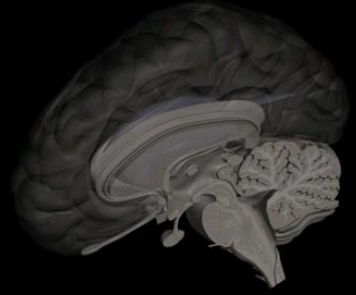
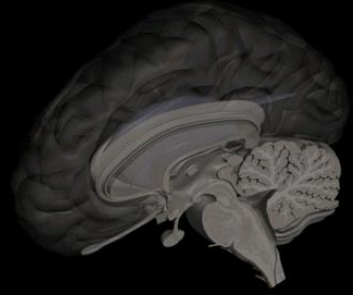


SÍNDROME CONVULSIVO EN PEDIATRÍA

DR. PATRICIO GUERRA
NEURÓLOGO INFANTIL Y ADOLESCENTES
MAGÍSTER NEUROCIENCIAS
ESCUELA DE MEDICINA UNIVERSIDAD SAN SEBASTIÁN PUERTO MONTT

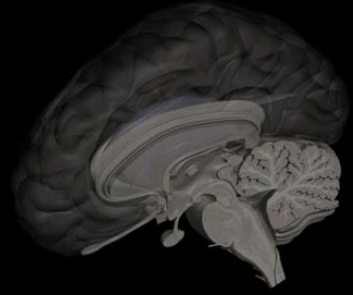
CRISIS CONVULSIVAS EN LA INFANCIA
TRES GRANDES GRUPOS ETIOLÓGICOS:



1. CRISIS PROVOCADAS O SECUNDARIAS A FENÓMENOS ORIGINADOS INICIALMENTE FUERA DEL SISTEMA NERVIOSO CENTRAL (REFLEJAS):

-ALTERACIONES METABÓLICAS
-HIPOXIA-ANOXIA
-TRASTORNOS HIDROELECTROLÍTICOS
-INTOXICACIONES
-SÍNDROME DE PRIVACIÓN OH
-TRAUMATISMOS (TEC)
-INFECCIONES
-OTRAS CAUSAS

*FENÓMENOS **AGUDOS** Y QUE REQUIEREN QUE SE SOSPECHE QUE SON PRODUCIDOS POR UNA NOXA ESPECÍFICA Y QUE REQUIEREN UNA TERAPIA ESPECÍFICA
*TAMBIÉN LLAMADAS CRISIS CONVULSIVAS NO EPILÉPTICAS



CRISIS CONVULSIVAS EN LA INFANCIA
GRANDES GRUPOS ETIOLÓGICOS:

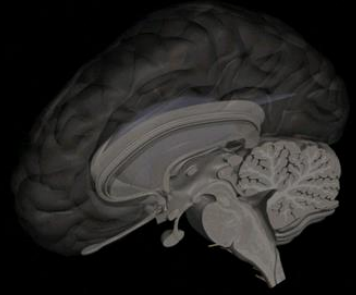
2. CONVULSIONES FEBRILES:

CRISIS EN RELACIÓN A LA PRESENCIA DE FIEBRE, EN EL CONTEXTO HABITUAL DE UN CUADRO INFECCIOSO, ANTES DE LOS 5 AÑOS DE EDAD, SIGNO DE UNA SUSCEPTIBILIDAD INDIVIDUAL FRENTE A ESTOS

CLASIFICADAS COMO SIMPLES O COMPLEJAS

FRECUENTES EN LA PRÁCTICA CLÍNICA HABITUAL

NO NECESARIAMENTE RELACIONADAS A MAYOR RIESGO DE EPILEPSIA



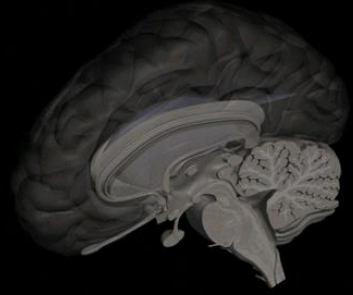
CRISIS CONVULSIVAS EN LA INFANCIA
GRANDES GRUPOS ETIOLÓGICOS:

3. CRISIS EPILÉPTICAS:

HABITUALMENTE RELACIONADAS A CRISIS CONVULSIVAS, PERO QUE ADEMÁS DE LAS CLÁSICAS CONVULSIONES TÓNICAS, CLÓNICAS O TÓNICO-CLÓNICAS, PUEDEN PRESENTARSE CON UNA AMPLIA GAMA DE SIGNOS O SÍNTOMAS, MOTORES O NO MOTORES, FOCALES O GENERALIZADOS

SE ORIGINAN POR ACTIVIDAD BIOELÉCTRICA EXCESIVA Y ANORMAL DE UN GRUPO DE NEURONAS, CORTICALES, SUBCORTICALES O INCLUSO DE TRONCO-ENCÉFALO

EL CONCEPTO DE EPILEPSIA CONLLEVA RIESGO DE RECURRENCIA



CRISIS CONVULSIVAS EN LA INFANCIA
TERMINOLOGÍA

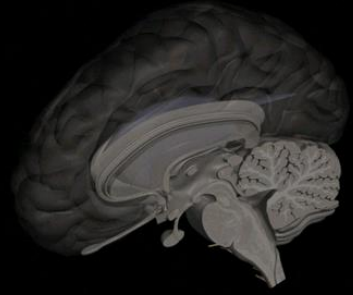
-CONVULSIÓN: MOVIMIENTO Y AGITACIÓN ALTERNADA DE CONTRACCIÓN Y ESTIRAMIENTO DE UNO O MÁS MÚSCULOS O MIEMBROS DEL CUERPO

-SEIZURE: DETENCIÓN, “ATAQUE”

-CRISIS: OCURRENCIA TRANSITORIA DE SIGNOS O SÍNTOMAS DEBIDO A ACTIVIDAD NEURONAL ANORMAL EXCESIVA O SINCRÓNICA NEURONAL EN EL CEREBRO

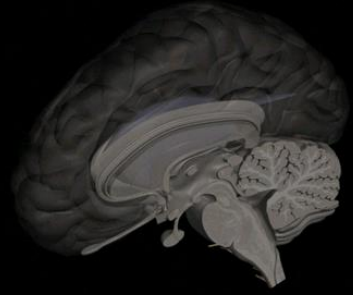
-PAROXISMO: FENÓMENOS RECURRENTES DE SIGNOS O SÍNTOMAS FUERA DEL CONTEXTO DE CONDUCTAS ASUMIDAS HABITUALMENTE COMO “NORMALES” EN UN SUJETO, ESTEREOTIPADAS O RELATIVAMENTE SIMILARES, CUYA NATURALEZA DESCONOCEMOS

CRISIS CONVULSIVAS EN LA INFANCIA
TRASTORNOS PAROXÍSTICOS EN LA INFANCIA



-GRUPO DE ALTERACIONES CONDUCTUALES, MOVIMIENTOS, SIGNOS O SÍNTOMAS QUE PUEDEN LLEVAR AL DIAGNÓSTICO ERRÓNEO DE FENÓMENOS DE TIPO EPILÉPTICO:

ALTERACIONES CARDIOLÓGICAS (SÍNCOPE)
TICS
TREMOR
DISTONÍAS
TRASTORNOS DEL SUEÑO-NARCOLEPSIA
FENÓMENOS DIGESTIVOS
CALAMBRES
FENÓMENOS EMOCIONALES-PSIQUIÁTRICOS
APNEAS EMOTIVAS
ALTERACIONES ELECTROLÍTICAS RECURRENTES
FENÓMENOS AUTONÓMICOS (LIPOTIMIAS)
JAQUECAS
VÉRTIGO



CRISIS CONVULSIVAS EN LA INFANCIA
MANEJO AGUDO DE CRISIS:

CRISIS SINTOMÁTICAS → **SUMARLE MANEJO CAUSA ESPECÍFICA**

CRISIS EPILÉPTICAS

CRISIS CONVULSIVAS FEBRILES → **BAJAR Tº**

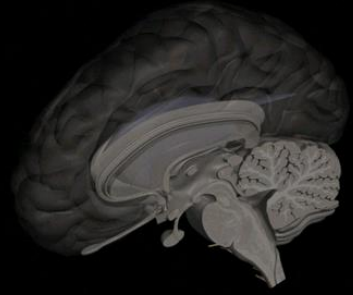
TRATAMIENTO URGENTE

CON ANTICONVULSIVANTES

DEMORA EN TRATAMIENTO

-RIESGO DAÑO SINAPSIS
-POTENCIACIÓN CRISIS
-RIESGO VITAL

CRISIS CONVULSIVAS EN LA INFANCIA
MANEJO DE CRISIS FUERA DEL HOSPITAL



-DEJAR ACOSTADO, DECÚBITO LATERAL (RIESGO ASPIRACIÓN VÓMITO)

-DESPEJAR OBSTÁCULOS MECÁNICOS VÍA AÉREA (ROPA, PRÓTESIS DENTAL)

-PROTEGER LA CABEZA DE GOLPES

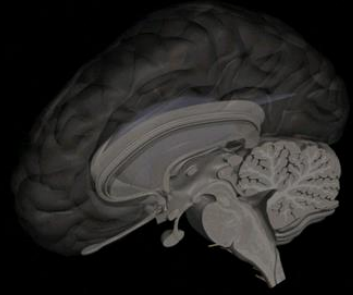
-SOLICITAR AYUDA

-NO INTRODUCIR NADA EN LA BOCA

-NO TIENE UTILIDAD AFIRMAR BRAZOS O LIMITAR MOVIMIENTOS

-CRISIS DURAN MENOS DE 1 MINUTO EN UN 91% DE CASOS: SIN RIESGO VITAL

-USO DE FÁRMACOS SI LOS TUVIERA INDICADO (RESCATAR ANTECEDENTES)



CRISIS CONVULSIVAS EN LA INFANCIA
MANEJO DE CRISIS DENTRO DEL HOSPITAL

ABC (PRIORIDADES Y MANEJO EN PARALELO)

-DESPEJAR VÍA AÉREA, EVENTUAL USO DE CÁNULA MAYO Y ASPIRACIÓN

-APLICAR OXÍGENO MASCARILLA AL 100% - OXIMETRÍA DE PULSO

-EVALUAR RESPIRACIÓN Y FRECUENCIA CARDÍACA –SIGNOS VITALES

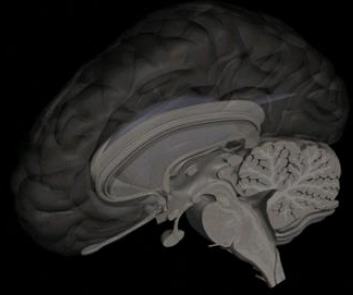
-YUGULAR CRISIS CONVULSIVA

-VÍA VENOSA PERMEABLE

-ADMINISTRAR SUERO GLUCOSADO AL 10%

-HEMOGLUCOTEST

-EVALUACIÓN CLÍNICA Y LABORATORIO



PRIORIDADES:

