ERN EpiCARE

A European Reference Network for Rare and Complex Epilepsies

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Network Epilepsies (ERN EpiCARE)



Disclosure



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I have no actual or potential conflict of interest in relation to this program/presentation

The Epilepsies



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A group of conditions characterised by tendency to recurrent epileptic seizures

Treatments

- Antiepileptic drugs
- Dietary treatment
- Stimulation

Epilepsy surgery (curative) in carefully selected individuals

Changing concepts: a group of rare diseases for which targeted treatment are possible

Early myoclonic encephalopathy PIGA, SETBP1, SIK1, SLC25A22

Dravet syndrome

SCN1A, GABRA1, GABRG2, HCN1, KCNA2,SCN1B, STXBP1

Lesional focal epilepsy

Non-lesional focal epilepsy ENFL1, ENFL2, LGI-1

Early-onset epileptic encephalopathy

KCNQ2, AARS, CACNA2D2, NECAP1, PIGA,

QARS, SCN8A

Early infantile ep

encephalopathy

(Ohtahara syndro

KCNT1, PIGQ

KCNQ2

CDKL5, SCN2A STXBP1, GNAO1 ARX, DOCK7, SLC25A22, SLC35A2, WWOX Infantile spasms

ALG13, DNM1, FOXG1 duplications, GABRA1,GABRB3, GRIN1, GRIN2A, GRIN2B, IQSEC2, KCNT1, MAGI2, MEF2C, NEDDL4, NDP, NRXN1, PIGA, PLCB1, PTEN, SCA2,SCN1A, SCN2A, SCN8A, SETBP1, SIK1, SLC25A22, SLC35A2, SPTAN1, ST3GAL3, STXBP1, TBC1D24, TCF4 Epilepsy with myoclonic-atonic seizures SLC2A1, SLC6A1, GABRA1, GABRG2, SCN1A, SCN1B

Rasmussen Syndrome , onset > 2 years

Lennox-Gastaut syndrome
ALG13, DNM1, FLNA, GABRB3, GLI3, HNRNPU, SCN1A,
SCN2A, SCN8A, STXBP1

Tuberous sclerosis complex
TSC1, TSC2, DEPDC5, GATOR1

Epilepsy–aphasia spectrum *GRIN2A*

Epilepsy of infancy with migrating focal seizures

KCNT1, SCN2A, SCN1A PLCB1, QARS, SCN8A, SLC25A22, TBC1D24, SLC12A5

Progressive myoclonic epilepsy EPM1, EPM2A, CLN

Other predominantly myoclonic epilepsies

Onset 0–1 years: EEF1A2, MEF2C, SCN1A, SLC2A1, SPTAN1, SYNGAP1, TBC1D24

Other predominantly myoclonic epilepsies, onset >1 year: CHD2, MEF2C, SYNGAP1, UBE3A

Other predominantly focal or multifocal epilepsies, onset 0-6 mon ARHGEF9, DEPDC5, SCN1A, TBC1D24, PNKP, SLC2A1 Other predominantly multifocal / focal epilepsies, onset 6-12 mon ARHGEF9, DEPDC5, FOXG1

Other predominantly focal or multifocal epilepsies, onset > 1 year ARHGEF9, DEPDC5, MBD5, PCDH19, POLG, TNK2, ZEB2

<6 mon 6-12 mon 1-4 y 5-10 y >11 y

Definitions





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Rare epilepsies: an epilepsy with incidence < 5 in 10,000 population, either defined as epilepsy syndrome or aetiologically driven epilepsy

Complex epilepsies: an epilepsy requiring multidisciplinary management through care pathway or comorbidity, with or without known aetiology eg surgically treatable epilepsies needing a high level of multidisciplinary expertise (including video-EEG analysis, functional and/or structural neuroimaging etc.)

