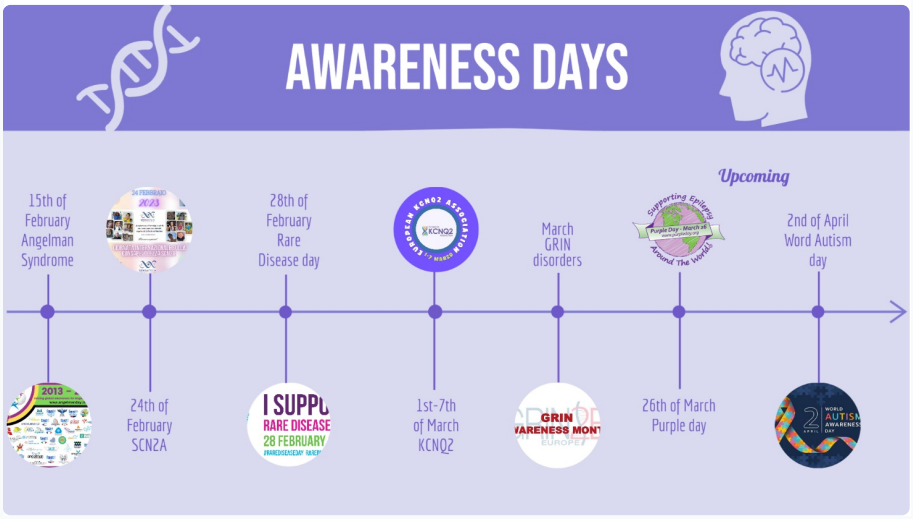
European Patient Advocacy Group

ePAG EpiCARE Newsletter
March 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



Angelman syndrome Awareness Day

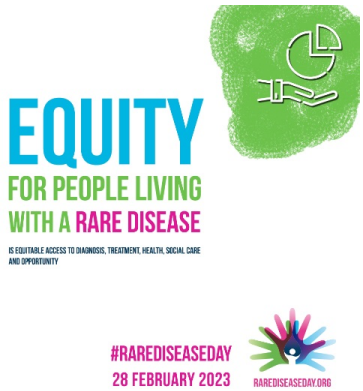
On February 15th, 10th International Angelman syndrome Awareness Day was celebrated!
The purpose of marking day is to: raise awareness worldwide of the condition, mobilise people to action & encourage fundraising for the AS organisation in their country, promote research and educational resources in the organisation's own country and remember those people with Angelman Syndrome who are no longer with us. On this date some cities in Europe were lit up in blue in support to Angelman community!

SCN2A Awareness day!

SCN2A is located on chromosome 2 at position 24 hence 24th of February was picked to celebrate the awareness day!

Symptoms of this complex conditions vary and can include: hypotonia, seizures, breathing difficulties, intellectual disability and autistic features.
There is no cure for this condition yet, but clinical trials are undergoing!

Rare disease day



On 28th of February we celebrated Rare disease day! Among our EpiCARE Network we distinguish more than 130 rare and complex epilepsies, but besides our diversities we share many common grounds that highly affect the quality of life of all patients!
Our ultimate goal is equality, equality in access to diagnosis, treatment, health and social care! To achieve these goals, we are taking actions to connect and collaborate more with many associations for rare and complex epilepsies across Europe. Only together we can achieve this for our patients across Europe!

KCNQ2 Awareness week

KCNQ2 Awareness week was marked from 1st to 7th of March!
KCNQ2-DEE is a severe form of neonatal epilepsy caused by a mutation on KCNQ2 gene located on chromosome 20. It is the most common cause of neonatal epilepsy with encephalopathy since seizures are only a part of it, and it affects the intellectual abilities, speech and language skills and motor function among others. Autistic features can also be present. To learn more about the KCNQ2-DEE check out the Patient Journey prepared by European KCNQ2 association. Stay tuned as the new publication on KCNQ2-DEE is coming soon!

GRI disorders Awareness month

March is dedicated for raising awareness about GRI related disorders! This group includes many genes such as: GRIN1A, GRIN2A, GRIN2B, GRIN2D, GRIA1, GRIA2, GRIA3, GRIA4 and GRIK2. It is roughly estimated that 1 500 people worldwide have been diagnosed with GRI related disorders although there could be a lot more people out there who haven't been diagnosed yet or have been misdiagnosed.
Epilepsy is more common in specific GRI mutations, but most of the patients are likely to develop intellectual disability, speech impairment, mobility impairment, sleep difficulties, low muscle tone etc.

Meet our ePAGs

Irena Bibić is the president of Dravet syndrome Croatia, and has been an ePAG member since 2020.

Have a look on a video to meet our ePAG, her view on EpiCARE network, role of ePAGs and benefits it brings for the patient community!

Irena Bibic presentation video

News

New leaflet published!

New leaflet for healthcare professionals and patients and caregivers is dedicated to Rett syndrome. You can view and download the leaflet here.
Thank you Rett Syndrome Europe for investing your time in the preparation of these documents!