INICIO TEMPRANO DEL TRATAMIENTO Y CONTROL DE CRISIS:
MENOR POSIBILIDAD SE, MENOR MORTALIDAD, MENOS SECUELAS

(North London Pediatric Convulsive SE Study: Por cada minuto de demora entre el inicio del SE a la llegada al SU, 5% de riesgo acumulado de que el episodio demore más de 60 minutos)

ABC + Glucosa 10% 5ml/k bolo iv (glucosa 30% 2ml/k vía central)
↓
Enfrentamiento etiológico

PRIMERA LÍNEA: BENZODIAZEPINAS (x1-2 veces)

-LORAZEPAM 0,1 MG/K/DOSIS IV-MUCOSA ORAL, DOSIS MÁXIMA 2 MG HASTA 6 AÑOS (30k)

-MIDAZOLAM 0,2 MG/K/DOSIS IV-VO-IM-IN, DOSIS MÁXIMA 5 MG EN NIÑOS (30k)

-DIAZEPAM 0,3 MG/K/DOSIS IV-0,5 A 0,6 MG/K/DOSIS RECTAL, DOSIS MÁXIMA 10 MG

SEGUNDA LÍNEA: FENITOINA IV-FENOBARBITAL IV-LEVETIRACETAM IV BOLO
(30-40%) - INFUSIÓN CONTÍNUA MIDAZOLAN

TERCERA LÍNEA: COMA ANESTÉSICO (TIOPENTAL-PROPOFOL-PENTOBARBITAL-
(20%) ISOFLUORANO)

Inpatient Guidelines for Management and Evaluation of Status Epilepticus

TIME

0

Witnessed or suspected seizure
Goal is to intervene for seizures lasting more than 5 minutes

STABILIZE AND ASSESS THE PATIENT

1. **Check ABCs**
   - Evaluate and maintain the airway - (reposition patient's head/suction)
   - Provide 100% oxygen (non-rebreather). Place pulse oximeter.
   - Assess and support ventilation
   - Check and establish monitoring of vital signs
   - (RR, BP, pulse, temperature, pulse oximetry)
2. **Request for Crash Cart and Seizure Medication Box**
   (Note: Fosphenytoin and Lorazepam are stored in the medication refridgerator)
3. **Check vascular access**
4. **Note the time and check time of seizure onset**
5. **Check bedside glucose**
   - If glucose < 40 mg/dl, administer 5 ml/kg D10%W
6. **Administer antipyretics as indicated**

General Principles
1. Assess ABCs at each step
2. Get a good history and description from a witness
3. Determine **time of onset** of seizure and **whether this is a seizure**
4. Follow sequence of benzodiazepine, Fosphenytoin, Midazolam.
5. **Substitute Phenobarbital for Fosphenytoin in neonates.**
6. Assess risk of morbidity (see page 2)

3 min

SEIZURE DURATION NOW 5 MINUTES
START INITIAL IV OR PR THERAPY
RE-ASSESS ABCs

1. LORAZEPAM 0.1 mg/kg IV (Rate 2 mg/min)
   OR
2. DIAZEPAM PR Maximum: 20 mg

Ages 2 - 5 yr:	0.5 mg/kg PR
Ages 6 - 11 yr:	0.3 mg/kg PR
Age > 12 yr	0.2 mg/kg PR

WAIT 3-5 minutes

Keys to effective treatment
1. Begin treatment early, within 3-5 minutes of seizure onset if possible
2. Use adequate doses of effective drugs.

Select Initial labs
1. Electrolytes (Glucose, Na, Ca, Mg)
2. Antiepileptic drug levels
3. CBC

8 min

SEIZURES CONTINUE

Neonates < 1 month age
1. Load with Phenobarb 20mg/kg IV
2. RE-ASSESS ABCs
3. Call NICU and page Neurology
4. Repeat up to 40 mg/kg total dose

1. LOAD Fosphenytoin 20 mg PE/kg IV
   RATE = 3 mg PE/kg/minute.
   If patient is already on PHT, give 10 mg PE/kg
2. RE-ASSESS ABCs
3. CALL PICU and PAGE NEUROLOGY

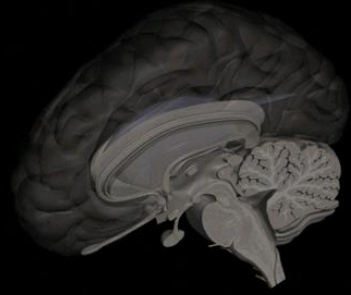
SEIZURES STOPPED

20 min

SEIZURES CONTINUE

1. Fosphenytoin 10 mg mg PE/kg IV or Phenobarbital (neonates) up to total of 40 mg/kg IV
2. CALL PICU or NICU
3. Maintain airway, re-assess ABCs.

1. Check vital signs
2. Additional diagnostic testing
3. Consider maintenance Fosphenytoin:
   5 mg PE/kg/d ÷ q8 hr
   or Phenobarbital
   3-5 mg/kg/d

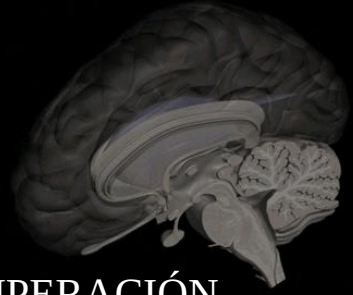


ELSEVIER
Clin Ped Emerg Med 9:96-100 © 2008
CLINICAL Pediatric Emergency Medicine

Status Epilepticus in the Pediatric Emergency Department

Joshua Goldstein, MD

STATUS EPILÉPTICO (SE)



MORTALIDAD ALREDEDOR 20%

-SE CLÁSICO: 30 MINUTOS O MÁS DE CRISIS O CRISIS ITERATIVAS SIN RECUPERACIÓN DE CONCIENCIA ENTRE ELLAS

-SE OPERACIONAL: 5 MINUTOS O MÁS DE CRISIS (SOBRE ESE PLAZO NO ES ESPERABLE QUE CRISIS CESEN EN FORMA ESPONTÁNEA)

-SE INMINENTE (“AMENAZA DE STATUS): MÁS DE 5-10 MINUTOS (USO DE BZD)

-SE ESTABLECIDO: AL MENOS 30 MINUTOS (FNT-FBB-AV-LVT-LACOSAMIDA)

-SE REFRACTARIO: CRISIS PROLONGADAS QUE PERSISTEN DESPUÉS DE LA ADMINISTRACIÓN DE 2 FAE DE DIFERENTE MECANISMO DE ACCIÓN O FÁRMACOS DE ADMINISTRACIÓN CONTÍNUA INDEPENDIENTEMENTE DE LA DURACIÓN (60 MINUTOS) MANEJO CON ANESTÉSICOS (PROPOFOL-TIOPENTAL)

-SE SUPER-REFRACTARIO: STATUS DE 24 HORAS O MÁS DESPUÉS POST MEDICAMENTOS ANESTÉSICOS (KETAMINA-$MgSO_4$-INMUNOMODULADORES)

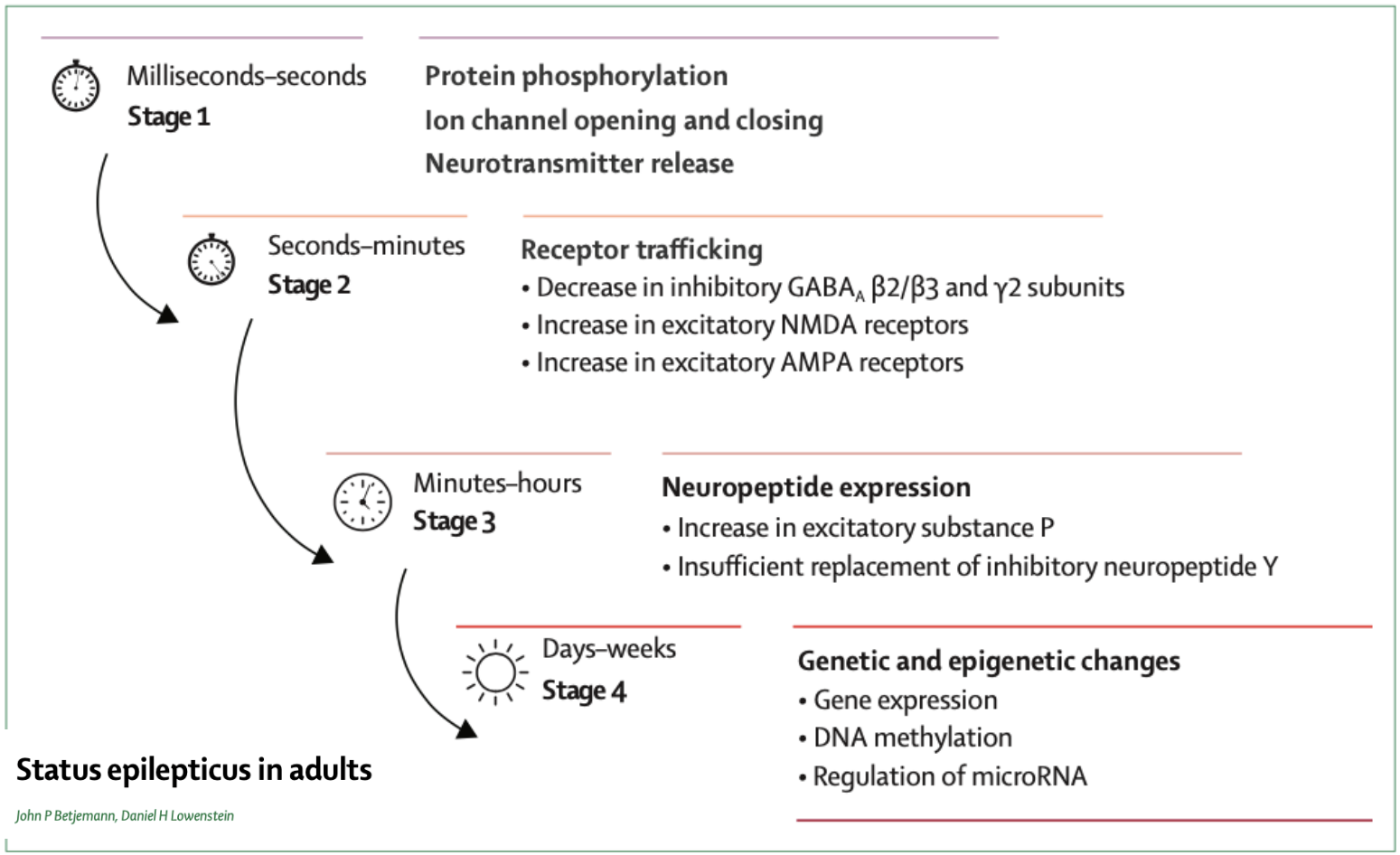
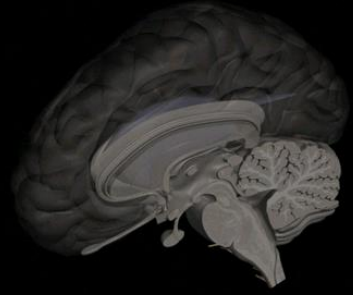