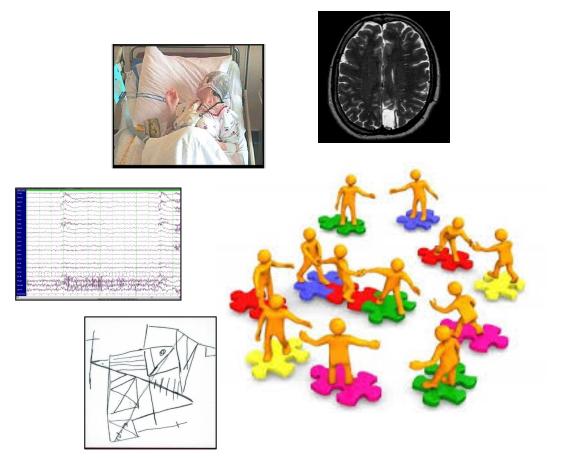




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multidisciplinary expertise









for rare or low prevalence complex diseases

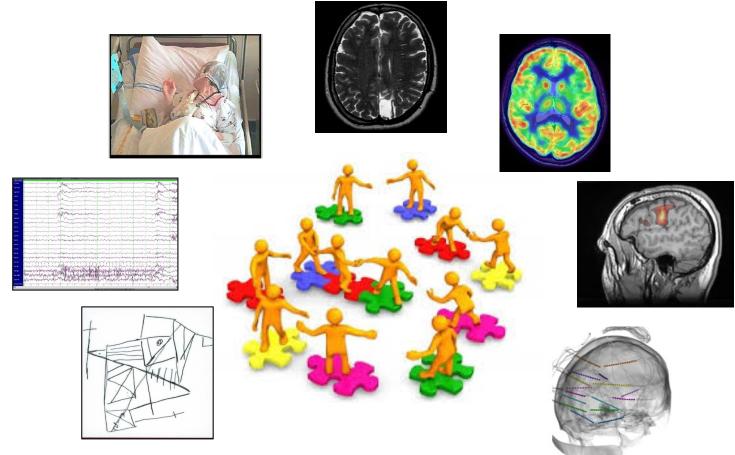
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complex diseases

Epilepsies (ERN EpiCARE)



multidisciplinary expertise

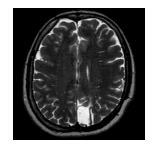


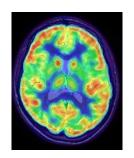


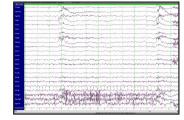
for rare or low prevalence complex diseases

Epilepsies (ERN EpiCARE)

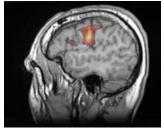


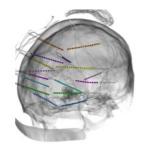
















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With gelastic seizures – ORPHA:86906

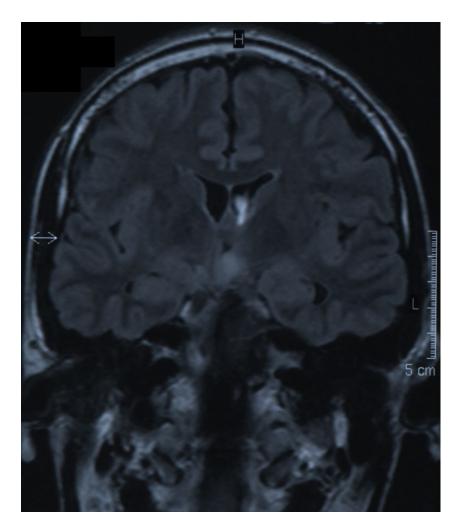
- non-neoplastic developmental malformation
- other seizure types including focal and generalized seizures
- concomitant cognitive decline and behavioral disorders
- some patients also present a precocious puberty



complex diseases

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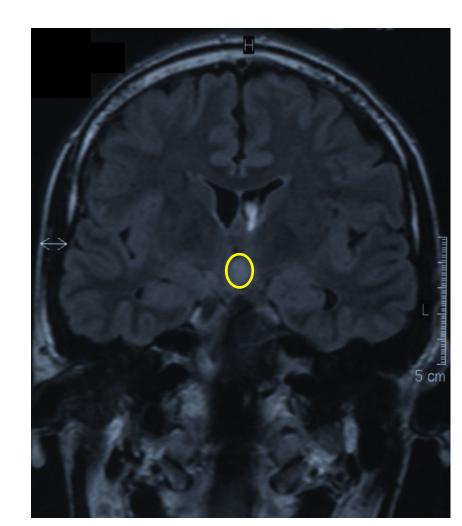
MRI – FLAIR, coronal section



