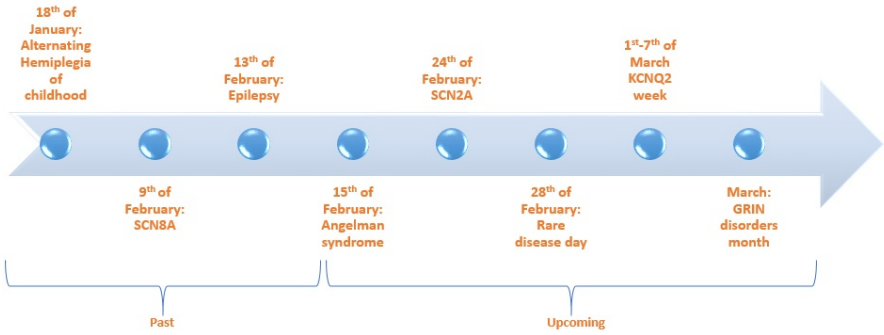
# ePAG EpiCARE Newsletter
# February 2023

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.

What a better day to start the Newsletter but on Valentine's day, as saint Valentine is a patron saint of epilepsy!

If you wish to disseminate the information about your association, projects, activities and events you are planning, please contact: epag.epicare@gmail.com

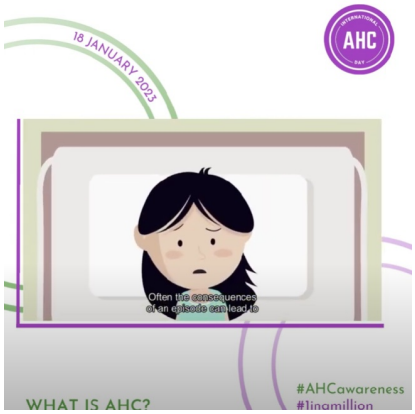
## AWARENESS DAYS




### Alternating Hemiplegia of Childhood (AHC): International Awareness Day 18th of January 2023

Alternating hemiplegia od childhood Federation of Europe (AHCFE) released an excellent video for AHC awareness day!

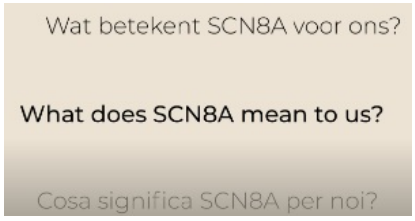

To learn more about AHC, check out our leaflets and Patient Journey, which were prepared by AHC-UK, IAHCRC and AHC18+ e.V.




Click on the picture above to view an excellent video prepared by Alternating hemiplegia of childhood Federation of Europe (AHCFE)

## SCN8A: International Awareness Day 9th of February 2023


Click on picture to view Awareness video prepared by SCN8A Italia!

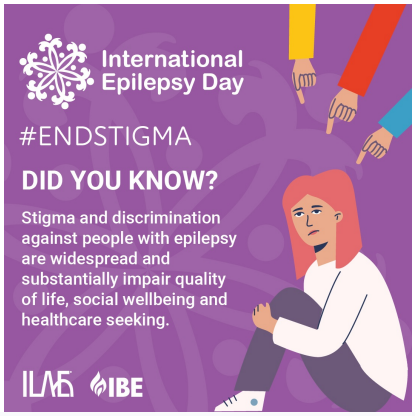



On the 9th of February, we celebrated International SCN8A Awareness Day! At this date, the SCN8A European Federation was officially established!

Click on picture to view a video of the new SCN8A European Federation!

This date was chosen for many reasons, including the Awareness day as well as celebrating and honoring Shay and her life, the first known case of SCN8A who unfortunately passed away before her father's discovery.