Figure 1: **Cascade of selected mechanisms involved in the transition of a single seizure to status epilepticus**

www.thelancet.com/neurology **Published online April 21, 2015** **http://dx.doi.org/10.1016/S1474-4422(15)00042-3**

CRISIS CONVULSIVAS EN LA INFANCIA
CONVULSIONES FEBRILES



-CRISIS, HABITUALMENTE MOTORAS, PRODUCIDAS EN LA INFANCIA TEMPRANA , RELACIONADAS A ALZAS FEBRILES EN EL CONTEXTO DE CUADRO INFECCIOSO, EXCLUYENDO INFECCIONES DEL SISTEMA NERVIOSO CENTRAL

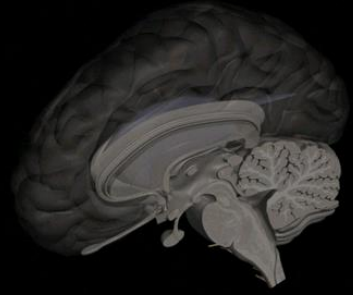
-FRECUENTES (2-5% POBLACIÓN PEDIÁTRICA OCCIDENTAL)

-EDAD CARACTERÍSTICA DE APARICIÓN: 18 MESES

-RANGO HABITUAL PRESENTACIÓN: 1-4 AÑOS

-SI BIEN PUEDEN APARECER EN MENORES DE 1 AÑO, NO ES LO HABITUAL, POR LO CUAL DEBEN EXTREMARSE LAS MEDIDAS PARA DESCARTAR UN CUADRO INFECCIOSO DEL SNC (MENINGITIS, ENCEFALITIS): CONSIDERAR QUE BAJO EL AÑO DE EDAD, LA AUSENCIA DE SIGNOS MENÍNGEOS NO LAS DESCARTA

-SOBRE LOS 6 AÑOS DE EDAD, O SE ESTÁ FRENTE A UNA CONVULSIÓN DE TIPO EPILÉPTICA GATILLADA POR LA FIEBRE, O SE TRATA DE UNA MENINGITIS O ENCEFALITIS



CRISIS CONVULSIVAS EN LA INFANCIA
CONVULSIONES FEBRILES

-SUSCEPTIBILIDAD GENÉTICA

-PUEDEN OCURRIR EN ETAPA PREVIA AL INICIO DEL CUADRO FEBRIL

-SE DIVIDEN EN SIMPLES Y COMPLEJAS (MÁS DE 10 MINUTOS, FOCAL, MÁS DE UNA CRISIS EN 24 HRS, PACIENTE CON RDSM , EEG ALTERADO)

-ANTE UNA PRIMOCONVULSIÓN FEBRIL SIMPLE: 30% RECURRENCIA EN 6 MESES

-RIESGO DESARROLLO EPILEPSIA, CF SIMPLE 2%, CF COMPLEJA 7%, RIESGO AUMENTA EN RELACIÓN A ANTECEDENTES FAMILIARES DE EPILEPSIA, CF COMPLEJA Y LA PRESENCIA DE RETRASO PSICOMOTOR

-SIN INDICACIÓN DE USO DE FÁRMACOS PROFILÁCTICOS

-FRENTE A PRIMERA CRISIS SIN INDICACIÓN DE EEG NI NEUROIMÁGENES

-LO MÁS IMPORTANTE: NO DEJAR PASAR UNA MENINGITIS (RIESGO 0,2-0,6%)

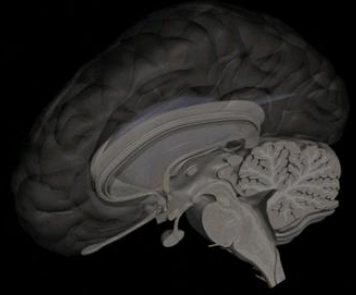
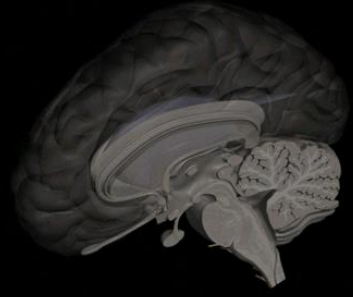


TABLE 3-1 Key Action Statements on the Indications of Lumbar Puncture (Cerebrospinal Fluid Examination) in a Child Who Presents With Seizure and Fever[a]

In Any Child Who Presents With a Seizure and Fever, a Lumbar Puncture:	Level of Evidence
1a: Should be performed if the child has meningeal signs and symptoms or history or examination raises a possibility of meningitis or intracranial infection	B (Overwhelming evidence from observational studies)
1b: Is an option in an infant 6–12 months of age when the child is considered deficient in *Haemophilus influenzae* type b (Hib) or *Streptococcus pneumoniae* immunizations or when immunization status cannot be determined	D (Expert opinion, case reports)
1c: Is an option when the child has been pretreated with antibiotics, because antibiotic treatment can mask the signs and symptoms of meningitis	D (Reasoning from clinical experience, case series)

[a] Data from Subcommittee on Febrile Seizures; American Academy of Pediatrics, Pediatrics.[3]

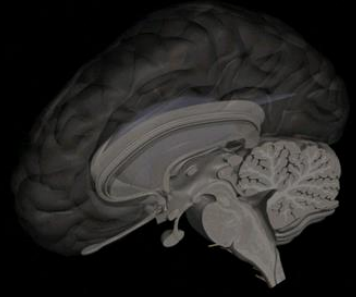
Gupta A. Continuum (Minneap Minn) 2016;22(1):51–59
American Academy of Pediatrics. Pediatrics 2011;127(2):389Y394



BMJ 2015;351:h4240 doi: 10.1136/bmj.h4240 (Published 18 August 2015)

Box 1: Red flags suggestive of central nervous system infection[31-33]

- History of irritability, decreased feeding, or lethargy
- Complex febrile seizures
- Any physical signs of meningitis or encephalitis (bulging fontanelle, neck stiffness, photophobia, focal neurological signs)
- Prolonged postictal altered consciousness or neurological deficit (>1 hour)
- Drowsiness with limited response to social cues (lasting >1 hour)
- Previous or current treatment with antibiotics
- Incomplete immunisation in children aged 6-18 months against *Haemophilus influenzae* b and *Streptococcus pneumoniae*
- In children <2 years old, symptoms and signs of meningeal irritation such as meningism and photophobia may be absent in meningitis and further assessment by a senior paediatrician (or general practitioner with suitable training, depending on the setting) may be required. If there is genuine uncertainty, a lumbar puncture should be performed but postponed if there is reduced consciousness

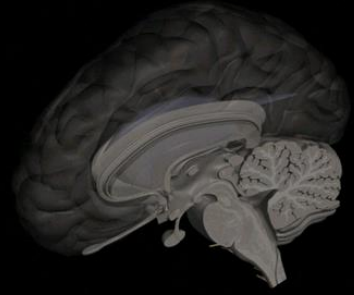


BMJ 2015;351:h4240 doi: 10.1136/bmj.h4240 (Published 18 August 2015)

The bottom line

- Febrile seizures are the commonest childhood seizure
- There is a low risk (1 in 40) of developing epilepsy in simple febrile seizures
- Benzodiazepines can be used as rescue treatment for recurrent prolonged febrile seizures
- There is no evidence of benefit for prophylactic antiepileptic drugs
- Children with simple febrile seizures have good cognitive outcomes
- Some children with recurrent or prolonged febrile seizures may have some memory impairment. It is not yet clear if this is permanent or if they "catch up" in time

CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA - DEFINICIÓN

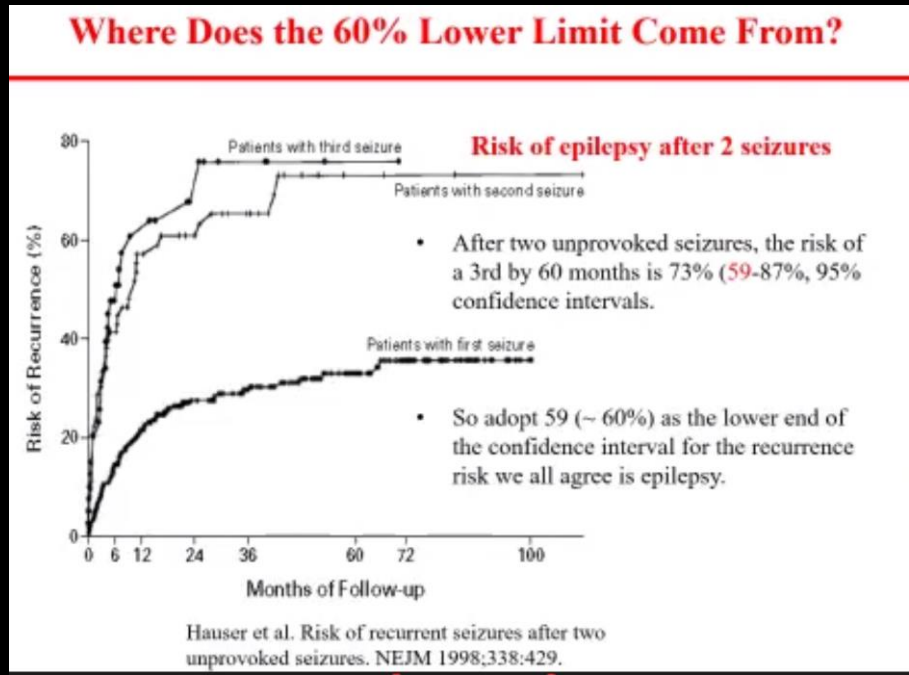


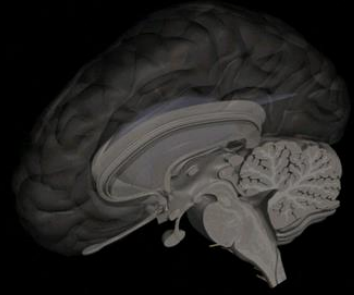
-ENFERMEDAD DEL CEREBRO DEFINIDA POR CUALQUIERA DE LAS SIGUIENTES CONDICIONES:

1. PRESENCIA DE DOS CRISIS NO PROVOCADAS EN LAPSO MAYOR A 24 HRS (UNA PRIMOCONVULSIÓN AFEBRIL TIENE UN 50% DE RECURRENCIA)

