

Epilepsy Ontology

Aristea S. Galanopoulou MD PhD

Albert Einstein College of Medicine, Bronx NY

American Epilepsy Society, Research and Training Council co-chair

ICARE Ontology, considerations

- ICARE has been extremely valuable tool to the American Epilepsy Society in assessing the AES-funded research and visualizing where funds is allotted to.
- Discussions for possible updates of the ontology were prompted by realizing its utility and potential to evaluate funded research and recognizing that:
 - Current ontology terms were not encompassing all research areas and epilepsy/seizure types, etiologies.
 - Interim revisions in the working classification and coding of human epilepsies and seizures.
 - Distinction of human vs nonhuman, basic vs translational research is not easily/accurately derived from the current system. Having this would be very useful (different funding, design, expectations and expertise; could allow faster tracking of experts in clinical vs preclinical translational and basic science research).
 - Use of certain terms in various platforms (ICARE, Benchmarks, researchers) occasionally is done with variable meaning and could benefit of refinement.
 - Enhancement of the capability to query the database with more specific questions would greatly enhance strategic funding decisions and collaborations, attract more organizations to participate in ICARE and possibly eventually compare funded research to publications.

iCARE

ICARE Portfolio Data Contributors



National Institutes of Health



Centers for Disease Control



United States Department
of Veterans Affairs



Health Resources and
Services Administration



Citizens United for Research
in Epilepsy



American Epilepsy Society



Epilepsy Foundation



Dravet Syndrome
Foundation



Tuberous Sclerosis Alliance



PCDH19 Alliance



Phelan-McDermid
Syndrome Foundation



LGS Foundation



Patient-Centered Outcomes
Research Institute



Pediatric Epilepsy Research
Foundation

ADEAF - Autosomal Dominant Epilepsy with
Auditory Features
ADNFLE - Autosomal-Dominant Nocturnal Frontal
Lobe Epilepsy
Alpers Syndrome
Angelman Syndrome
BECTS - Benign Epilepsy with Centrotemporal
Spikes
BFNE - Benign Familial Neonatal Epilepsy
CAE - Childhood Absence Epilepsy
Catamenial Seizures
Childhood Epilepsy
Dravet Syndrome
Early Life Seizures
EME - Early Myoclonic Encephalopathy
Encephalitis Acquired Epilepsy
Epilepsy/Seizures associated with other disorders
(like Alzheimer's, Autism, Fragile X, Malaria, ...)
Epilepsy/Seizures in pregnant women
Epilepsy/Seizures in the elderly
Epileptic Encephalopathies
Febrile Seizures
Focal Epilepsy
GEFS+ - Generalized Epilepsy with Febrile Seizures
plus
Genetic Epilepsy
Hemiconvulsion–Hemiplegia–Epilepsy
Hypothalamic Hamartoma with Gelastic Seizures
IS - Infantile Spasms
JAE - Juvenile Absence Epilepsy
JME - Juvenile Myoclonic Epilepsy
KCNQ2 Encephalopathy

Lafora Disease
LGS - Lennox -Gastaut Syndrome
LKS - Landau-Kleffner syndrome
Malformations of Cortical Development
Neonatal Seizures
Neurocysticercosis
Nodding Syndrome
Non-Epileptic Seizures
Ohtahara Syndrome
PCDH19 Epilepsy
PME - Progressive Myoclonus Epilepsies
PMSE - Polyhydramnios, Megalencephaly and
Symptomatic Epilepsy Syndrome
PTE - Post Traumatic Epilepsy
Rasmussen Syndrome
Reflex Epilepsies
Seizures
Status Epilepticus
Sturge-Weber Syndrome
Succinic Semialdehyde Dehydrogenase Deficiency
SUDEP
TLE - Temporal Lobe Epilepsy
TSC - Tuberous Sclerosis Complex
West Syndrome
Epilepsy - not otherwise specified

ICARE Search

Search Results

— ADJUST YOUR SEARCH CRITERIA

Full Text Search

epileptogenesis

Funding Organization

Project Number

Funding Year(FY)

Choose some options

Funding Agency/Organization

Choose some options

Researcher and Awardee Organization

Principal Investigator (PI)

PI Organization

Research Categories

Research Type

Choose some options

Research Classification

Choose some options

Epilepsy or Seizure condition

Choose some options

NINDS Epilepsy Research Benchmarks

Benchmark Area

Choose some options

Search

Clear

ICARE search terms and classifications

YOUR SEARCH CRITERIA: FULL TEXT: epileptogenesis

FURTHER REFINE SEARCH RESULTS

Funding Year (FY) ?

- ☐ 2013 (202)
- ☐ 2014 (220)
- ☐ 2015 (208)
- ☐ 2016 (208)

Funding Agency ?

- ☐ NIH (640)
- ☐ CURE (76)
- ☐ AES (35)
- ☐ Veterans Affairs (35)
- ☐ Dravet Syndrome Foundation (11)
- ☐ Phelan-McDermid Syndrome Foundation (11)
- ☐ DOD (10)
- ☐ TS Alliance (8)
- ☐ EF (7)
- ☐ Wishes for Elliott (2)
- ☐ Batten Disease Support and Research Association (1)
- ☐ LGS Foundation (1)
- ☐ PCORI (1)

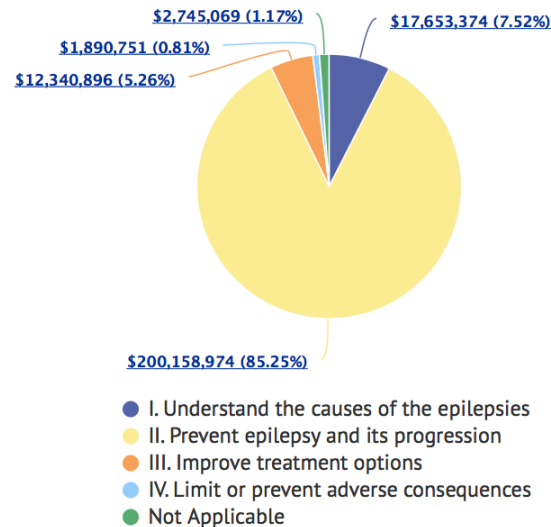
Visualizing data for 838 Total Projects

[View Results](#)

[Export](#)

Distribution of Funding by Benchmark

Click on a Benchmark Area slice to view the distribution of funding by Individual Benchmark.



Funding by Research Classification

Research Association (1)

- ☐ LGS Foundation (1)
- ☐ PCORI (1)

Research Type ?