Network

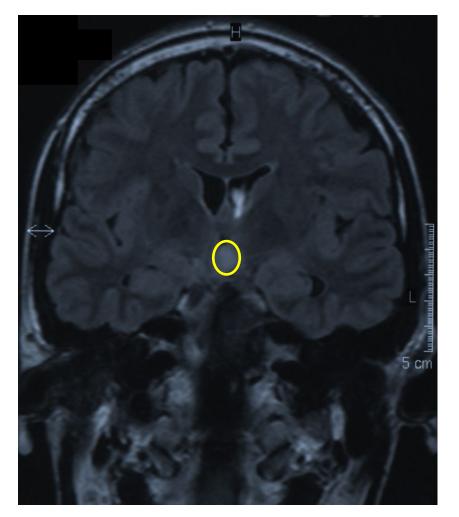


complex diseases



MRI – FLAIR, coronal section







Surgical resection

- transcallosal
- pterional
- endoscopic

Stereotactic

- thermoablation
- thermocoagulation

Gama Knife[©] radiosurgery

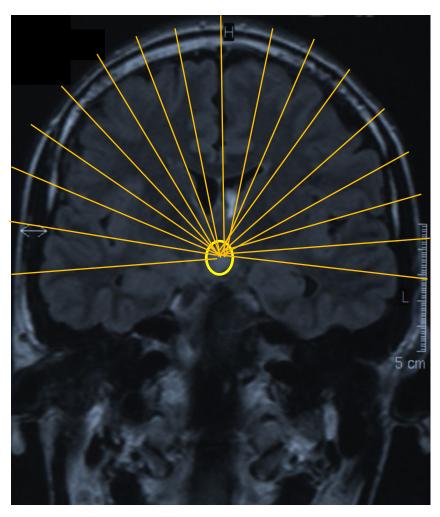


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MRI – FLAIR, coronal section

Gama Knife[©] radiosurgery

- many beams of radiation that converge on a single point
- individual beam is not powerful enough to destroy healthy tissue

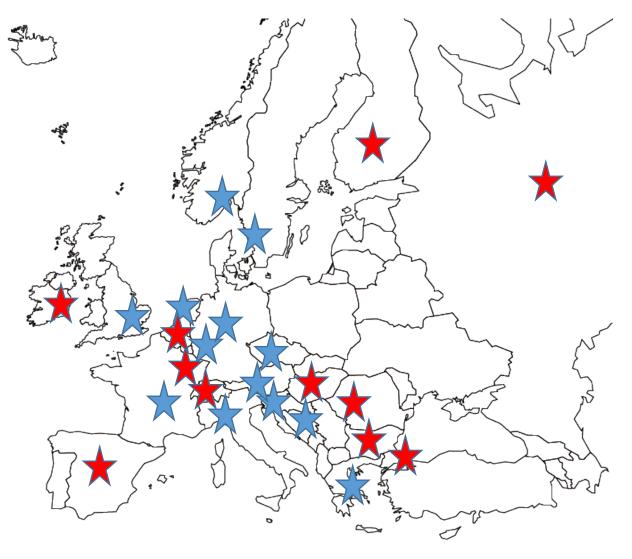




A pilot ERN: to raise awareness & accessibility of epilepsy surgery

13 members11 associatepartners

2013-2017



- Systematic reviews, guidelines and recommendations
- E-PILEPSY website
- E-Database
- E-Care platform (web-based epilepsy conference)
- E-Neuropsych
- E-Processing
- E-Eligibility
- Model of sustained development
- Exchange program
- Final conference





for rare or low prevalence complex diseases

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Summary EpiCARE showcase



NetworkEpilepsies (ERN EpiCARE)



The epilepsies: a group of rare diseases for which targeted treatment are possible

Vison: deliver highly-specialized care to improve diagnosis and outcome in individuals with rare and complex epilepsies

Multidisciplinary epilepsy surgery case discussions: European collaboration has already improved care for individual patients and their families

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A European Reference Network for Rare and Complex Epilepsies

J Helen Cross
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London



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