2. UNA CRISIS NO PROVOCADA Y UN RIESGO ESTIMADO DE RECURRENCIA SIMILAR AL DE LA RECURRENCIA GENERAL DESPUÉS DE DOS CRISIS NO PROVOCADAS (AL MENOS 60% EN LOS PRÓXIMOS 10 AÑOS):
   -DÉFICIT NEUROLÓGICO
   -EEG CON ACTIVIDAD EPILEPTIFORME INTERICTAL INEQUÍVOCA
   -NEUROIMÁGENES ANORMALES
   -PACIENTE/FAMILIA/MÉDICO CONSIDEREN RIESGO INACEPTABLE EL PRESENTAR UNA SEGUNDA CRISIS CONVULSIVA

3. DIAGNÓSTICO DE UN SÍNDROME EPILÉPTICO





CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA – CONCEPTOS GENERALES

-AFECTA AL 1-2 % DE LA POBLACIÓN

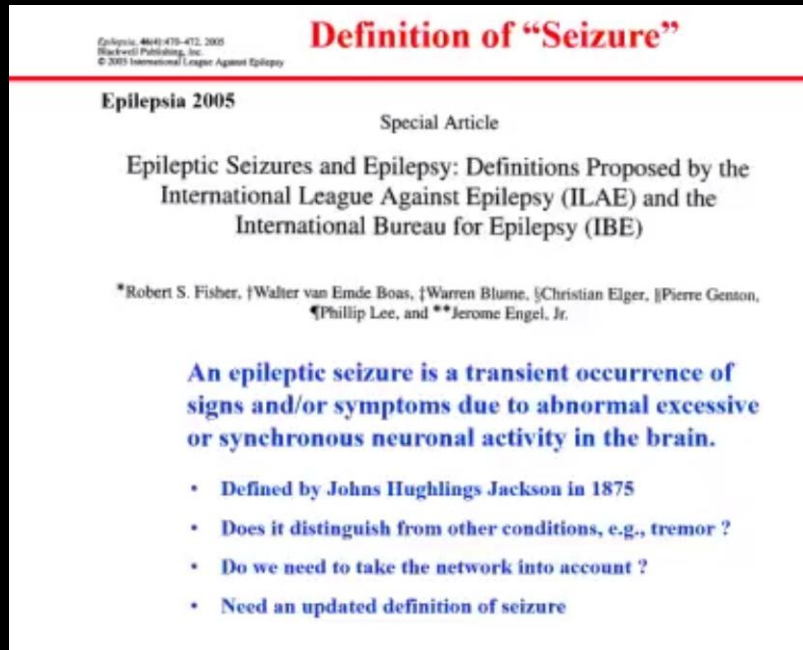
-IMPORTANTE CARGA DE ENFERMEDAD

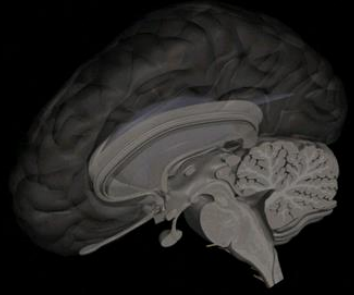
-UN PORCENTAJE DE PACIENTES EVOLUCIONA A LA CRONICIDAD

-UN PORCENTAJE DE PACIENTES EVOLUCIONA A LA CURACIÓN

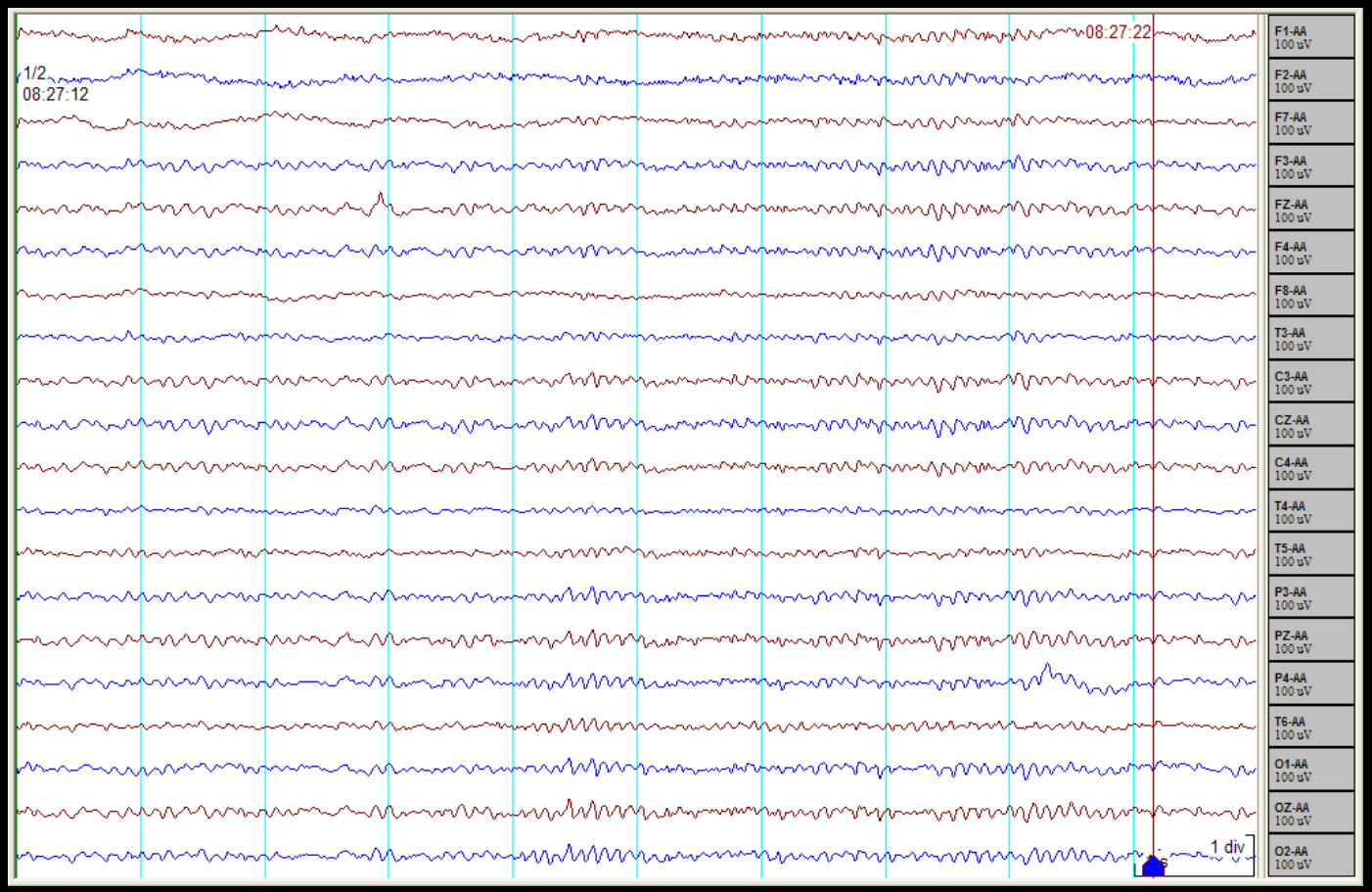
-GRAN HETEROGENEIDAD CLÍNICA

-ESTIGMA SOCIAL IMPORTANTE

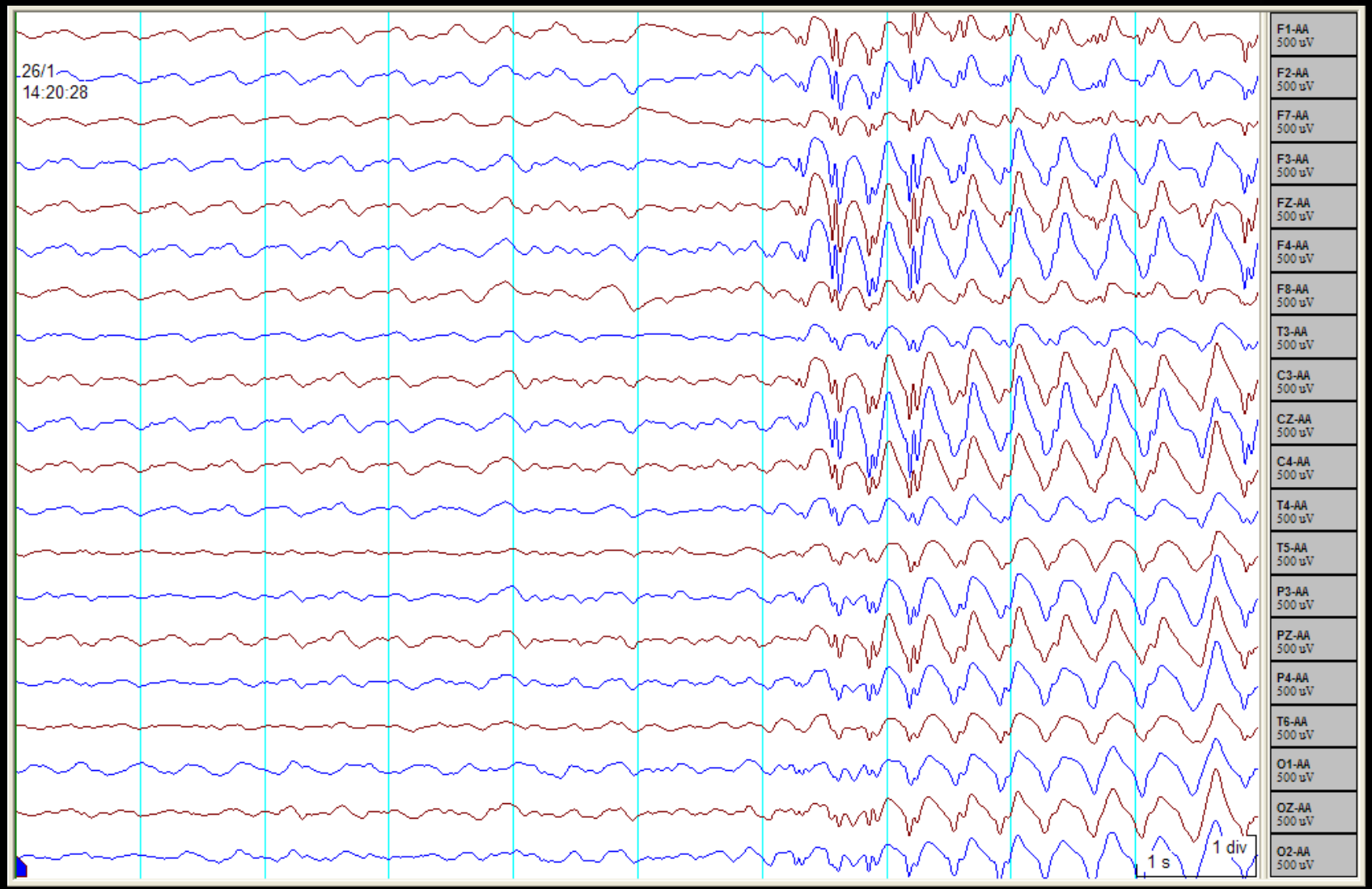


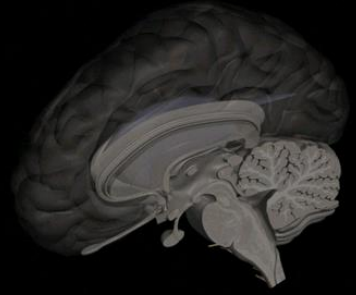


TRAZADO ELECTROENCEFALOGRÁFICO NORMAL

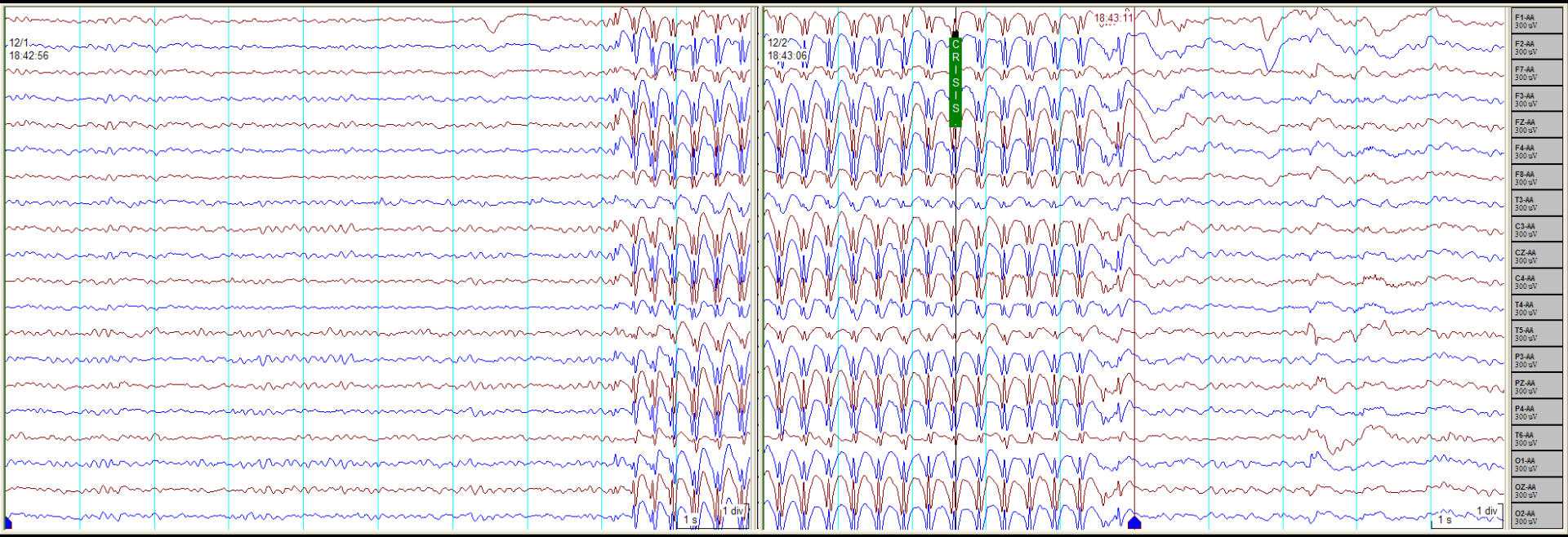