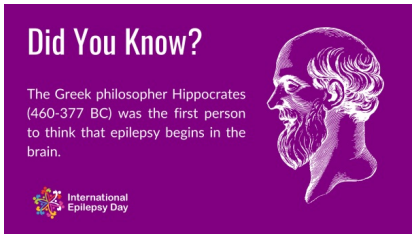
## Epilepsy: International Awareness Day 13th of February 2023




This year's theme for International Epilepsy Day (13 February), "Step Up Against Stigma", provided a platform for people with epilepsy, and those who care for them, to dispel myths with facts. "Epilepsy affects almost every aspect of the life of the person diagnosed with the condition," said Dr. Francesca Sofia, president of the International Bureau for Epilepsy (IBE). "For many people living with epilepsy, the stigma attached to the condition is often more difficult to deal with than the disease itself." In many countries, lack of awareness about epilepsy as a treatable brain condition remains a central public health challenge.

Educating people about epilepsy and dispelling myths can reduce stigma and discrimination at school, at work, in the clinic, across the community and at home.
International Epilepsy Day is celebrated in more than 100 countries, with epilepsy organizations and advocates scheduling ministerial meetings, informational workshops, awareness-raising campaigns, fundraising events, and social media activity.
It is estimated that more than 50 million people are living with epilepsy around the world. The provision of safe, effective, and highly affordable ($5 per person/year) anti-seizure medicines could control seizures in up to 70% of people with epilepsy. On the other hand, around 30% of epilepsy patients have drug resistant epilepsy which limits the effects of the treatments as well as the quality of life. Epilepsy ranks among the top five causes of global neurological disability; an estimated 125,000 deaths per year globally are related to the condition. Untreated epilepsy is a life-threatening disorder.
The death rate for people with epilepsy is three times that of the general population, and for some specific syndromes even higher. Every fourth case of epilepsy could be prevented by addressing and minimizing risk factors, such as

stroke, brain infections, birth injuries, and head trauma. However, prevention strategies remain limited and underexplored.




People with epilepsy are disproportionately affected by other conditions as well, such as depression, anxiety, dementia, migraine, heart disease, peptic ulcers, and arthritis. Many more additional conditions affect the ones with rare and complex epilepsies.

## Meet the patients' associations

ePAG EpiCARE decided to dedicate 2023 to patients' associations of rare nad complex epilepsies. Having more that 130 rare and complex epilepsy conditions, we wish to connect more, a help in making every voice heard.

Although we are many, and within each there are diversities, with years of advocacy we saw that we also share many common grounds, regardless of a syndrome name or gene mutations. We want to learn more about rare and complex epilepsies you represent and highlight the common issues we share in order to set common goals in providing better quality of life for all patients!
So, for starters, we decided to establish a newsletter dedicated to associations in order for all of us to connect better, collaborate and make each's voice heard.
We hope that with this newsletter, patient's associations will reach wider community of patients and caregivers, healthcare professionals and public.

We are looking forward to hearing you and disseminating your information.
Join us!

Isabella Brambilla - Chair of the epags

Presentation of the ePAGS - Video available here

### New leaflets for patients and caregivers and healthcare professionals

A new leaflet for SYNGAP1 is out and available on the EpiCARE website!

We would like to thank Famiglie Syngap1 Italia APS for their work on providing information about SYNGAP1 together with physicians!

Leaflet translations are actively being published. Recently added translations include Romanian and

Croatian language. If you are interested in translating leaflets into your language, please contact: epag.epicare@gmail.com


## New EpiCARE webinar: PCOMS in clinical trial

Webinar "PCOMS in clinical trials" was held on 2nd of February. With prof. Rima Nabbout, our ePAG chair, Isabella Brambilla, was also a speaker on this great webinar.
If you weren't able to join the webinar, the recording is available at the following site: https://epi-care.eu/past-webinars/



## New article!

Members of ePAG EpiCARE and patient representatives joined forces in preparing and publishing an article titled: Rare. The importance of research, analysis, reporting and education in 'solving' the genetic epilepsies: A perspective from the European patient advocacy group for EpiCARE.

Authors of the article are: Emma Nott (Hope for Hypothalamic Hamartomas), Katherine Behl (Alternating Hemiplegia of Childhood UK), Isabella Brambilla (Dravet Italia Onlus), Monica Lucente (Associazione Italiana GLUT1 Onlus), Rosaria Vavassori (International Alternating Hemiplegia of Childhood Research Consortium) and Allison Watson (Ring20 Research and Support UK) together with the physician: prof. Green, prod. Dalla Bernardina and prof. Hidelbrand.

