ERDERA
European Rare Diseases Research Alliance

Challenges in diagnosis and care

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Views and opinions expressed are those of the author(s) only and do not necessarily reflect those of the European Union or any other granting authority, who cannot be held responsible for them.

Disclosures

- RN has received consulting and speaking fees from Nutricia, Zogenix, Advicenne, Eisai , Jazz and GW Pharmaceuticals, Neuraxpharma, Stoke, newbridge pharmaceuticals, UCB, Biocodex
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NOTE: The views on these slides are personal and are not affected by any conflict of interest

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Challenges facing patients and families

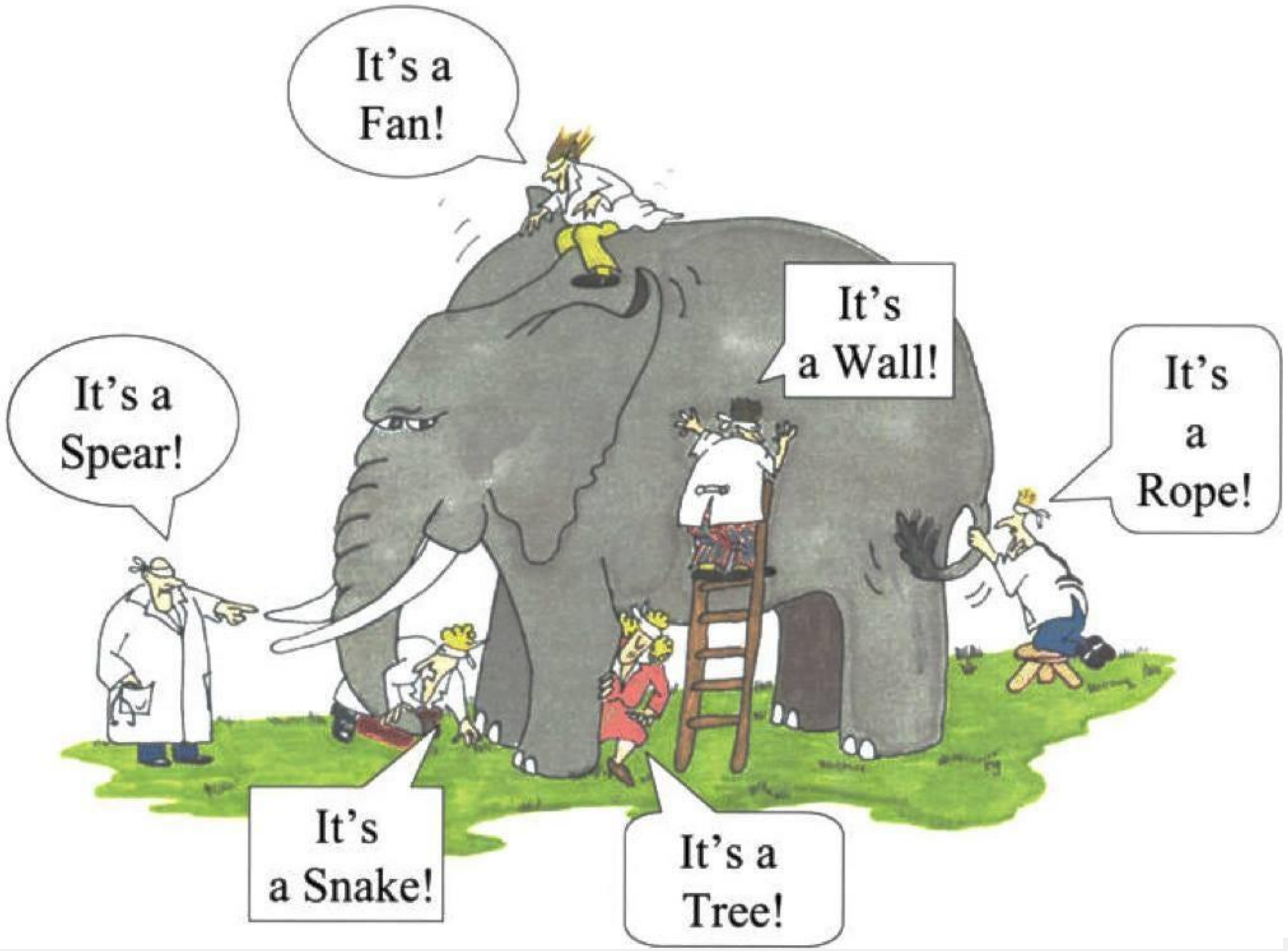
And Physicians!