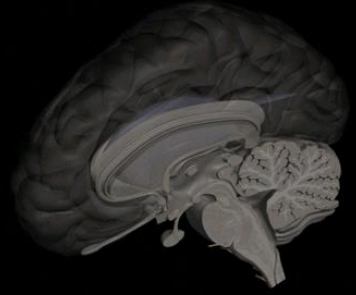
REGISTRO ELECTROENCEFALOGRÁFICO CON CRISIS DE INICIO FOCAL





REGISTRO ELECTROENCEFALOGRÁFICO CON CRISIS DE INICIO GENERALIZADO





CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA – ETIOLOGÍA

1. GENÉTICA
2. INFECCIOSA
3. METABÓLICA
4. INMUNE
5. DESCONOCIDA

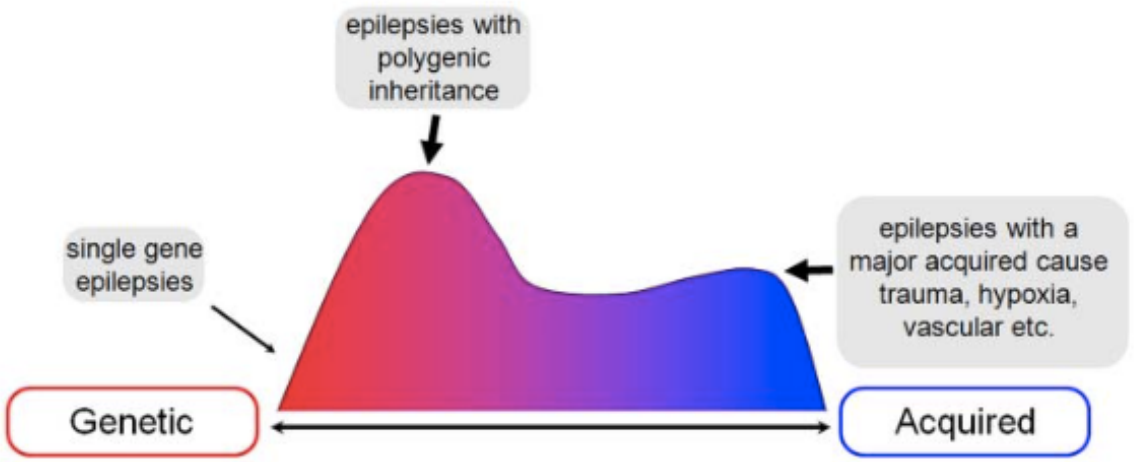


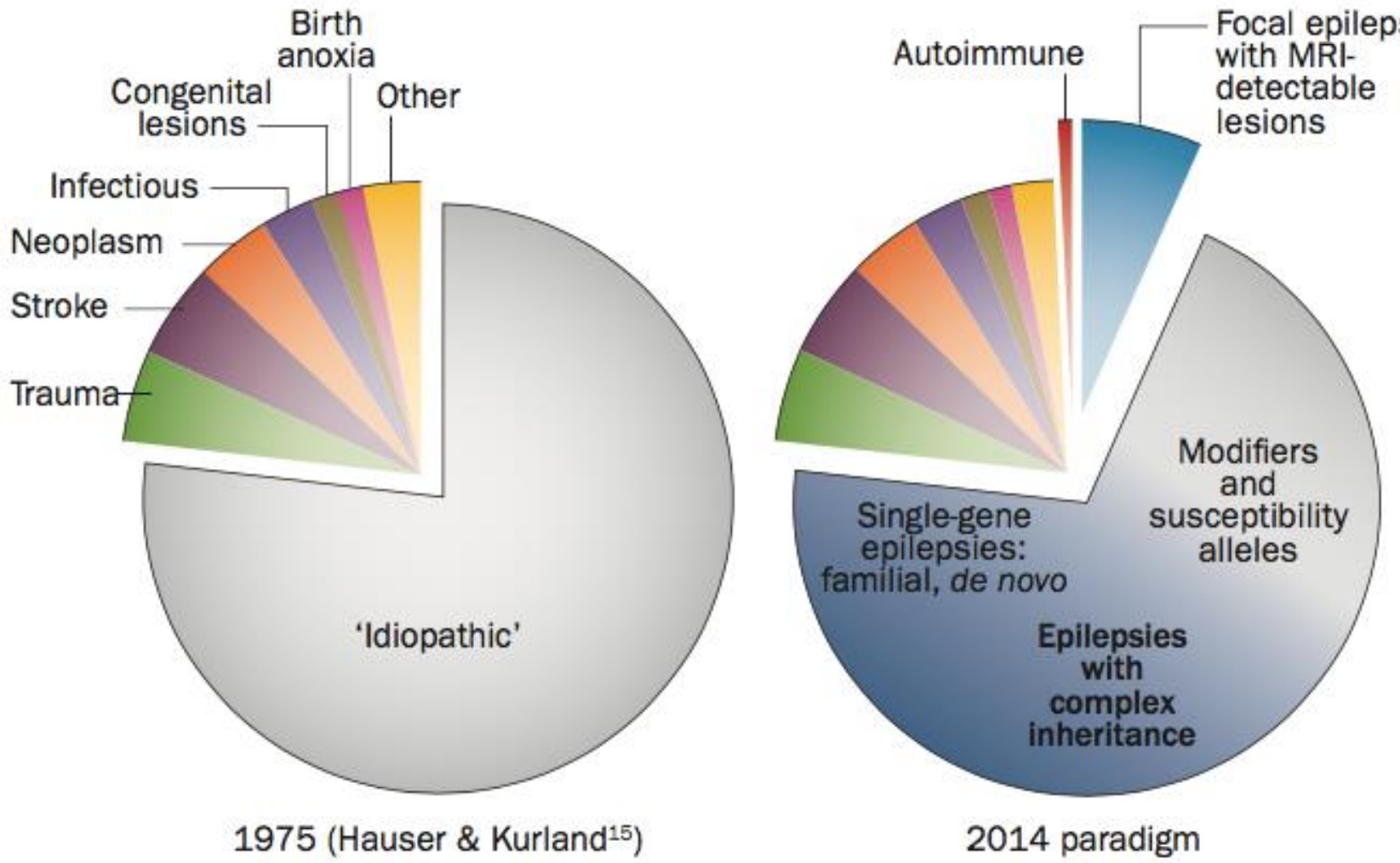
Figure 1 Contributions to common epilepsies. The causation of common epilepsies is a biological continuum due to the overlap between genetic and acquired cases. The vertical axis represents approximate frequency. Among genetic epilepsies polygenic cases are predominant and the vast majority remain unsolved. Adapted from.[5]

Hildebrand MS, *et al*. *J Med Genet* 2013;**50**:271–279. doi:10.1136/jmedgenet-2012-101448

The hidden genetics of epilepsy—a clinically important new paradigm

Rhys H. Thomas and Samuel F. Berkovic

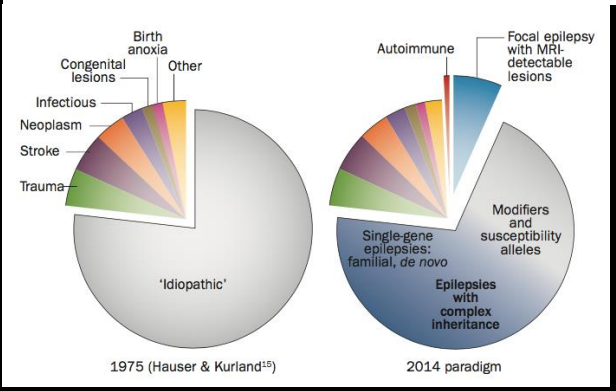
Thomas, R. H. & Berkovic, S. F. *Nat. Rev. Neurol.* advance online publication 15 April 2014;