Article was published in the European Journal for Medical Genetics, and highlights the transformative effect that early genetic diagnosis on the quality of life of individuals with epilepsy, discusses the significant gaps in knowledge and care that currently exist from the patient perspective, and describes some of the initiatives that patient-advocacy organisations have undertaken, and would promote, to fill those gaps. The full article is available as open access and can be downloaded in pdf until next March 11th, 2023 here.

# Meet the patients' associations

### UK Infantile Spasms Trust (UKIST)

The UK Infantile Spasms Trust (UKIST) provides a network of support to families with children affected by the rare seizure disorder infantile spasms, also known as West syndrome. The charity offers a lifeline to anxious parents via its online community, and works alongside medical professionals to raise awareness of this little known condition, which can be very difficult to recognise – but potentially very damaging if left untreated.
Our current major project is collaborating with a medical education company to produce digital training resources for health professionals. We have over 2000 members in our support group and

we continue to provide peer support, online resources and grants to aid in their children's recovery.
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Facebook / Youtube / Twitter: @uk_is_t

UKIST
THE UK INFANTILE SPASMS TRUST

## Dravet Syndrome Foundation

SÍNDROME DE DRAVET
FUNDACIÓN
www.dravetfoundation.eu

Our organization was born with the aim of promoting, encouraging and connecting the world's leading research centers on Dravet syndrome and other related diseases. It is based on the principle of a collaborative model uniting professionals, patients, researchers, physicians, volunteers and sponsors in the search for an effective therapy. . The main objective is to eliminate the barriers that prevent research on Dravet syndrome from advancing, as well as to find effective drugs and treatments that allow eliminating, mitigating and even curing the disease. One of our active projects is the Dravet Syndrome Conference 2023 will be held in March 2023. The purpose of this conference is for all stakeholders involved in the treatment of this rare syndrome to learn about the latest advances in drugs, treatments and therapies from renowned researchers and scientists, including Dr. Charlotte Dravet, who gives her name to this rare disease. The meeting also offers the opportunity to interact with other families living with this syndrome called "familial condition" and exchange concerns and hopes. For more information: dravetconference.com

Nuestra organización nació con el objetivo de promocionar, incentivar y conectar los principales centros de investigación mundiales del síndrome de Dravet y otras enfermedades relacionadas. Se sustenta en el principio de un modelo colaborativo uniendo a profesionales, pacientes, investigadores, médicos, voluntarios y patrocinadores en la búsqueda de una terapia efectiva. El objetivo principal es eliminar las barreras que impiden que la investigación sobre el síndrome de Dravet avance, así como encontrar fármacos y tratamientos efectivos que permitan eliminar, mitigar e incluso curar la enfermedad. En marzo 2023 se llevará a cabo la Dravet Syndrome Conference 2023. La finalidad de estas jornadas es que se todos los stakeholders implicados en el tratamiento de este raro síndrome conozcan los últimos avances en medicamentos, tratamientos y terapias, de la mano de renombrados investigadores y científicos, incluyendo a la doctora Charlotte Dravet, que da nombre a esta enfermedad rara. El encuentro ofrece también la oportunidad de interactuar con otras familias que viven con este síndrome denominado «condición familiar» e intercambiar preocupaciones y esperanzas. Para más información ingresar a dravetconference.com

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+34 622 88 35 58
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## Stichting GRIN syndroom