- ☐ Basic (616)
- ☐ Clinical (60)
- ☐ Translational (162)

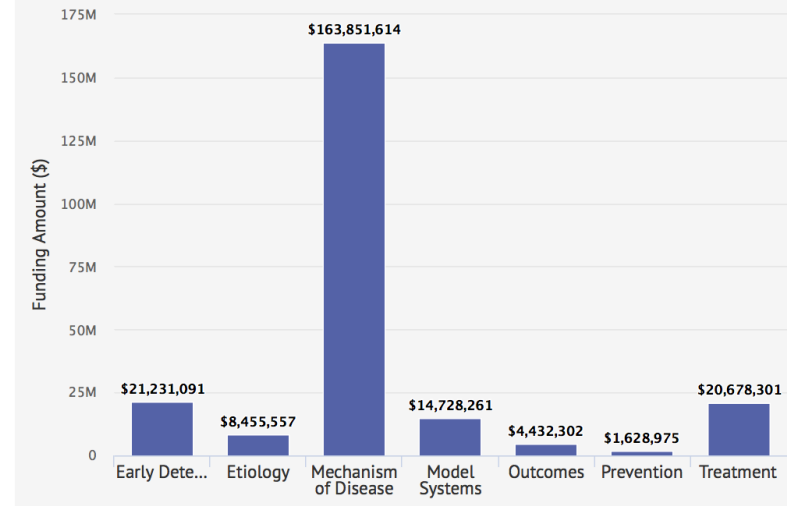
Research Classification ?

- ☐ Early Detection / Diagnosis / Prognosis (70)
- ☐ Etiology (23)
- ☐ Mechanism of Disease (557)
- ☐ Model Systems (76)
- ☐ Outcomes (11)
- ☐ Prevention (17)
- ☐ Treatment (84)

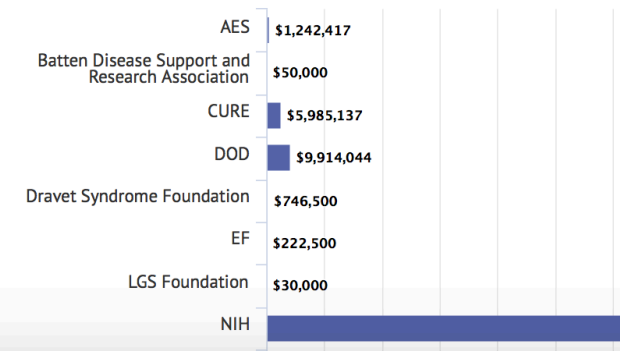
Condition ?

- ☐ TLE - Temporal Lobe Epilepsy (163)
- ☐ PTE - Post Traumatic Epilepsy (129)
- ☐ Epilepsy - not otherwise specified (117)
- ☐ Genetic Epilepsy (60)
- ☐ Dravet Syndrome (43)
- ☐ Epileptic Encephalopathies (30)
- ☐ Epilepsy/Seizures associated with other disorders (like Alzheimer's, Autism, Fragile X, Malaria, ...) (28)
- ☐ TSC - Tuberous Sclerosis

Funding by Research Classification



Total Funding by Organization



Benchmark ?

- ☐ I. Understand the causes of the epilepsies (35)
- ☐ II. Prevent epilepsy and its progression (702)
- ☐ III. Improve treatment options (75)
- ☐ IV. Limit or prevent adverse consequences (20)
- ☐ Not Applicable (5)

Research Type	
Basic	Basic research is the systematic study of the fundamental aspects of phenomena and of observable facts without specific development of processes, products or clinical applications. Projects typically include studies of the mechanisms of normal or disease related processes at the molecular, cellular, systems or organ level.
Translational	Translational research is the process of developing ideas, insights, and discoveries generated through basic scientific inquiry for the treatment or prevention of human disease.
Clinical	<i>Patient-oriented research.</i> <u>Research conducted with human subjects</u> (or on material of human origin such as tissues, specimens and cognitive phenomena) for which an investigator directly interacts with human subjects. <u>Excluded from this definition are in vitro studies that utilize human tissues that cannot be linked to a living individual.</u> Patient-oriented research typically includes therapeutic interventions and applications of new technologies, clinical trials, epidemiologic and behavioral studies, outcomes research and health services research.

1. Separating research in humans vs in animals / models

Issues:

- Basic and translational research may utilize animal/model systems or human subjects which may confound the reporting of funded research in each of these categories.
- Research using human tissue is not always clinical.
- Using only keywords for search for human vs animal/model research may not sufficiently differentiate the two different types of research (keyword hits are not always specific for keywords).

Suggestions:

Separating the two types of research may help visualize and compare more directly:

- the value, productivity, and results of animal/model vs human epilepsy research
- Expertise in animal vs human research

→ Suggest to create Refine Search Criteria for:

Organism/Model:

- Nonhuman organism
- Human
- Other model system

Research Type

2. Improving distinction of basic and translational research

Issues:

Expectations from basic vs translational (preclinical) research may be different in terms of grant review, study design and performance.

Distinction is not always clear producing overlap of hits when using the current system.

Suggestions:

→ May consider more specific definitions of how to differentiate and log basic vs translational research.

Research classification

Classification	Definitions
Etiology	Research included in this category aims to identify the <u>causes</u> or origins of epilepsy - genetic, infectious, metabolic, environmental, or other factors, and the interactions between these factors
Mechanism of Disease	Research included in this category looks at the biology of <u>how epilepsy/seizures starts and progresses</u> as well as normal biology relevant to these processes
Prevention	Research included in this category looks at <u>identifying interventions which reduce the risk of developing epilepsy</u> by reducing exposure to risk factors and/or increasing protective factors. Interventions aimed at prevention of <u>complications of epilepsy or its co-occurring conditions</u> may also be included. Interventions may target lifestyle or may involve drugs or vaccines
Early Detection/ Diagnosis/Prognosis	Research included in this category focuses on <u>identifying and testing biomarkers, technology methods or predictive models</u> that are helpful in detecting and/or diagnosing as well as predicting the outcome or chance of recurrence
Treatment	Research included in this category focuses on <u>identifying and testing treatments</u> , such as novel therapeutics, devices or other interventions.
Outcomes	Research included in this category includes a broad range of areas: surveillance and epidemiology; ethics, education and communication approaches for health care professionals, patients and families, and community members; patient care and health care services research; effectiveness research and phase 4 trials
Model Systems	Research included in this category looks at the development of new animal models, cell cultures and computer simulations and their application to other studies across the spectrum of epilepsy research

Funding Year (FY) ?

- ☐ 2013 (19)
- ☐ 2014 (23)
- ☐ 2015 (26)
- ☐ 2016 (30)

Funding Agency ?