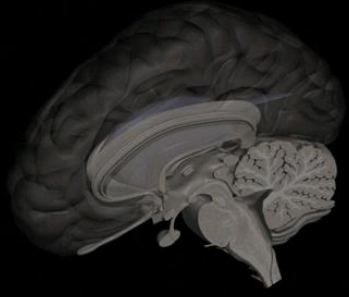


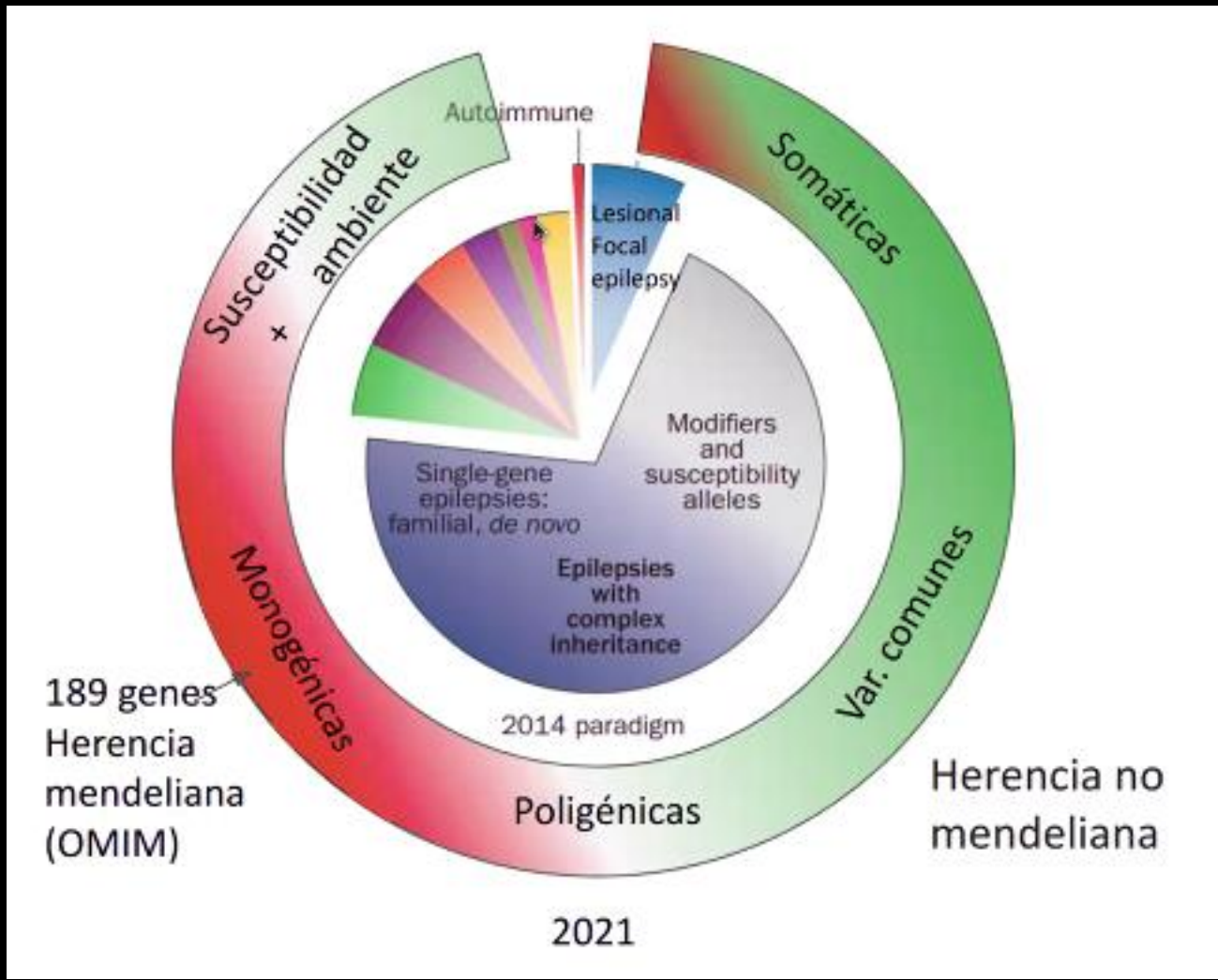
The hidden genetics of epilepsy—a clinically important new paradigm

Rhys H. Thomas and Samuel F. Berkovic

Thomas, R. H. & Berkovic, S. F. *Nat. Rev. Neurol.* advance online publication 15 April 2014;







CRISIS CONVULSIVAS CLASIFICACIÓN ILAE 1981

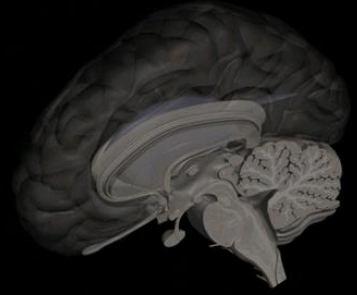


Table 4. International Classification of Epileptic Seizures

Partial (Focal, Localized) Seizures

- Simple partial seizures
  - —With motor signs
  - —With somatosensory or special sensory systems
  - —With autonomic symptoms and signs
  - —With psychic symptoms
- Complex partial seizures
  - —Simple partial onset followed by impairment of consciousness
  - —With impairment of consciousness at onset
- Partial seizures evolving to secondarily generalized seizures
  - —Simple partial seizures evolving to generalized seizures
  - —Complex partial seizures evolving to complex partial seizures evolving to generalized seizures

Generalized Seizures (Convulsive or Nonconvulsive)

- Absence seizures
  - —Typical absences
  - —Atypical absences
- Myoclonic seizures
- Clonic seizures
- Tonic seizures
- Tonic–clonic seizures
- Atonic seizures

Unclassified Epileptic Seizures

Adapted from the Commission on Classification and Terminology of the International League Against Epilepsy. Proposal for revised clinical and electroencephalographic classification of epileptic seizures. *Epilepsia*. 1981;22:489–501.

CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA – CLASIFICACIÓN 2017

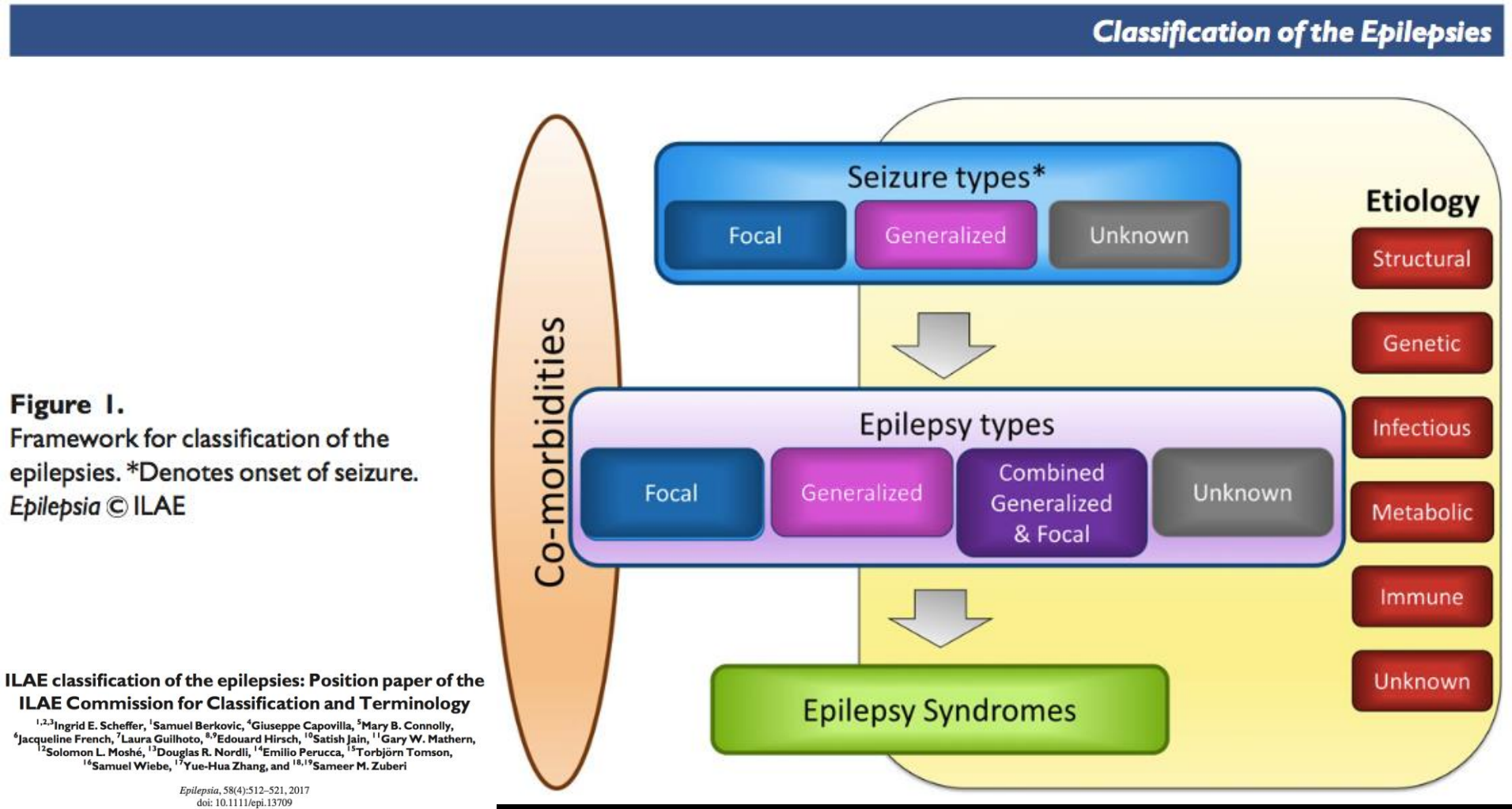


Figure 1.
Framework for classification of the epilepsies. *Denotes onset of seizure.
Epilepsia © ILAE

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

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

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

Table 1. Changes in seizure type classification from 1981 to 2017

1. Change of "partial" to "focal"
2. Certain seizure types can be either of focal, generalized, or unknown onset
3. Seizures of unknown onset may have features that can still be classified
4. Awareness is used as a classifier of focal seizures
5. The terms dyscognitive, simple partial, complex partial, psychic, and secondarily generalized were eliminated
6. New focal seizure types include automatisms, autonomic, behavior arrest, cognitive, emotional, hyperkinetic, sensory, and focal to bilateral tonic–clonic seizures. Atonic, clonic, epileptic spasms, myoclonic, and tonic seizures can be either focal or generalized
7. New generalized seizure types include absence with eyelid myoclonia, myoclonic absence, myoclonic–tonic–clonic, myoclonic–atonic, and epileptic spasms