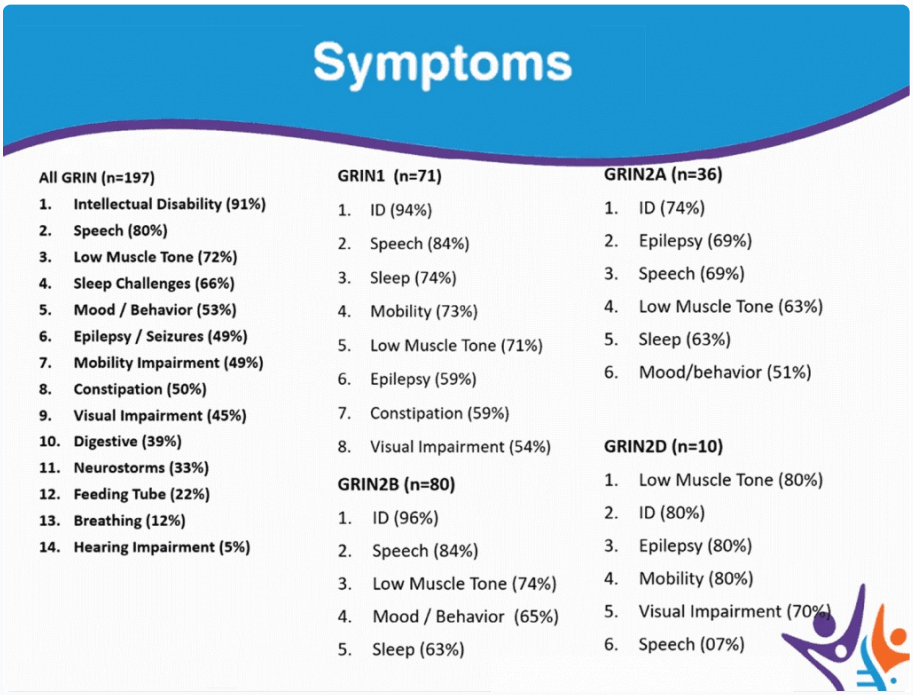
GRIN verbindt
www.grinsyndroom.nl

Our foundation aims to represent and support parents and patients with GRIN and GRIA mutations especially in the Netherlands and in Belgium. We do this by providing information in Dutch on our website www.grinsyndroom.nl (translations available by clicking the flag) and in our newsletter 4x per year.
In this newsletter you will receive the latest development in research and clinical trials, it will also include updates on behalf of the Dutch center of expertise for GRIN and GRIA: ENCORE.
Subscription to our foundation is free can be done by leaving your email address at: https://grinsyndroom.nl/nieuwsbrief/
Our foundation is an easy platform to connect with other parents and patients. This can be done by joining Facebook group for parents and patients only or by joining one of our (online) meetings. For the latest meeting dates, see: https://grinsyndroom.nl/nieuws/.
As shown below in a survey by CureGRIN, not all GRIN and GRIA patients suffer from epilepsy. However some case of epilepsy are severe and hard to tread with generic medication for epilepsy. In future new treatments for GRIN and GRIA could become available that target special GRIN and GRIA proteins. So if you or your child suffers from epilepsy due to a GRIN/GRIA mutation. Make sure to stay connected.

Onze stichting vertegenwoordigd en ondersteund ouders en patiënten met GRIN en GRIA mutatie en is op de eerste plaats gericht op Nederland en België. Dit doen we door het delen van Nederlandstalige informatie via onze websiteen 4x per jaar in onze nieuwsbrief. In deze nieuwsbrief

vind je de laatste ontwikkelingen op het gebied van onderzoek en klinische studies voor nieuwe medicatie. Ook vind je er updates vanuit het Nederlandse expertise centrum voor GRIN and GRIA: ENCORE.
Aanmelding is gratis en gebeurd door het achterlaten van je emailadres op: https://grinsyndroom.nl/nieuwsbrief/
Onze stichting is ook een handige manier om in contact te komen met andere ouders en patiënten. Dit kan je doen door je aan te sluiten bij onze besloten Facebook groep: (allen voor ouders en patiënten) of door je aan te melden voor een van onze (online) meetings. Voor de laatste data van meetings zie: https://grinsyndroom.nl/nieuws/.
CureGRIN heeft een enquête gehouden welke symptomen er het meeste voorkomen voor GRIN en GRIA patiënten. In de onderstaande uitslag is het duidelijk dat niet alle patenten epilepsie hebben. Echter als epilepsie voorkomt is het vaak lastig te behandelen met standaard epilepsie medicatie. In de toekomst komen er mogelijk nieuwe behandelingen voor GRIN en GRIA beschikbaar die specifiek werken voor deze aandoeningen. Dus als jij of je kind getroffen bent door epilepsie veroorzaakt door een GRIN/GRIA mutatie, zorg dat je je aanmeld en op de hoogte blijft.

