes multicenter high-quality data collection. In the future, such integrated platform may represent an invaluable asset for easing access to cohorts of patients that may benefit from clinical trials with emerging novel therapies, for drug safety monitoring, and for delineating natural history. Its framework makes it improvable based on growing experience with its use and easily adaptable to other rare and complex epilepsy syndromes.
Full article is available here.

Upcoming events

2nd SCN2A and SCN8A Conference

Save the date! More information coming soon!

Families attending the 2nd European Conference 26-27th May, please register (free of charge) by sending an email to Dr Francesca Furia.

If you have special requests please contact Francesca and/or Roland

Inaugural family and scientific conference on CHD2-related disorders

The conference by Coalition to Cure CHD2, will be held on June 2nd - 4th, 2023 at Disney's Coronado Springs Resort in Lake Buena Vista, Florida! CHD2 families and researchers from across the globe will come together for this two day event to learn about the latest developments and research in CHD2. Virtual attendance is also possible!

More information about the conference and registration here

Meet the patients' associations

Asociación de Afectados CDKL5

Main objectives:

- To raise funds for research into CDKL5 and other related diseases.
- To obtain and manage resources to provide support and improve the quality of life of those affected and their families.
- To give visibility to the disorder in the clinical, health and social spheres.
- To promote meetings between affected relatives, medical specialists and researchers at national and international level.
- To support and participate in international cooperation programmes Member of CDKL5 Alliance.

Most outstanding or important project:
The AACDK5 is currently funding the following study: Search for treatments for CDKL5 deficiency syndrome: From the Centro de Investigación Príncipe Felipe de Valencia, the team of PI: Máximo Ibo Galindo Orozco (Developmental Biology and Disease Models) are working with the fruit fly or vinegar fly "Drosophila melanogaster" with the aim of studying CDKL5 deficiency syndrome in this biological model, in order to find new therapies for CDKL5.

Principales objetivos:

- Recaudar fondos destinados a la investigación del CDKL5 y otras enfermedades relacionadas.
- Obtener y gestionar recursos para proporcionar apoyo y mejora de la calidad de vida de las personas afectadas y sus familias.
- Dar visibilidad del trastorno en el ámbito clínico sanitario y social.
- Promover encuentros entre familiares afectados, médicos especialistas e investigadores de ámbito nacional e internacional.
- Apoyar y participar en programas de cooperación Internacional Member of CDKL5 Alliance

Proyecto más destacado o importante:
La AACDK5 financia en la actualidad el siguiente estudio:
Búsqueda de tratamientos para el síndrome por deficiencia de CDKL5: Desde el Centro de Investigación Príncipe Felipe de Valencia, el equipo del IP: Máximo Ibo Galindo Orozco (Biología del Desarrollo y Modelos de Enfermedad) están trabajando con la mosca de la fruta o mosca del vinagre "Drosophila melanogaster" con el objetivo de estudiar el síndrome de deficiencia de CDKL5 en este modelo biológico, a fin de encontrar nuevas terapias para CDKL5.

Contacts and social networks:
Web: News aacdkl5.org
Facebook: https://www.facebook.com/CDKL5-España
Teaming: https://www.teaming.net/asociaciondeafectadoscdkl5
Twitter: @cdkl5spain
Instagram: cdkl5_spain
Youtube: https://www.youtube.com/channel/UCImR1ge8tmp6LDcYmft6eEQ

Coalition to Cure CHD2

We're called Coalition to Cure CHD2. Our mission is to improve the lives of those affected by CHD2-related disorders by funding research necessary for uncovering a cure.
Our most important project is our inaugural family and scientific meeting 2nd-4th of June 2023. We have also awarded two pre-clinical research grants last year to further the basic science understanding of our disorder.

Our email: info@cureCHD2.org.
Website: CHD2 Epilepsy | Coalition to Cure CHD2
twitter: @cureCHD2
FB: CHD2 Support and Research Group (private) and Coalition to Cure CHD2
Instagram: cureCHD2.
Linkedin: Coalition to Cure CHD2.

Alternating hemiplegia of childhood Association Denmark

The purpose of the association:

- To bring together the families where AHC occurs.
- To increase their knowledge and support the fight against problems with the disease.
- To disseminate knowledge about the treatment options and the options for help for the families.

To create unity between the families, establish contact with and collaborate with international groups, organizations or associations that work with AHC.
E.g. through participation in international courses and conferences.
To create more knowledge in the health system about AHC.
The most important projects are family meetings, theme days and international conferences.

Contact: Suffia Madsen, president - suffiam@gmail.com
Homepage: https://ahckids.dk/
Facebook: https://www.facebook.com/AHCkids.dk

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