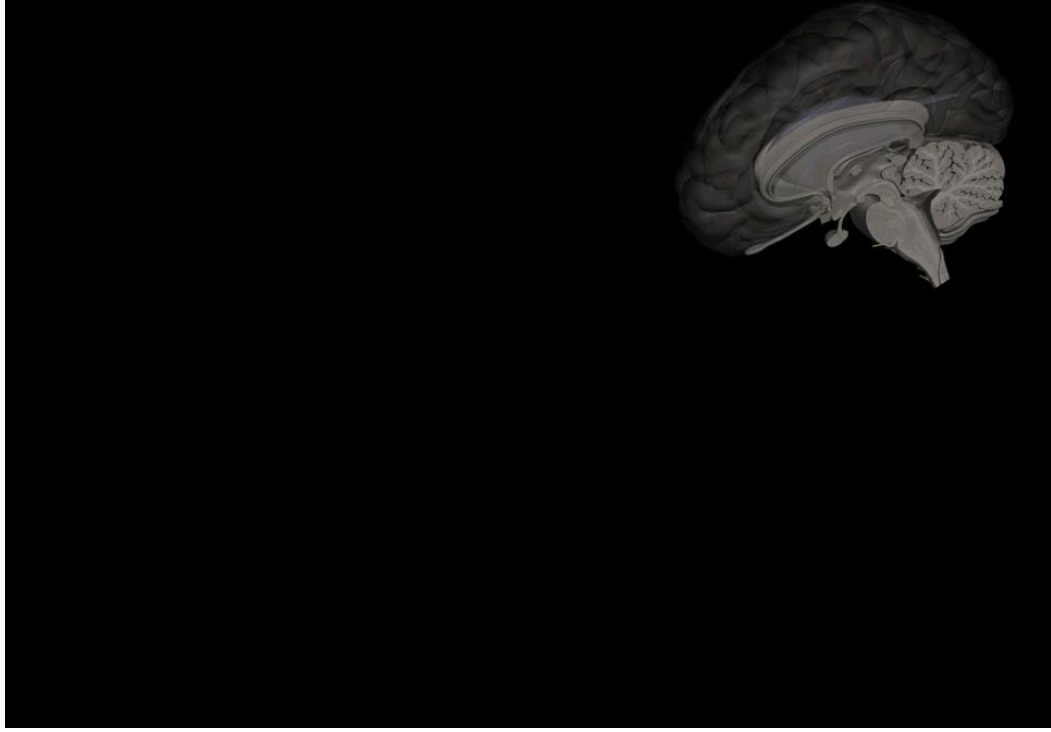
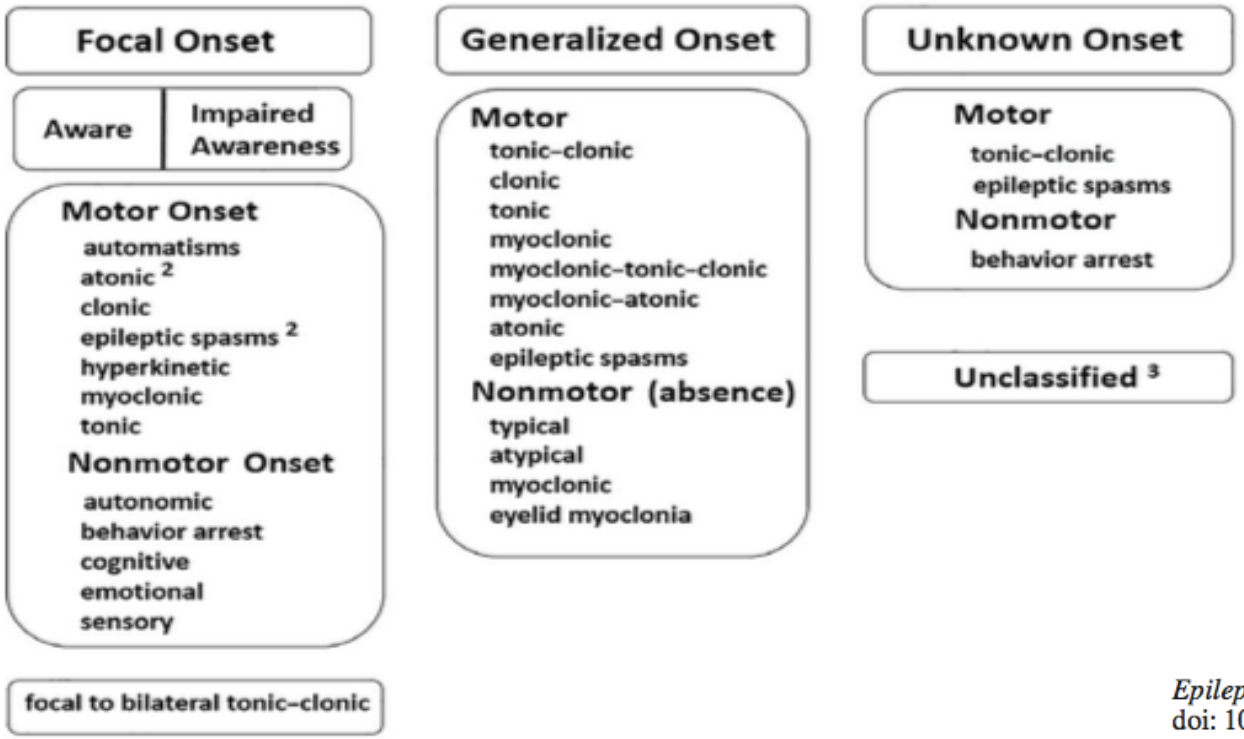


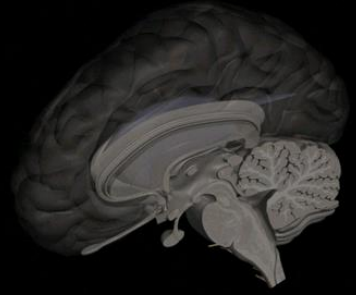
Figure 1.
The basic ILAE 2017 operational classification of seizure types. [1]Definitions, other seizure types and descriptors are listed in the accompanying paper and glossary of terms. [2]Due to inadequate information or inability to place in other categories.
Epilepsia © ILAE

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



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

Figure 2.
The expanded ILAE 2017 operational classification of seizure types. The following clarifications should guide the choice of seizure type. For focal seizures, specification of level of awareness is optional. Retained awareness means the person is aware of self and environment during the seizure, even if immobile. A focal aware seizure corresponds to the prior term simple partial seizure. A focal impaired awareness seizure corresponds to the prior term complex partial seizure, and impaired awareness during any part of the seizure renders it a focal impaired awareness seizure. Focal aware or impaired awareness seizures optionally may further be characterized by one of the motor-onset or nonmotor-onset symptoms below, reflecting the first prominent sign or symptom in the seizure. Seizures should be classified by the earliest prominent feature, except that a focal behavior arrest seizure is one for which cessation of activity is the dominant feature throughout the seizure. A focal seizure name also can omit mention of awareness when awareness is not applicable or unknown and thereby classify the seizure directly by motor onset or nonmotor-onset characteristics. Atonic seizures and epileptic spasms would usually not have specified awareness. Cognitive seizures imply impaired language or other cognitive domains or positive features such as déjà vu, hallucinations, illusions, or perceptual distortions. Emotional seizures involve anxiety, fear, joy, other emotions, or appearance of affect without subjective emotions. An absence is atypical because of slow onset or termination or significant changes in tone supported by atypical, slow, generalized spike and wave on the EEG. A seizure may be unclassified due to inadequate information or inability to place the type in other categories. [1]Definitions, other seizure types and descriptors are listed in the accompanying paper and glossary of terms. [2]Degree of awareness usually is not specified. [3]Due to inadequate information or inability to place in other categories.



CRISIS CONVULSIVAS EN LA INFANCIA
SÍNDROMES EPILÉPTICOS (SÍNDROMES ELECTRO-CLÍNICOS)

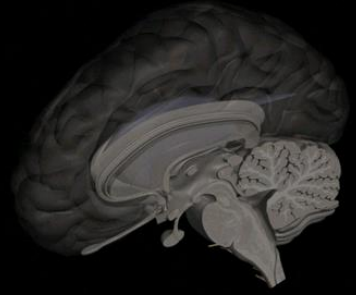
-GRUPO DE CARÁCTERÍSTICAS CLÍNICAS, ELECTROENCEFALOGRÁFICAS, IMAGENOLÓGICAS QUE TIENDEN A AGRUPARSE

-A MENUDO SON EDAD-DEPENDIENTE EN SU CLÍNICA EN CUANTO A INICIO Y REMISIÓN

-COMPARTEN CARÁCTERÍSTICAS COMO TIPOS DE CRISIS,DESENCADENANTES, VARIACIÓN HORARIA Y PRONÓSTICO

-PUEDEN TENER DIFERENTES COMORBILIDADES, POR EJEMPLO, DISFUNCIÓN COGNITIVA O PSIQUIÁTRICA

-CLASIFICAR A UN SUJETO EN UN SÍNDROME EPILÉPTICO TIENE IMPLICANCIAS EN ETIOLOGÍAS ASOCIADAS, PRONÓSTICO Y TRATAMIENTO



SÍNDROMES EPILÉPTICOS EN PEDIATRÍA

-SÍNDROME DE WEST

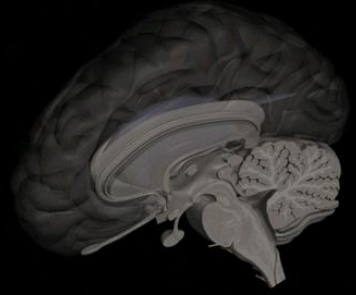
-SÍNDROME DE LENNOX-GASTAUT

-AUSENCIAS CLÁSICAS

-EPILEPSIA ROLÁNDICA O BENIGNA DE LA INFANCIA (ESPIGAS CENTRO-TEMPORALES)