https://grinsyndroom.nl / info@grinsyndroom.nl
Facebook (for families only)




## Upcoming events

### Third International KCNB1 Conference

The team of the Rare Epilepsies Reference Center, Necker Enfants Malades and the Developmental and Epileptic Encephalopathies research group of INSERM Unit 1163, Institut Imagine, along with the KCNB1 France association, are pleased to invite you to the 3rd KCNB1 conference that will be held on Saturday - March 11th 2023 from 9 am to 5 pm (CET).
This day dedicated to families is open to all

health professionals wishing to better understand this pathology and its impact and to follow new avenues of research around this pathology. Interventions will be in French, Powerpoint presentations will be written in English. Registration here.
The conference will happen: - Face to face at the Imagine Institute, Paris, France (from 10am to 4pm) - By video conferencing for people unable to attend (from 10am to 1pm).

**PROGRAMME/PROGRAM**

**9:15** Ouverture de la connexion/ Remote connection open

**9:30** Bienvenue/ Welcome
Mot de l'association KCNB1/ Welcome of the KCNB1 association **(M Cassard)**
Mot du **Pr R Nabbout**, coordinatrice du Centre de référence épilepsies rares, Hôpital Necker, Institut Imagine, Université de Paris cité/ Welcome of the **Pr R Nabbout, CRéER Necker, Chair GEEN-DS, Institut Imagine.**

**10:00** L'Etiologie des épilepsies et des encéphalopathies épileptiques développementales/ Epilepsies DEEs (Developmental and Epileptic Encephalopathy)- the place of etiology **(Pr R Nabbout)**

**10:30** Le gène KCNB1 et ses variants pathogènes / KCNB1 gene and its pathogenic variants **(Dr G Barcia, Necker and Dr Marina Trivisano, Bambino Gesu, Rome)**

**11:00** Pause/ Break

**11:30** Corrélation génotype/ phénotype : Le besoin de données / Corrélation genotype-phenotype- need for data **(Dr Claire Bar / Delphine Breuillard, PhD )**

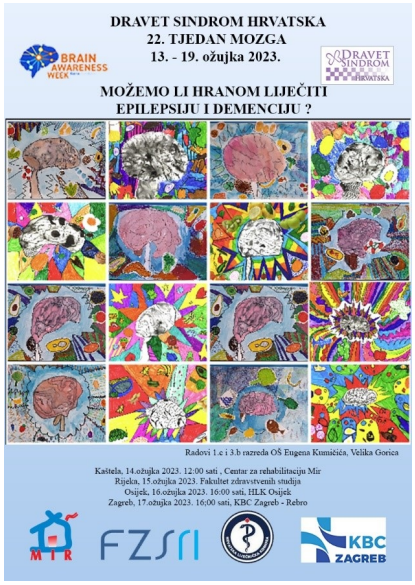
**12:30** Intelligence artificielle et data des forums des patients/ The use of data from patients' forum **(E Le Priol, PhD Student)**

**13:00** Déjeuner/ Break

**14:00** Les modèles animaux de recherche – Imagine/ Animal models-Imagine **(E Kabashi, director of research, Institut Imagine/ L Robichon, PhD Student)**

**15:00** Le programme de recherche sur la mutation du gène KCNB1 au sein de l'hôpital Necker/ KCNB1 programme at CRéER-Necker-Chaire GEEN-DS, Institut Imagine

Discussion générale/ General discussion




## Dravet syndrome Croatia

Brain week: Can we treat epilepsies and dementia with food?
Tjedan mozga: Možemo li hranom liječiti epilepsiju I demenciju?

14th- 18th of March 2023 in Split, Rijeka, Osijek and Zagreb
Od 14. do 18. ožujka 2023. u Splitu, Rijeci, Osijeku I Zagrebu.

For more information follow our webpage and social media!



## Dravet syndrome Foundation

Dravet syndrome conference 2023
March 23rd; for more information and registration, visit: dravetconference.com

Follow us on social media!

EpiCARE

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