Diagnosis

Research

Care

A good diagnosis is always holistic and patient centered!



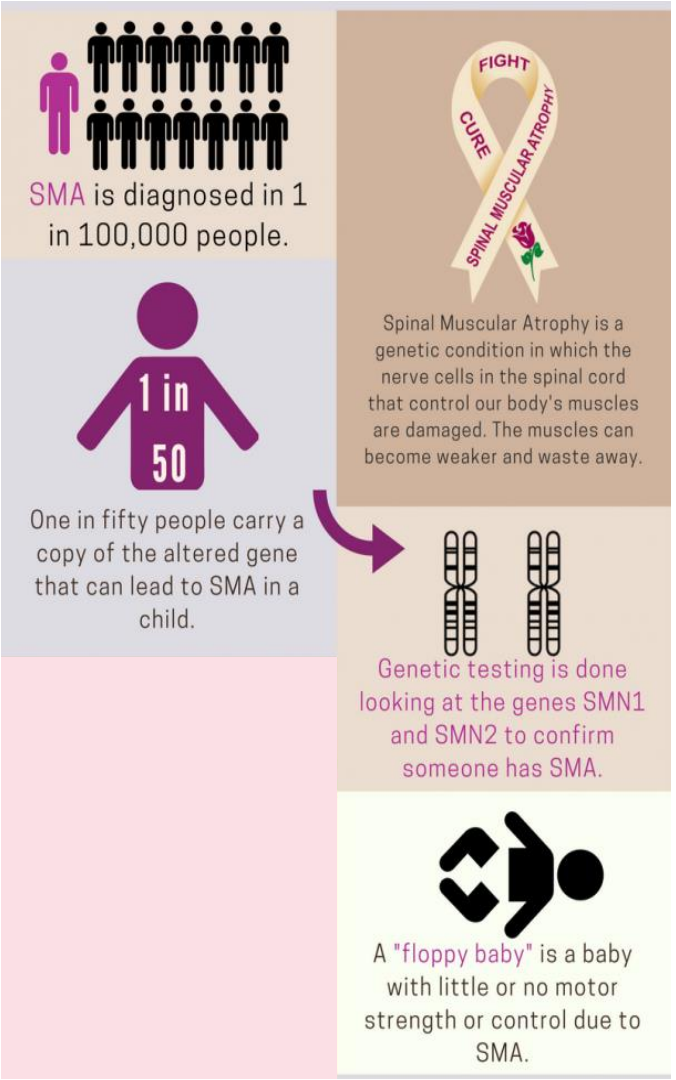
A train can hide another

The clinical examination and the history from the family are major steps!