- ☒ AES
- ☐ NIH (1620)
- ☐ CURE (191)
- ☐ EF (83)
- ☐ Veterans Affairs (59)
- ☐ CDC (41)
- ☐ Dravet Syndrome Foundation (36)
- ☐ HRSA (32)
- ☐ TS Alliance (25)
- ☐ DOD (18)
- ☐ Pediatric Epilepsy Research Foundation (16)
- ☐ Phelan-McDermid Syndrome Foundation (12)
- ☐ PCORI (8)
- ☐ LGS Foundation (6)
- ☐ Wishes for Elliott (4)
- ☐ Batten Disease Support and Research Association (2)
- ☐ Dup15q Alliance (2)
- ☐ Epilepsy Study Consortium (2)
- ☐ IFCR (2)
- ☐ PCDH19 Alliance (2)

+ Show more

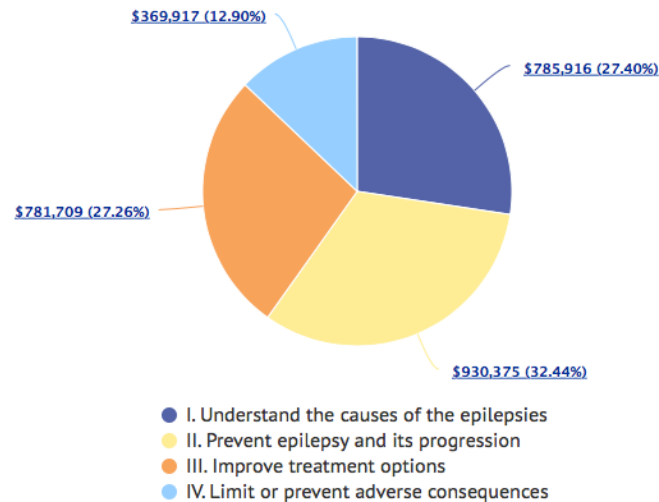
Research Type ?

- ☐ Basic (34)
- ☐ Clinical (27)
- ☐ Translational (37)

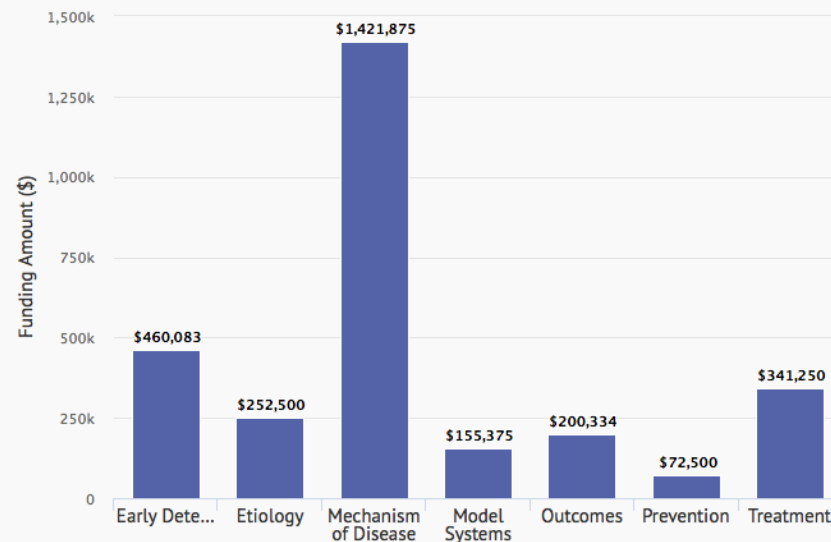
Research Classification ?

Distribution of Funding by Benchmark

Click on a Benchmark Area slice to view the distribution of funding by Individual Benchmark.



Funding by Research Classification



Research Classification

Same terms across ICARE, Epilepsy benchmarks epilepsy researchers are not used with the same meaning

Examples:

Prevention of epilepsy / co—occurring conditions and consequences:

- ICARE: intervention-oriented research.
- Benchmarks
 - II: includes mechanisms, biomarkers, interventions
 - I, III and IV: may also address prevention

Prevention research, AES-funded

Prevention Research:~145K

FURTHER REFINE SEARCH RESULTS

Funding Year (FY)

- ☐ 2014 (2)
- ☐ 2015 (4)

Funding Agency

- ☒ AES
- ☐ NIH (20)
- ☐ CURE (11)
- ☐ EF (3)
- ☐ Pediatric Epilepsy Research Foundation (1)
- ☐ Veterans Affairs (1)

Research Type

- ☐ Basic (4)
- ☐ Translational (2)

Research Classification

- ☒ Prevention

Condition

- ☐ TLE - Temporal Lobe Epilepsy (2)
- ☐ Epilepsy - not otherwise specified (1)
- ☐ Focal Epilepsy (1)
- ☐ Seizures (1)
- ☐ West Syndrome (1)

Benchmark

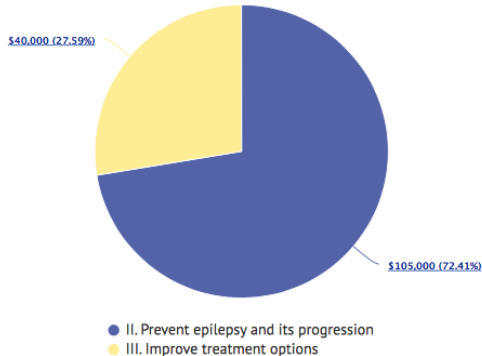
- ☐ II. Prevent epilepsy and its progression (4)
- ☐ III. Improve treatment options (2)

Visualizing data for 6 Total Projects

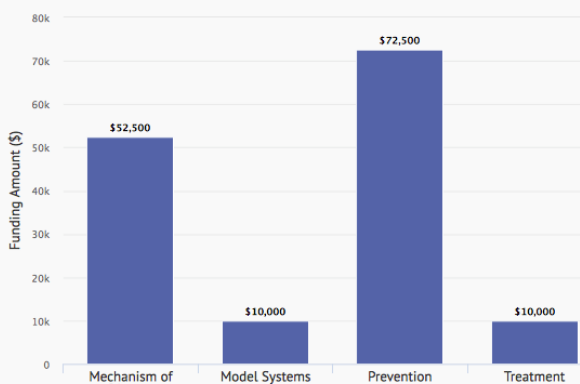
[View Results](#) [Export Data \(CSV\)](#)

Distribution of Funding by Benchmark

Click on a Benchmark Area slice to view the distribution of funding by Individual Benchmark.



Funding by Research Classification



Funding Year (FY)

- ☐ 2013 (9)
- ☐ 2014 (7)
- ☐ 2015 (5)
- ☐ 2016 (3)

Funding Agency

- ☒ AES
- ☐ NIH (590)
- ☐ CURE (72)
- ☐ TS Alliance (7)
- ☐ EF (5)
- ☐ Veterans Affairs (5)
- ☐ Dravet Syndrome Foundation (4)
- ☐ Batten Disease Support and Research Association (1)
- ☐ Child Neurology Foundation (1)
- ☐ DOD (1)
- ☐ LGS Foundation (1)

Research Type

- ☐ Basic (15)
- ☐ Clinical (3)
- ☐ Translational (6)

Research Classification

- ☐ Early Detection / Diagnosis / Prognosis (3)
- ☐ Mechanism of Disease (13)
- ☐ Model Systems (1)
- ☐ Prevention (4)
- ☐ Treatment (3)

Condition

- ☐ TLE - Temporal Lobe Epilepsy (5)
- ☐ Dravet Syndrome (3)
- ☐ Status Epilepticus (3)
- ☐ CAE - Childhood Absence Epilepsy (2)
- ☐ West Syndrome (2)
- ☐ Angelman Syndrome (1)
- ☐ Early Life Seizures (1)
- ☐ Encephalitis Acquired Epilepsy (1)
- ☐ Epilepsy - not otherwise specified (1)
- ☐ Epilepsy/Seizures in the elderly (1)