Upcoming events

Dravet syndrome Foundation

Endorsed by ERN EpiCARE, Dravet conference will be held on March 23rd.
For more information and registration, visit: dravetconference.com

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 on:
https://www.curechd2.org/conference

Meet the patients' associations

SCN2A Famiglie in Rete APS

Famiglie SYNGAP1 Italia APS

We represent the people with a mutation on the Syngap1 gene. The Syngap1 gene located on the sixth chromosome at position 21, encodes the protein of the same name, which is very important in nerve communication at synapses. Its deficiency and structural abnormalities can lead to the following symptoms:
intellectual disability, epilepsy, autism spectrum disorder, hypotonia, global development delay, gross and fine motor skill delays, speech and language disorders, visual abnormalities, feeding difficulties, gastrointestinal issues, high pain threshold, sensory processing disorder, sleep disturbance, severe behavior problems, self-injurious behaviors, dyspraxia, ataxia and gait abnormalities, anhidrosis, orthopedic issues and hip dysplasia.
We are continually looking for other people with a mutation to the Syngap1 gene in the knowledge that there are many more in Italy than the thirty that we know of. Especially in adulthood.
We have always been active in the search for funds to finance the necessary scientific research.
We maintain contacts with other international Syngap1 Associations to increase the necessary mutual knowledge to improve the quality of life of all of us affected by this syndrome.
We are working to realise the following two projects on 20 and 21 June 2023 in Rome in the beautiful setting of the Botanical Garden:

1. WORKSHOP with the participation of international Scientists, Clinicians, Support Teachers, Health Professions involved in care and assistance, Families and other interested operators;
2. Meeting of Syngapian families.

Ci interessiamo di Persone con mutazione al gene Syngap1. Il gene Syngap1 collocato nel sesto cromosoma dalla posizione 21, codifica la omonima proteina, molto importante nella comunicazione nervosa a livello delle sinapsi. La sua carenza e le sue anomalie strutturali possono determinare le seguenti sintomatologie: disabilità intellettuale, epilessia, disturbo dello spettro autistic, ipotonia, ritardo dello sviluppo globale, ritardi nelle capacità motorie grossolane e fini, disturbi della parola e del linguaggio, anomalie visive, difficoltà di alimentazione, problemi gastrointestinali, alta soglia del dolore, disturbo dell'elaborazione sensoriale, disturbi del sonno, gravi problemi di comportamento, comportamenti autolesivi, disprassia, atassia e anomalie dell'andatura, anidrosi, problemi ortopedici e displasia dell'anca.
Cerchiamo continuamente altre Persone con mutazione al gene Syngap1 nella consapevolezza che in Italia ce ne siano molto di più delle trenta che conosciamo. Specialmente nell'età adulta.
Siamo da sempre attivi nella ricerca di fondi per finanziare le necessarie ricerche scientifiche.
Manteniamo contatti con altre Associazioni internazionali di syngapiani per aumentare la necessaria conoscenza reciproca per migliorare la qualità della vita di tutti noi coinvolti da questa sindrome.
Stiamo lavorando per realizzare i seguenti due progetti il 20 e 21 giugno 2023 a Roma nella stupenda cornice dell'Orto Botanico:
1 WORKSHOP con la partecipazione di Scienziati internazionali, Clinici, Insegnanti di Sostegno, Professioni sanitari coinvolte nella cura eassistenza, Famiglie e altri operatori interessati;
2 Incontro tra famiglie di Syngapiani.

- E-mail : si.amo@syngap1.it / syngap1@pec.syngap1.it
- Websites : www.syngap1.it / syngapglobal.net/international-orgs
- Facebook : www.facebook.com/syngap1italia / www.facebook.com/groups/Syngap1Italia
- Instagram : www.instagram.com/syngap1_italia
- Twitter : https://twitter.com/Syngap1Italia?t=wKZpO5oML1Wbhc4jttNq7w&s=08

GRIN Europe

GRIN Europe is a pan-European non-profit association founded by a group of GRIN families actively involved in their local communities and across European countries whose main aim is to help and support other GRIN families, GRIN groups and GRIN associations in and beyond Europe. Our main

interest is to bring knowledge about GRIN related Disorders (GRD) to families, physicians, and researchers. Our mission is to improve the availability and quality of educational and scientific publications, guidelines, and recommendations, to make this an easily available resource for families and caregivers, and to improve the quality of care that a GRIN patient should receive across healthcare systems in Europe. Our aim is to help create a GRIN expert panel that consists of European neurologists and specialists in rare and complex disorders, such as epilepsies, behavioural and movement disorders, and to promote and advocate for a better understanding of GRIN disorders throughout Europe. We try to accomplish this through various initiatives such as scientific based conferences where families, doctors and researchers come together to exchange their experiences and insights and learn from each other. In order to better represent the entire European GRIN community, effective as of December 2022, we have changed our name from GRIN2B Europe to GRIN Europe. In this way we aim to provide support to a larger group of families affected by GRIN disorders.

- admin@grineurope.com
- Facebook: https://www.facebook.com/grineurope
- New website is coming out soon!

GRIN EUROPE Care 4 GRIN

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