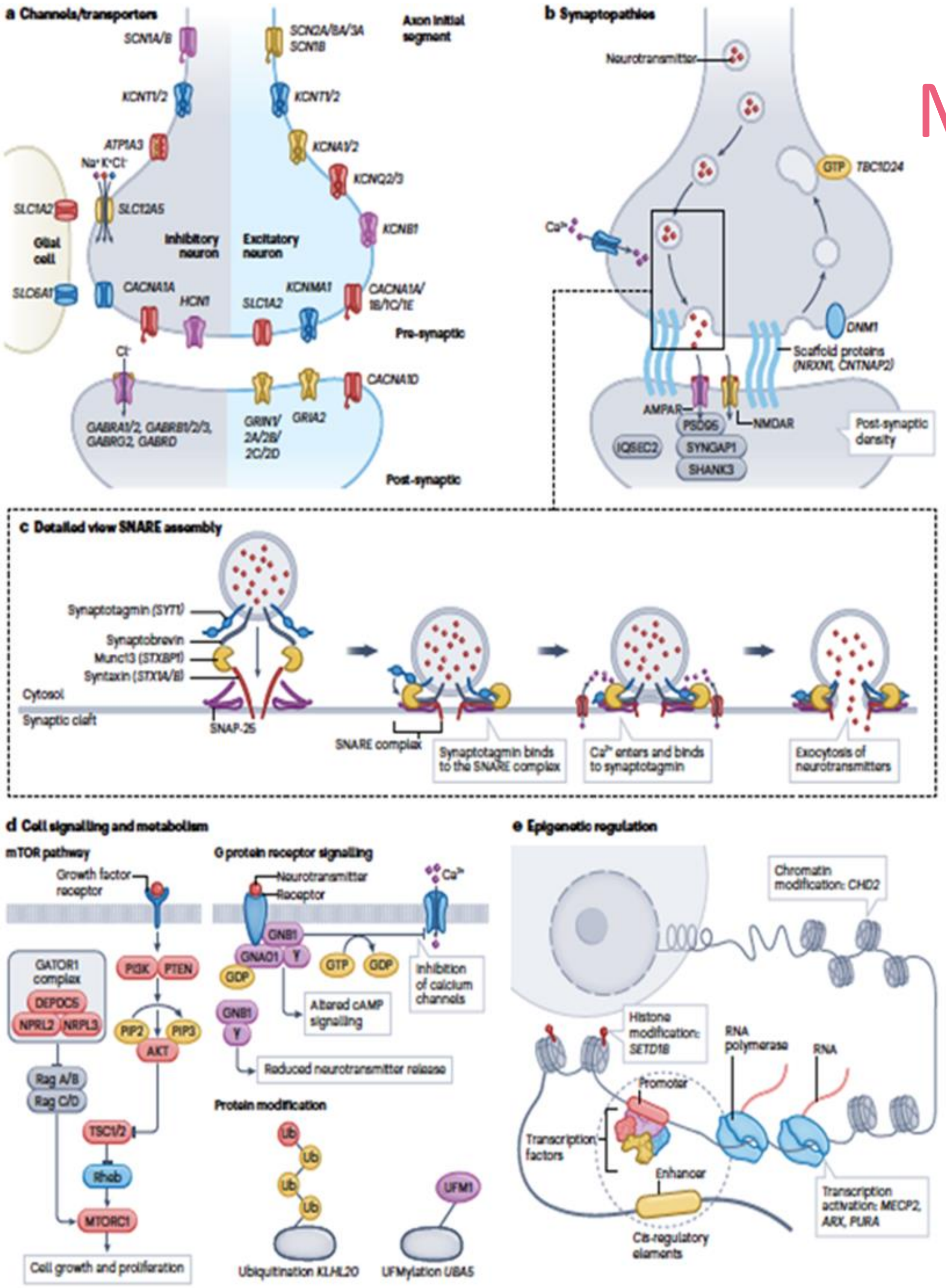
A train can hide another!

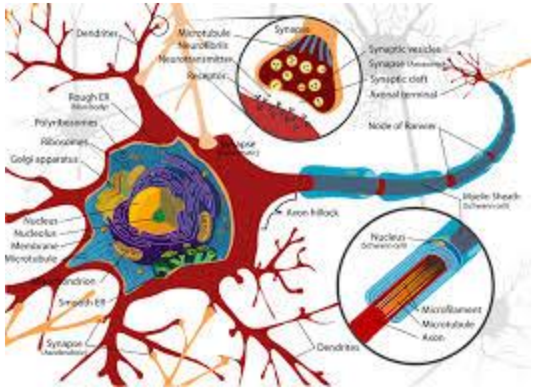
A train can hide another!