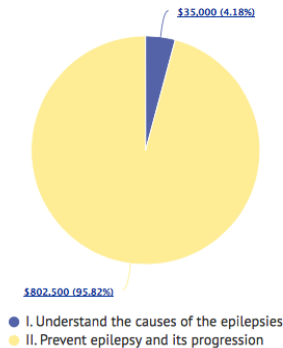
Benchmark

- ☒ II. Prevent epilepsy and its progression

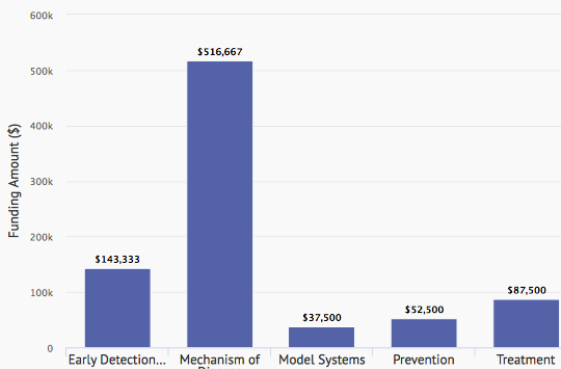
Benchmark II: ~837.5K

Distribution of Funding by Benchmark

Click on a Benchmark Area slice to view the distribution of funding by Individual Benchmark.



Funding by Research Classification



Total Funding by Organization



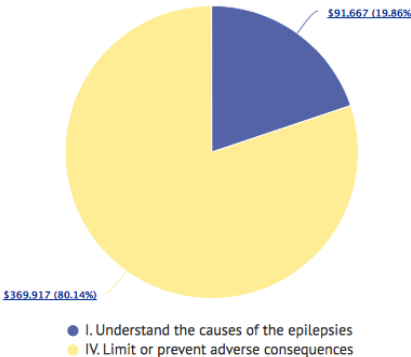
Benchmark IV:~461K

Visualizing data for 15 Total Projects

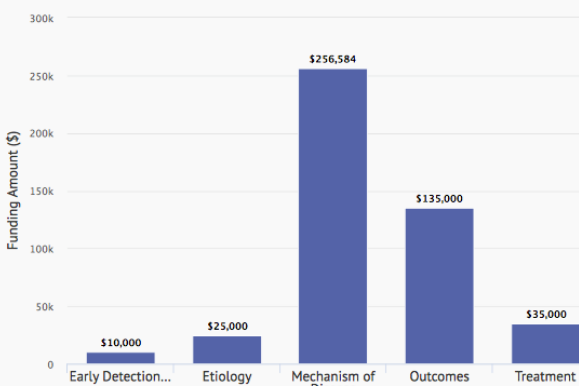
[View Results](#) [Export Data \(CSV\)](#)

Distribution of Funding by Benchmark

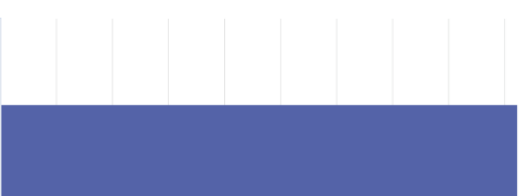
Click on a Benchmark Area slice to view the distribution of funding by Individual Benchmark.



Funding by Research Classification



Total Funding by Organization



FURTHER REFINE SEARCH RESULTS

Funding Year (FY)

- ☐ 2013 (3)
- ☐ 2014 (3)
- ☐ 2015 (4)
- ☐ 2016 (5)

Funding Agency

- ☒ AES
- ☐ NIH (197)
- ☐ CURE (28)
- ☐ CDC (24)
- ☐ EF (21)
- ☐ Veterans Affairs (12)
- ☐ Pediatric Epilepsy Research Foundation (8)
- ☐ Phelan-McDermid Syndrome Foundation (8)
- ☐ DOD (3)
- ☐ Dravet Syndrome Foundation (3)
- ☐ Wishes for Elliott (1)

Research Type

- ☐ Basic (2)
- ☐ Clinical (9)
- ☐ Translational (4)

Research Classification

- ☐ Early Detection / Diagnosis / Prognosis (1)
- ☐ Etiology (1)
- ☐ Mechanism of Disease (8)
- ☐ Outcomes (4)
- ☐ Treatment (1)

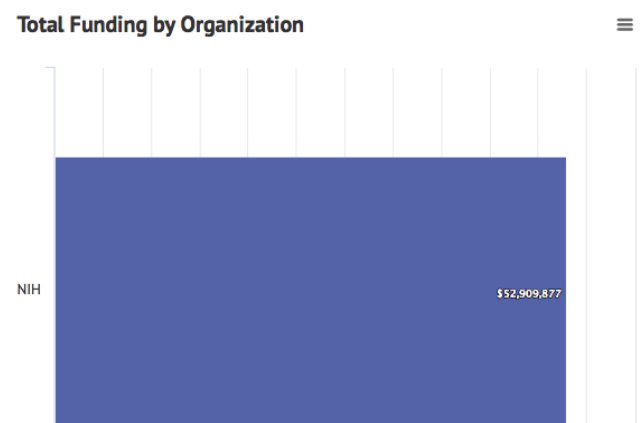
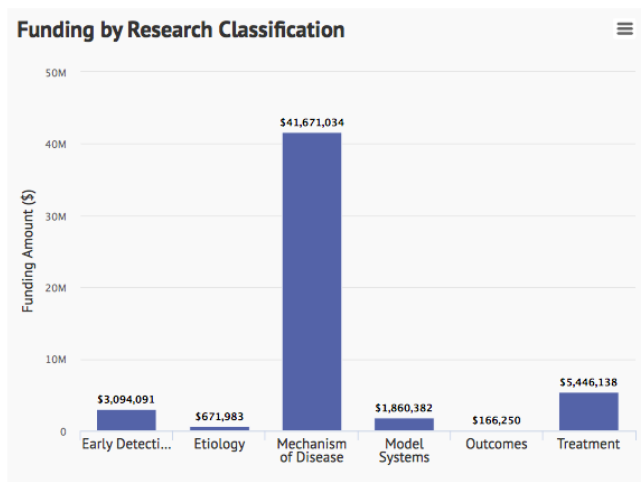
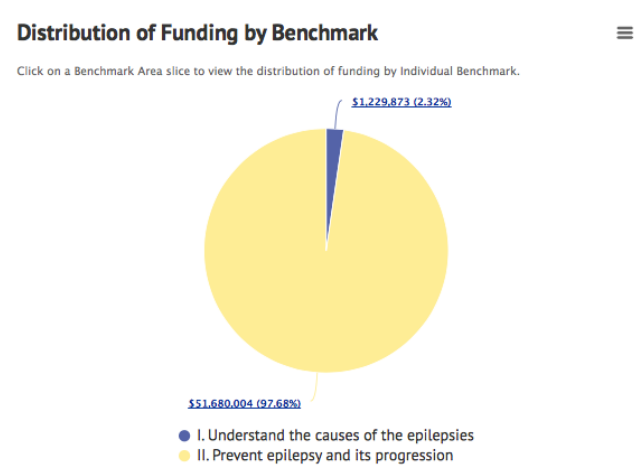
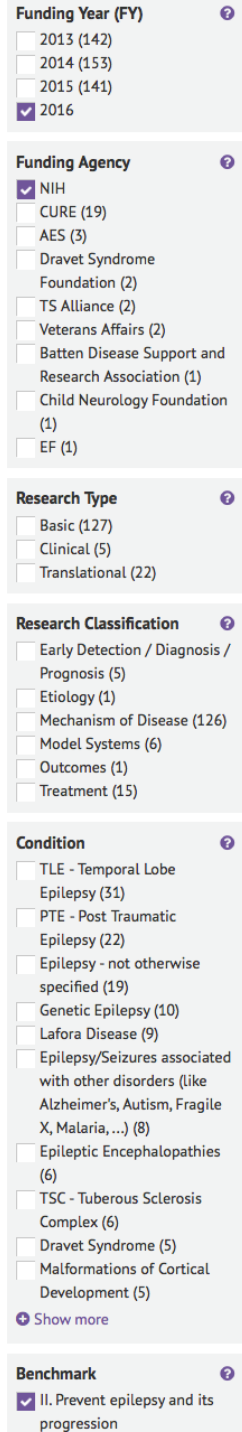
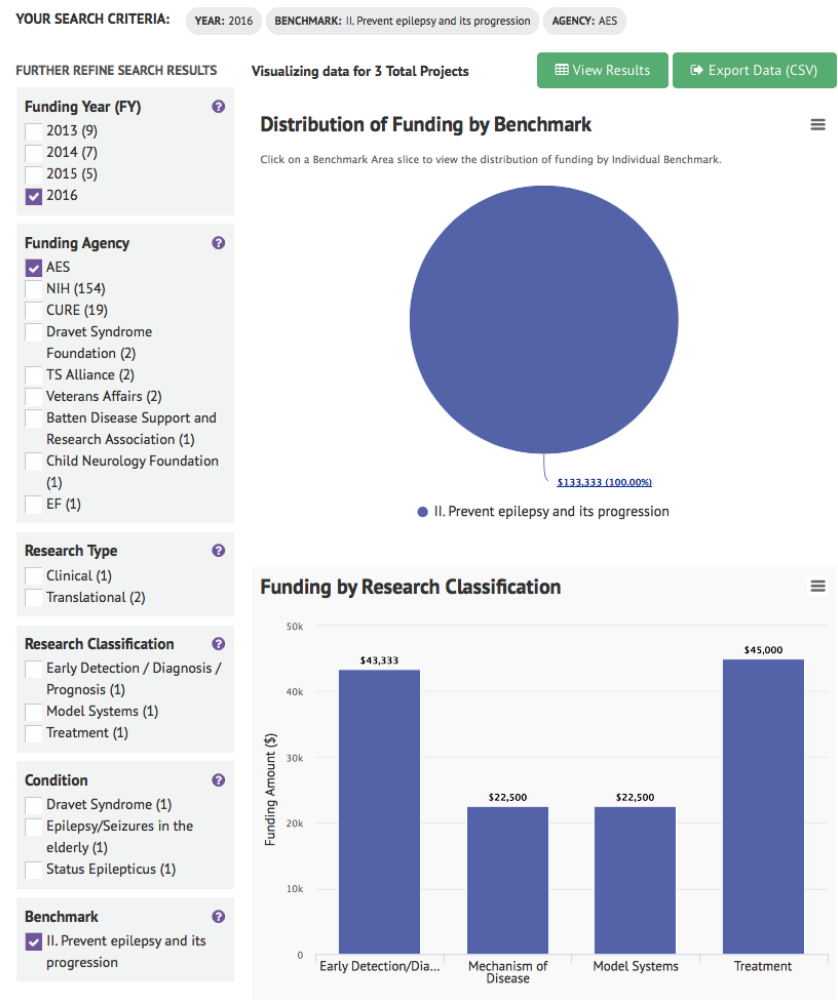
Condition

- ☐ Epilepsy - not otherwise specified (4)
- ☐ TLE - Temporal Lobe Epilepsy (4)
- ☐ Epilepsy/Seizures in pregnant women (2)
- ☐ SUDEP (2)
- ☐ Epilepsy/Seizures in the elderly (1)
- ☐ Epileptic Encephalopathies (1)
- ☐ Status Epilepticus (1)

Benchmark

- ☒ IV. Limit or prevent adverse consequences

Benchmark II funded research: Prevent epilepsy and its progression (2016) → no hits for prevention



Research Classification

Same terms across ICARE, Epilepsy benchmarks and in epilepsy research are not always used with the same meaning

Example: prevention of epilepsy / co—occurring conditions and consequences:

- ICARE: intervention-oriented research
- Epilepsy Benchmarks
 - II: includes mechanisms, biomarkers, interventions
 - III and IV: may also address prevention
- Search hits may not always capture the research classification done, as coded

Suggestions:

- Recoding may probably not be the best solution, since each coding method has its advantages and different information
- Perhaps:
 - More specific terms coding research classification to track key areas of prevention research (e.g, anti-epileptogenesis, disease modification, etc) ?
 - Refining search tools by allowing to select or exclude classifications or search keywords (AND, OR, NOT) ?

Epilepsy syndromes, seizures, special populations, consequences

SYNDROMES

Dravet Syndrome
EME - Early Myoclonic Encephalopathy
Epileptic Encephalopathies
Hemiconvulsion–Hemiplegia–Epilepsy
IS - Infantile Spasms
West syndrome
LGS - Lennox -Gastaut Syndrome
LKS - Landau Kleffner syndrome
Ohtahara Syndrome
PTE - Post Traumatic Epilepsy
Rasmussen Syndrome
Nodding Syndrome

SEIZURES

Epilepsy/Seizures associated with other disorders (like Alzheimer's, Autism, Fragile X, Malaria, ...)
Febrile Seizures
Non-Epileptic Seizures
Seizures
Status Epilepticus

SPECIAL POPULATIONS

Early Life Seizures
Neonatal Seizures
Childhood Epilepsy
Epilepsy/Seizures in pregnant women
Catamenial Seizures
Epilepsy/Seizures in the elderly

CONSEQUENCES

SUDEP

Condition: Current epilepsy ontology

GENETIC or GENETIC-STRUCTURAL

Genetic Epilepsy

ADEAF - Autosomal Dominant Epilepsy with Auditory Features

ADNFLE - Autosomal-Dominant Nocturnal Frontal Lobe Epilepsy

BECTS - Benign Epilepsy with Centrotemporal Spikes

BFNE - Benign Familial Neonatal Epilepsy

CAE - Childhood Absence Epilepsy

KCNQ2 Encephalopathy

Lafora Disease

PCDH19 Epilepsy

TSC - Tuberous Sclerosis Complex

PME - Progressive Myoclonus Epilepsies

PMSE – Polyhydramnios, Megalencephaly and Symptomatic Epilepsy

Reflex Epilepsies

GEFS+ - Generalized Epilepsy with Febrile Seizures plus

Alpers syndrome

ACQUIRED

Encephalitis Acquired Epilepsy

Hypothalamic Hamartoma with Gelastic Seizures

FOCAL vs GENERALIZED

Focal Epilepsy

TLE - Temporal Lobe Epilepsy

OTHER EPILEPSIES

Epilepsy - not otherwise specified
Epilepsy/Seizures associated with other disorders (like Alzheimer's, Autism, Fragile X, Malaria, ...)