Spinal muscular atrophy



MONOGENIC EPILEPSIES





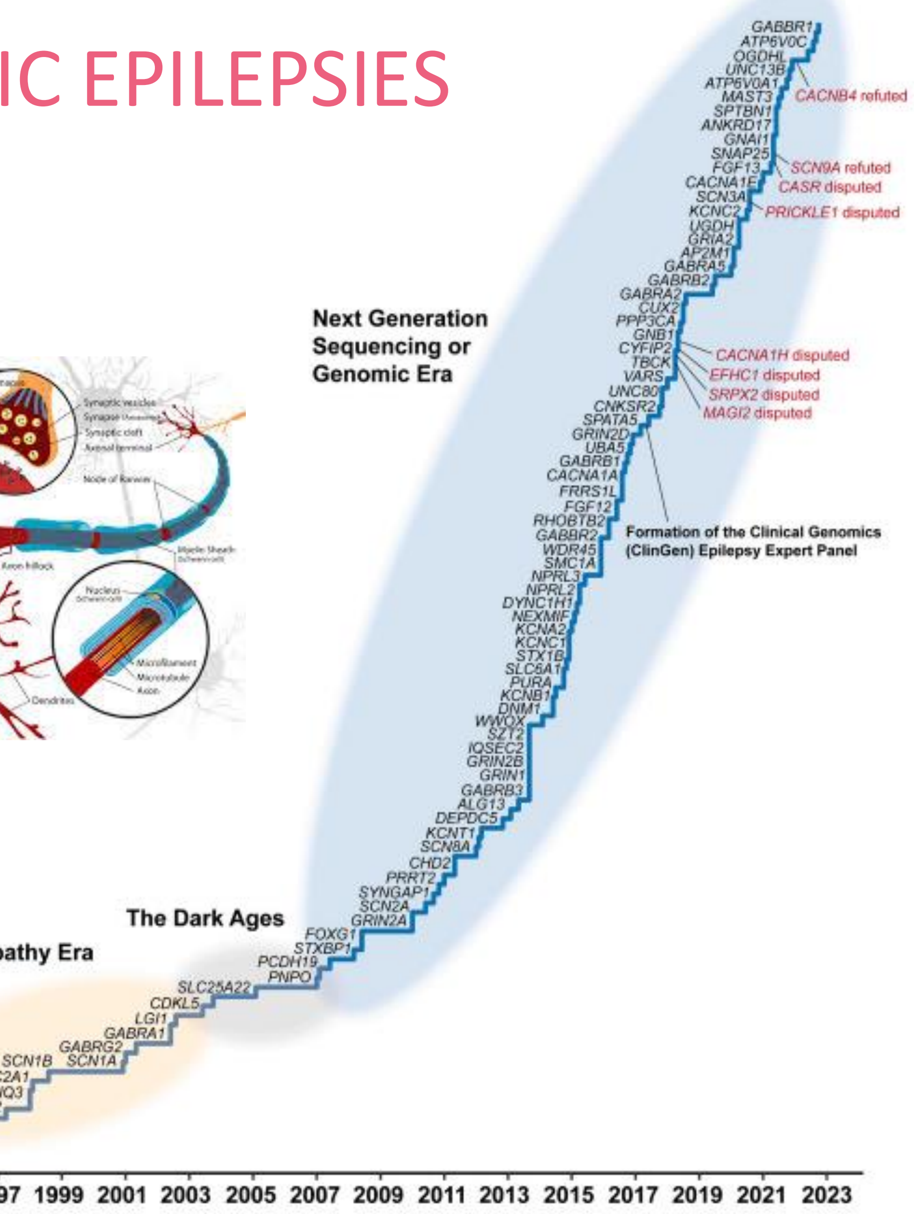


Figure from Scheffer IE, et al. Nat Rev Dis Primers. 2024;10:61 and Xian J and I Helbig, 2023

5 years to diagnosis… the brain does not wait!

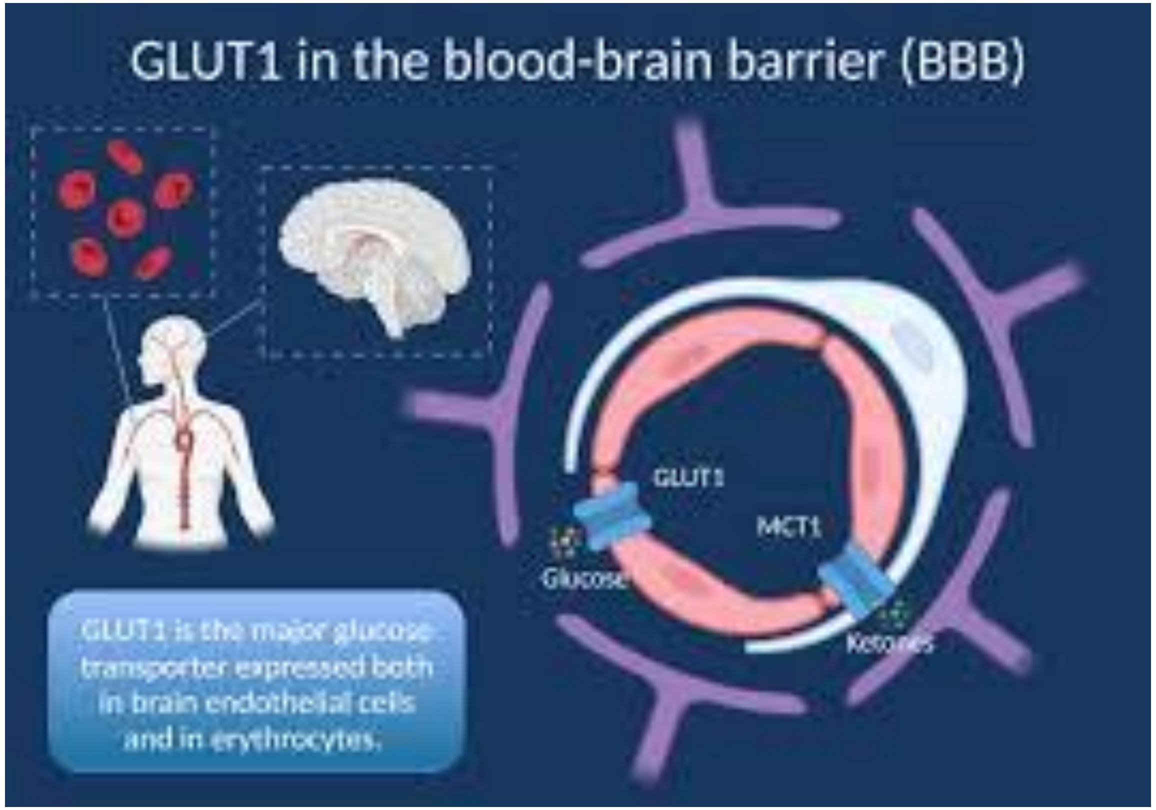
Abnormal eyes' movement
Ophtalmologist exam normal?