ETIOLOGY

Malformations of Cortical Development

Neurocysticercosis

Succinic Semialdehyde

Dehydrogenase Deficiency

Not complete list of epilepsies or etiologies

- Only TLE among focal epilepsies
- Focal but no generalized epilepsy coding

Some epilepsies are represented by specific etiologies only, eg

- ADNFLE (no FLE)

Etiologies are not systematically captured or listed in the same manner, eg

- metabolic etiologies
- neurocysticercosis vs epilepsy/seizures associated with other disorders

Some names have been revised or may have additional variations of names, eg

- benign vs self-limited
- GEFS+: *Genetic*....

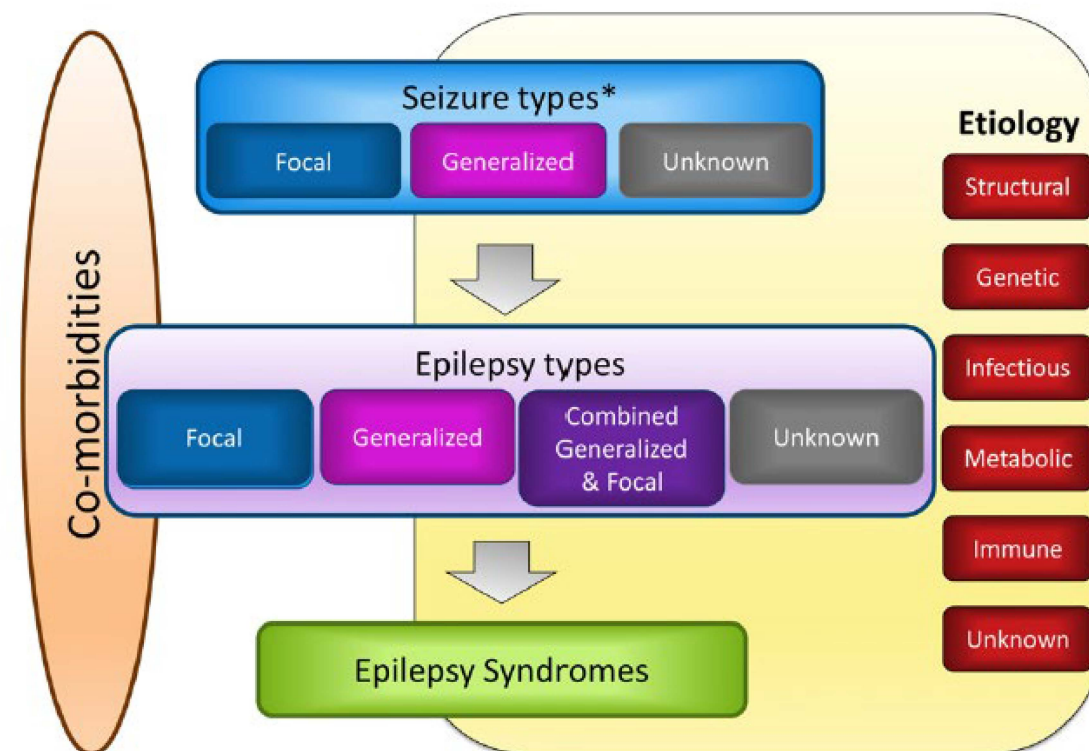
No coding for comorbidities, co-occurring conditions

ILAE classification of the epilepsies: Position paper of the ILAE Commission for Classification and Terminology

^{1,2,3}Ingrid E. Scheffer, ¹Samuel Berkovic, ⁴Giuseppe Capovilla, ⁵Mary B. Connolly, ⁶Jacqueline French, ⁷Laura Guilhoto, ^{8,9}Edouard Hirsch, ¹⁰Satish Jain, ¹¹Gary W. Mathern, ¹²Solomon L. Moshé, ¹³Douglas R. Nordli, ¹⁴Emilio Perucca, ¹⁵Torbjörn Tomson, ¹⁶Samuel Wiebe, ¹⁷Yue-Hua Zhang, and ^{18,19}Sameer M. Zuberi

Epilepsia, 58(4):512–521, 2017
doi: 10.1111/epi.13709

Classification of the Epilepsies



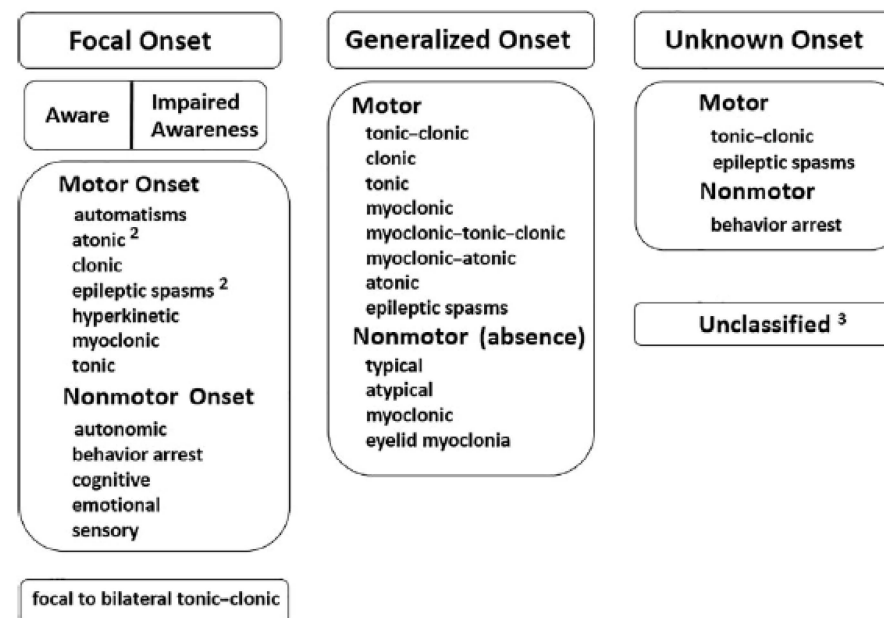
Operational classification of seizure types by the International League Against Epilepsy: Position Paper of the ILAE Commission for Classification and Terminology

*Robert S. Fisher, †J. Helen Cross, ‡Jacqueline A. French, §Norimichi Higurashi, ¶Edouard Hirsch, #Floor E. Jansen, **Lieven Lagae, ††Solomon L. Moshé, ‡‡Jukka Peltola, §§Eliane Roulet Perez, ¶¶Ingrid E. Scheffer, and ###**Sameer M. Zuberi

Epilepsia, 58(4):522–530, 2017
doi: 10.1111/epi.13670

Operational Classification of Seizure Types

ILAE 2017 Classification of Seizure Types Expanded Version ¹



EpilepsyDiagnosis.org



International League
Against Epilepsy



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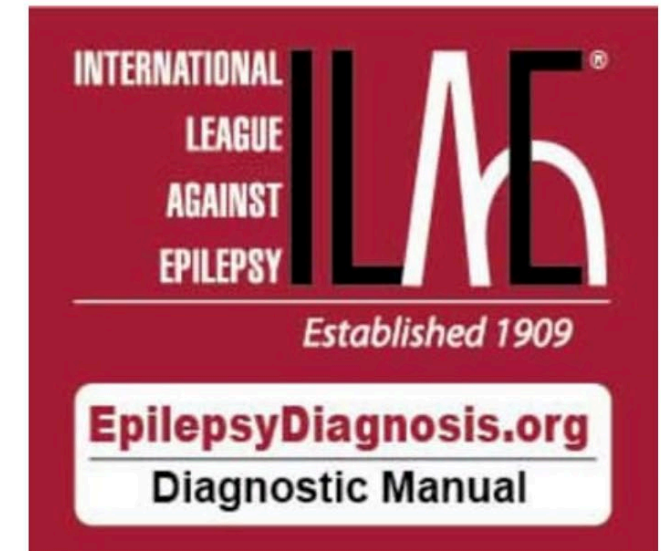
EpilepsyDiagnosis.org

Explore EpilepsyDiagnosis.org

The ILAE Commission on Classification and Terminology is pleased to announce the release of **EpilepsyDiagnosis.org** a cutting-edge online diagnostic manual of the epilepsies.

Manual Goal:

The goal of EpilepsyDiagnosis.org is to make available, in an easy to understand form, the latest concepts relating to seizures and the epilepsies. The principal goal is to assist clinicians who look after people with epilepsy anywhere in the world to diagnose seizure type(s), classify epilepsy, diagnose epilepsy



(from <https://www.ilae.org/education/diagnostic-manual/epilepsydiagnosis-org>)

Etiology

Most of the funded research addresses mechanisms / etiologies and yet the codes for such research are minimal and not systematically captured.

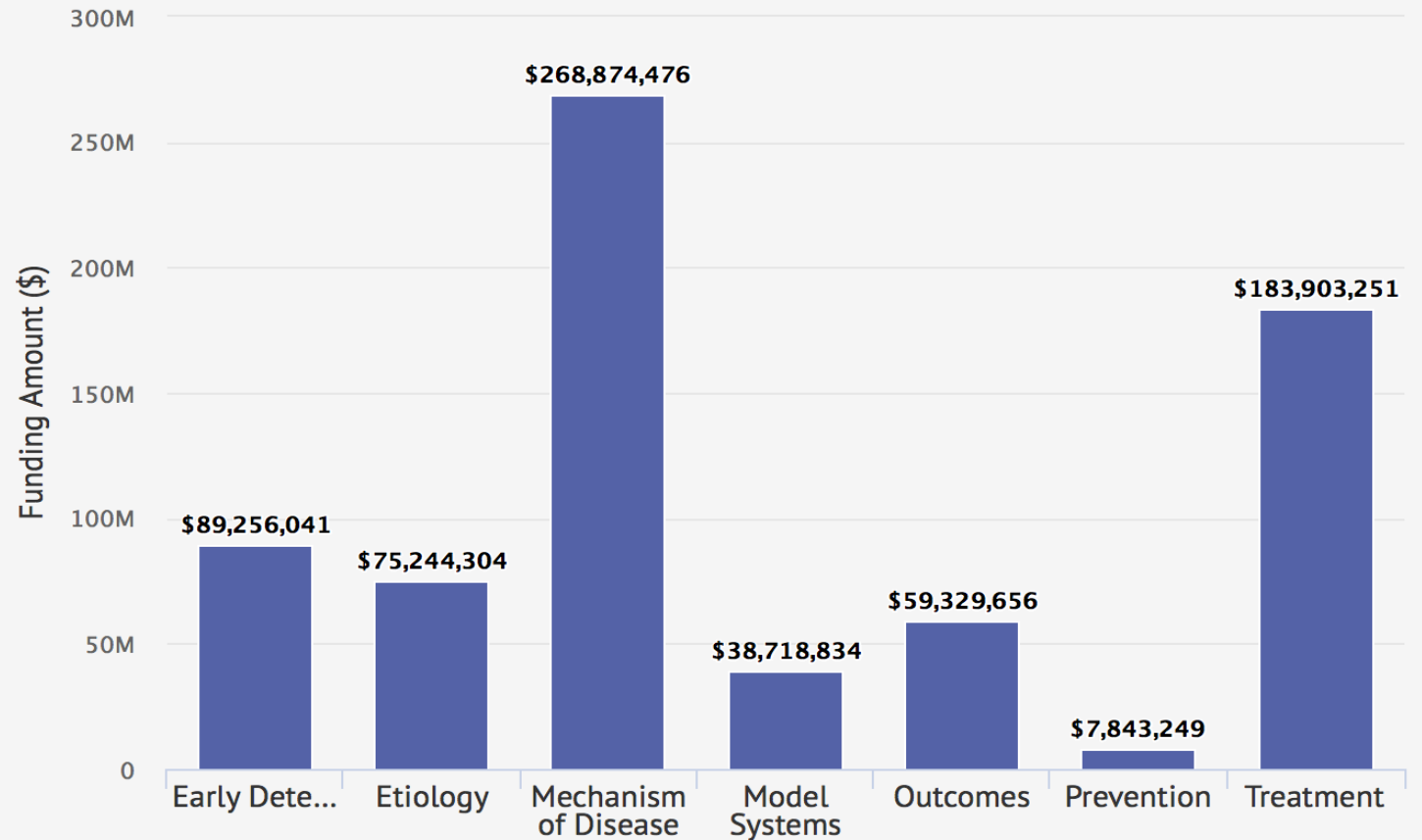
→ Suggest adding a crude sub-classification for :

“Etiology”:

- Genetic
- Infection
- Immune
- Structural
- Metabolic
- Other

Funding by Research Classification

2013-2016, all funding



Although these can be searched with keywords, adding this research classification may allow for (a) more specific search, (b) capturing epilepsies in these broader categories, when it may become too complex to add codes for all the specific causes

Condition: proposal for update

Genetic
Infection
Immune
Structural
Metabolic
Associated with other disorders
Other