Absences (epilepsy), abnormal EEG and slight delay in the development?

- Absences non responding to drugs, more frequent in the morning
- Delay in the development
- Small head circumference
- Abnormal movement after awakening

10 years delay… the brain doaes not wait as much!

End of the why?
Beginning of the How!



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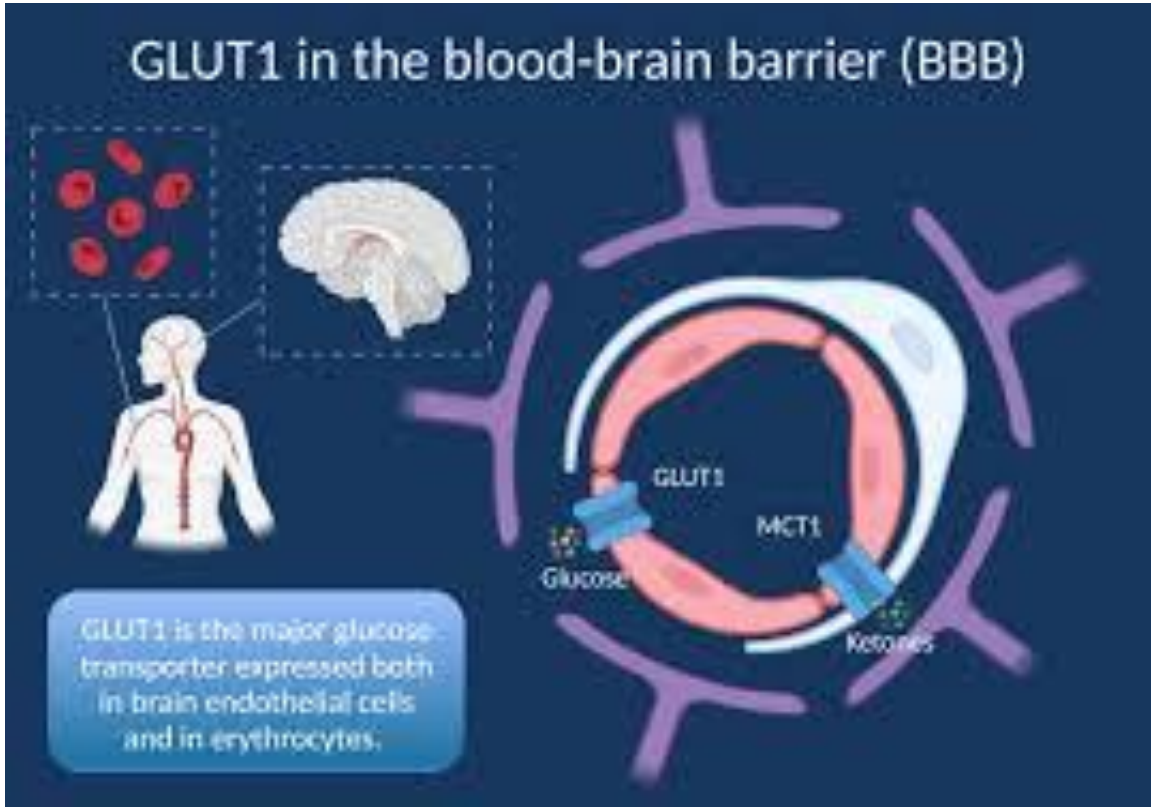
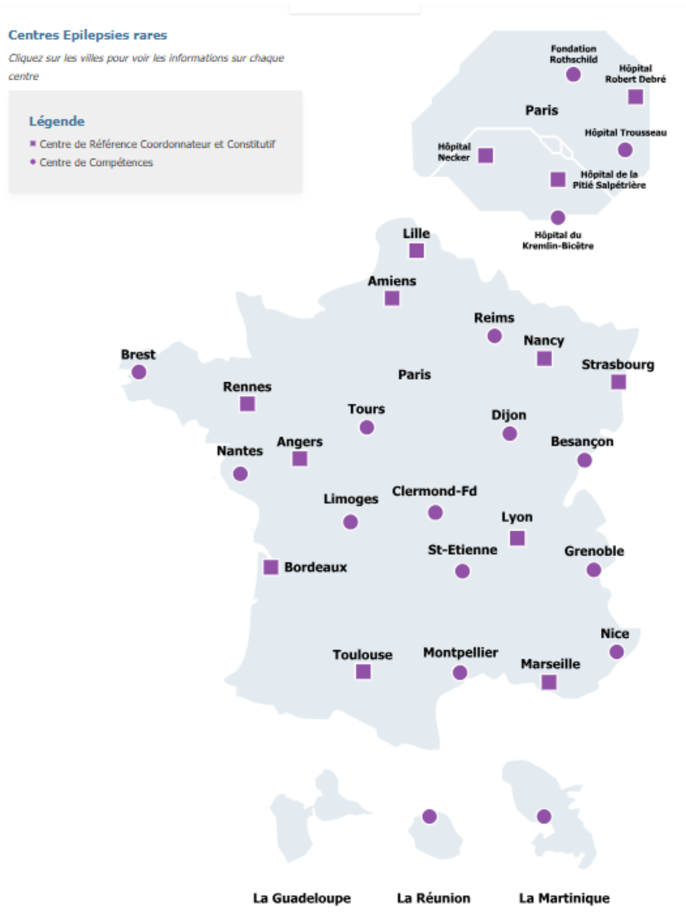
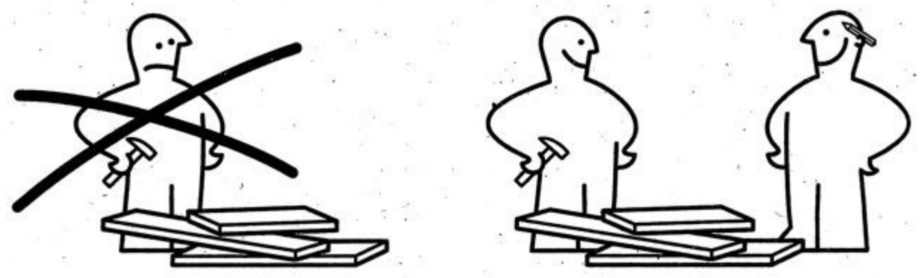