• Etiology

• Epilepsy

• Seizure

• Epilepsy syndrome

• Co-occurring condition

• Consequences

• Epilepsy imitators

Onset

Focal
Generalized
Combined
Unknown

Localization

Frontal
Parietal
Occipital
Temporal
Multifocal
Generalized

Populations

Neonatal/Infantile
Childhood
Adolescent/Adult
Special populations
Other

Cognitive
Behavioral
Affective
Endocrine
Other

SUDEP
Fetal/neonatal development
Quality of life
Other

Syncope and anoxic seizures
Behavioral / Psychological and Psychiatric disorders
Sleep related conditions
Paroxysmal movement disorders
Migraine associated disorders
Miscellaneous events

Epilepsies / Seizures / Syndromes by Population

NEONATAL/INFANTILE

Self-limited neonatal seizures

Self-limited familial neonatal epilepsy

Self-limited familial and non-familial infantile epilepsy

EME – Early myoclonic epilepsy

Ohtahara syndrome

West syndrome

Dravet syndrome

Myoclonic epilepsy in infancy

Epilepsy in infancy with migrating focal seizures

Myoclonic encephalopathy in non progressive disorders

Febrile seizures plus, genetic epilepsy with febrile seizures plus

Febrile seizures

CHILDHOOD

Epilepsy with myoclonic-atonic seizures

Epilepsy with eyelid myoclonias

Lennox-Gastaut syndrome

CAE - Childhood absence epilepsy

Epilepsy with myoclonic absences

Panayiotopoulos syndrome

Childhood occipital epilepsy (Gastaut syndrome)

Photosensitive occipital lobe epilepsy

BECTS - Childhood epilepsy with centrotemporal spikes

Atypical childhood epilepsy with centrotemporal spikes

Epileptic encephalopathy with continuous spike-and-wave during sleep

LKS – Landau Kleffner syndrome

Autosomal dominant nocturnal frontal lobe epilepsy

In blue: updates / revisions from existing ontology

Epilepsies / Seizures / Syndromes by Population

ADOLESCENT / ADULT

JAE – Juvenile absence epilepsy

JME – Juvenile myoclonic epilepsy

FAME – Familial adult onset myoclonic epilepsy

Epilepsy with generalized tonic-clonic seizures alone

Autosomal dominant epilepsy with auditory features

Other familial temporal lobe epilepsies

OTHER

Familial focal epilepsy with variable foci

Reflex epilepsies

PME - Progressive myoclonus epilepsies

Epilepsy not otherwise specified

Seizures

Status epilepticus

Nonepileptic events / seizures

SPECIAL POPULATIONS

Early Life

Neonatal / Infantile

Childhood

Pregnant women

Catamenial

Elderly

In blue: updates / revisions from existing ontology

Genetic epilepsies

EPILEPSIES BY ETIOLOGY

Genetic - Chromosomal

[15q13.3 MICRODELETION SYNDROME](#)

[18q- SYNDROME](#)

[INV-DUP \(15\) OR IDIC \(15\)](#)

[DEL 1p36](#)

[ANGELMAN SYNDROME](#)

[DOWN SYNDROME \(TRISOMY 21\)](#)

[KLEINFELTERS SYNDROME \(XXY\)](#)

[MILLER DIEKER SYNDROME \(DEL 17p\)](#)

[PALLISTER KILLIAN SYNDROME \(TETRASOMY 12p\)](#)

[RING 14 \(r14\) SYNDROME](#)

[RING 20 \(r20\) SYNDROME](#)

[TRISOMY 12p](#)

[WOLF-HIRSCHHORN SYNDROME \(DEL 4p\)](#)

Genetic - Gene abnormalities

[AKT3](#)

[ARFGEF2](#)

[ARHGEF9](#)

[ARX](#)

[CACNA1A](#)

[CACNB4](#)

[CDKL5](#)

[CHD2](#)

[CHRNA2](#)

[CHRNA4](#)

[CHRNA2](#)

[CLCN2](#)

[COL4A1](#)

[DCX](#)

[DEPDC5](#)

[EFHC1](#)

[FKTN](#)

[FLNA](#)

[FMR1 \(FRAGILE X SYNDROME\)](#)

[FOXP1](#)

[GABRA1](#)

[GABRD](#)

[GABRG2](#)

[GLI3](#)

[GNAQ](#)

[GRIN2A](#)

[KCNQ2](#)

[KCNQ3](#)

[KCNT1](#)

[LARGE](#)

[LGI1](#)

[LIS1](#)

[MECP2](#)

[NPRL3](#)

[PCDH19](#)

[PIK3CA](#)

[PIK3R2](#)

[PLCB1](#)

[PNKP](#)

[POMT1](#)

[POMT2](#)

[PRRT2](#)

[RELN](#)

[SCN1A](#)

[SCN1B](#)

[SCN2A](#)

[SLC2A1](#)

[SLC25A22](#)

[SPTAN1](#)

[STXBP1](#)

[TBC1D24](#)

[TCF4 \(PITT](#)

[HOPKIN](#)

[SYNDROME\)](#)

[TSC1](#)

[TSC2](#)

[TUBA1A](#)

[WDR62](#)

[ZEB2](#)

[\(MOWAT](#)

[WILSON](#)

[SYNDROME\)](#)

By Etiology

Structural –

Malformation of cortical development

Vascular malformations

Hippocampal sclerosis

Hypoxic-ischemic

Traumatic brain injury

Tumors

Porencephalic cyst

Metabolic –

Biotinidase and holocarboxylase synthase deficiency

Cerebral folate deficiency

Creatine disorders

Folinic acid responsive seizures

Glucose transporter 1 (GLUT1) deficiency

Mitochondrial disorders

Peroxisomal disorders

Pyridoxine dependent epilepsy / PNPO deficiency

Immune –

Rasmussen's

Antibody mediated

Anti-NMDA receptor

Voltage gated potassium channel

GAD65 antibody

GABAB receptor antibody

Steroid responsive encephalopathy with thyroid disease

Celiac disease, epilepsy and cerebral calcification syndrome

Other

Infectious -

Bacterial meningitis or meningoencephalitis

Malaria

Cerebral Toxoplasmosis

CMV

HIV

Neurocysticercosis

Tuberculosis

Viral encephalitis

Other (Lyme disease, toxocariosis, schistosomiasis)

Associated with other diseases

(Alzheimer's, Autism, Fragile X, Rett syndrome, Malaria, etc)

Unknown

Febrile infection related epilepsy

Considerations

- The coding is oriented towards human classifications of epilepsies / seizures.
- Working classification for animal models of seizures and epilepsies is in progress (ILAE/AES Joint Translational Task Force) and could be considered in the future.
- New revised terms could be added as alternatives / equivalent to existing ones so as not to revise coding from past years.
- It would be useful, if easily feasible, to allow:
 - multiple choices of ontology terms from same categories or keywords
 - enhanced search tools allowing direct comparisons, head to head, of data from various search keywords.

This additional search flexibility could minimize the need in the future for ontology revisions.

Thank you!

- AES
- Eileen Murray
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