Case discussion meetings at national level






NORWAY Norwegian National Unit for Epilepsy, Oslo
SWEDEN Sahlgrenska University Hospital, Gothenburg
FINLAND Kuopio University Hospital / Consortium ERNEpi Helsinki-Oulu
THE NETHERLANDS University Medical Center Utrecht (UMCU)
BELGIUM University Hospital Gasthuisberg KUL, Leuven / BRACE Brussels Rare and Complex Epilepsies Consortium
LUXEMBOURG Centre Hospitalier de Luxembourg
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ROMANIA Alexandru Obregia Clinical Hospital, Bucharest
CYPRUS The Cyprus Institute of Neurology and Genetics, Nicosia
MALTA Mater Dei Hospital


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Objectives of ERNs: the example of EPICARE

Accessibility

To improve accessibility of detailed diagnostics to individuals of all ages with rare and complex epilepsies across Europe, including clinical evaluation and investigation.

Developing treatment

To develop treatment protocols and monitor standardised outcomes of rare and complex epilepsies.

Awareness

To improve awareness and accessibility to protocols for physicians and individuals with rare and complex epilepsies across Europe for treatment.

Education

To enhance educational activities and training opportunities across Europe by interchange across the network.

Collaborative research

To enhance opportunities for registries, and collaborative research for the benefit of individuals with rare and complex epilepsies across Europe.

Stand up for your rights!

Early diagnosis and best care!

Diagnosis	Targeted therapy	Accessibility
The right to have a diagnosis and a diagnosis etiology when possible!	The right to have the best care!	The right to have accessibility for this therapy wherever and whoever you are!

EDUCATION – DISSEMINATION – EXPERTISE and REFERRAL

Hurdles at each step

Need to empower physicians/patients/families

ERDERA

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Thank you!

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