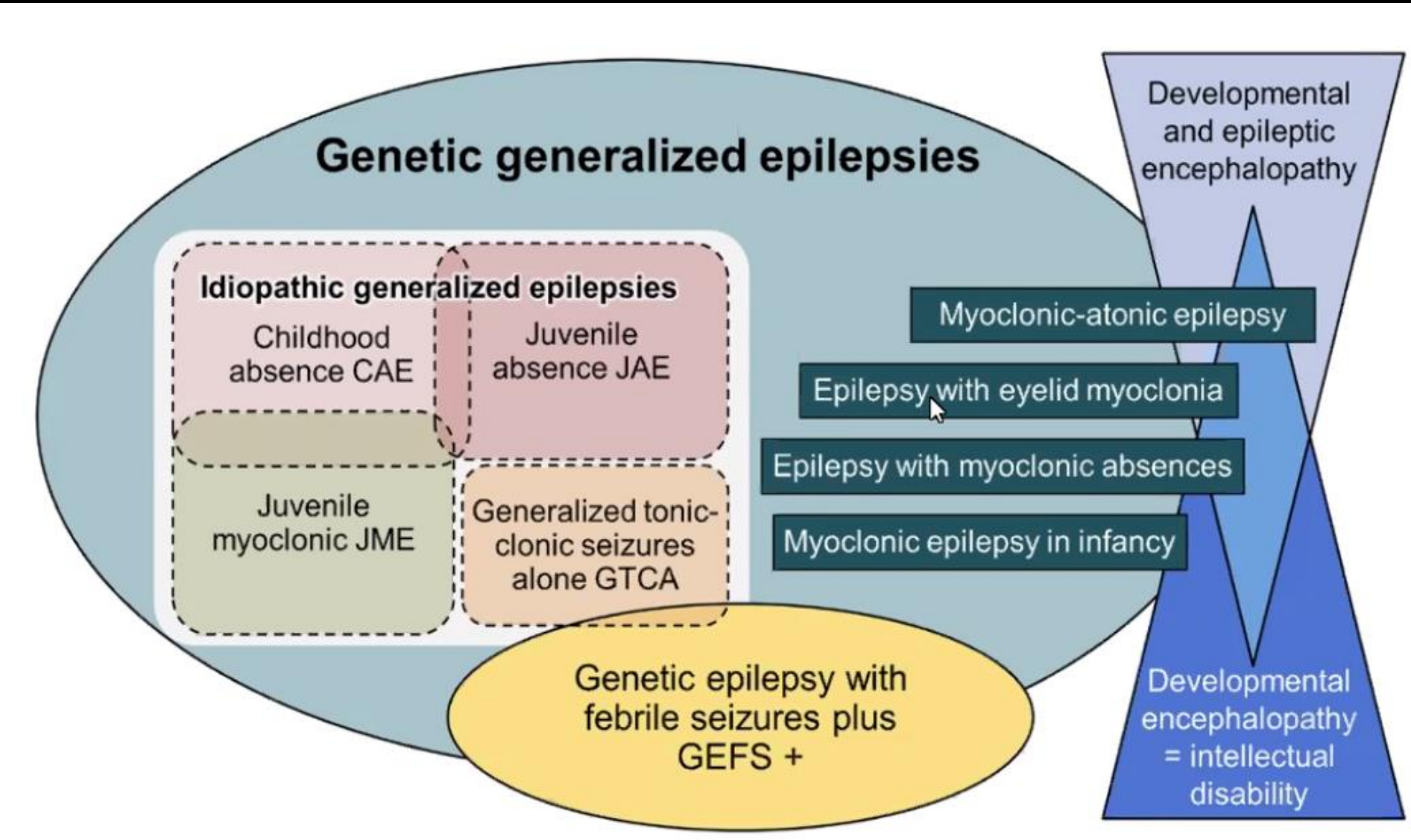
-EPILEPSIA MIOCLÓNICA JUVENIL

SÍNDROMES EPILÉPTICOS EN PEDIATRÍA
IMPORTANCIA DE DEFINIR SÍNDROMES EPILÉPTICOS



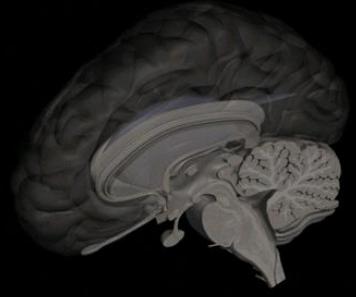
	Juvenile Myoclonic Epilepsy (JME)	Generalized Tonic-Clonic Seizures Alone (GTCA)
Age at onset **-Usual** **-Range**	 10-24 years 8-40 years	 10-25 years 5-40 years
Development	Typically normal but may have learning disorder or ADHD	Typically normal but may have learning disorder or ADHD
Main seizure type	Myoclonic seizures, seen predominantly on awakening	Generalized tonic-clonic seizures typically within 2 hours of awakening
Other seizure types **-Febrile seizures**	 May occur in approximately 15% Generalized tonic-clonic seizures in >90% which are often preceded by myoclonic jerks (myoclonic-tonic-clonic), and often occur on awakening Absence seizures in 33% - typically brief (3-8 seconds), infrequent (<daily) and with variable impairment of awareness	 May occur in approximately 15% Absence or myoclonic seizures are not present
Triggers	Sleep deprivation Photic stimulation	Sleep deprivation
EEG Background	Normal	Normal
Epileptiform Discharges	Irregular, generalized 3-5.5 Hz spike-wave and polyspike-wave seen in all states May fragment in sleep	Generalized 3-5.5 Hz spike-wave or polyspike-wave, which may be seen only in sleep May fragment in sleep
Photoparoxsmal response	Seen in 33% and may trigger myoclonic jerks or generalized myoclonic-tonic-clonic seizures	May be seen
Hypeventilation induction	33% have hyperventilation-induced generalized spike-wave discharge but rarely induces absence seizures	May be seen
Ictal EEG	Disorganized discharges 110 fold more common in absences with JME than CAE Generalized polyspike-wave with myoclonic jerks 3.5-6 Hz generalized spike-wave or polyspike-wave with absences Generalized spikes with tonic phase of generalized tonic-clonic seizure followed by spike-wave during clonic phase – but often obscured by muscle artifact	Generalized spikes with tonic phase followed by spike-wave during clonic phase – but often obscured by muscle artifact

SÍNDROMES EPILÉPTICOS EN PEDIATRÍA
IMPORTANCIA DE DEFINIR SÍNDROMES EPILÉPTICOS

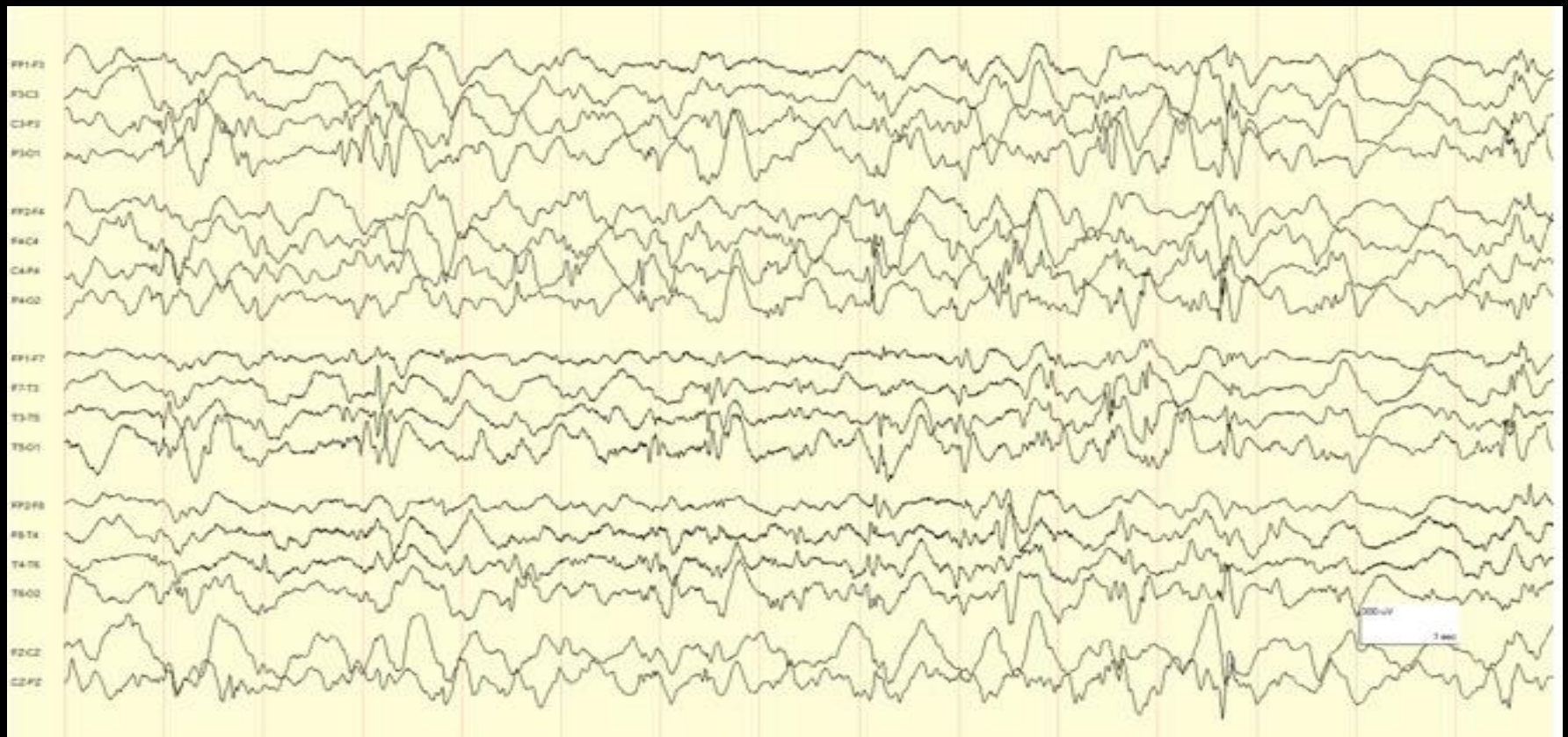


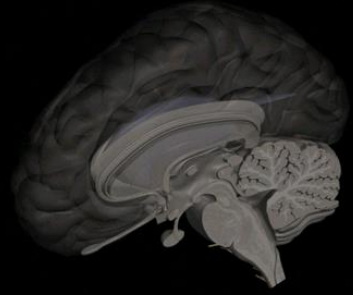
SÍNDROMES EPILÉPTICOS EN PEDIATRÍA

SÍNDROME DE WEST



-ESPASMOS EN FLEXIÓN, HIPSARRÍTMIA, RDSM (DETERIORO)
-LACTANTES MENORES: 6-18 MESES
-ENCEFALOPATÍA EPILÉPTICA
-URGENTE DIAGNÓSTICO Y MANEJO PRECOZ (ACTH)

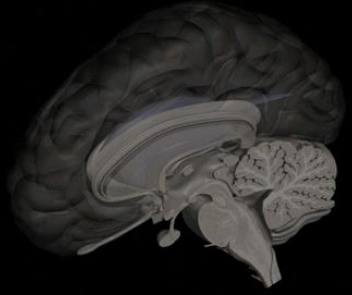


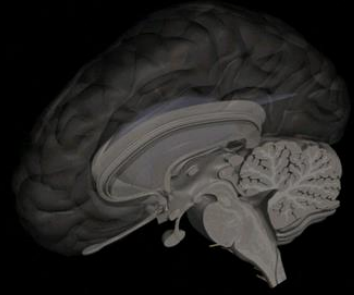


SÍNDROMES EPILÉPTICOS EN PEDIATRÍA

SÍNDROME DE LENNOX-GASTAUT

-PREESCOLARES (3-5 AÑOS)
-CRISIS ATÓNICAS, TÓNICAS, AUSENCIAS
-HABITUALMENTE NIÑOS CON DAÑO NEUROLÓGICO O RDSM PREVIO
-ALTA REFRACTARIEDAD A TRATAMIENTO
-ENCEFALOPATÍA EPILÉPTICA
-ETIOLOGÍA: DAÑO ESTRUCTURAL SNC-GENÉTICA

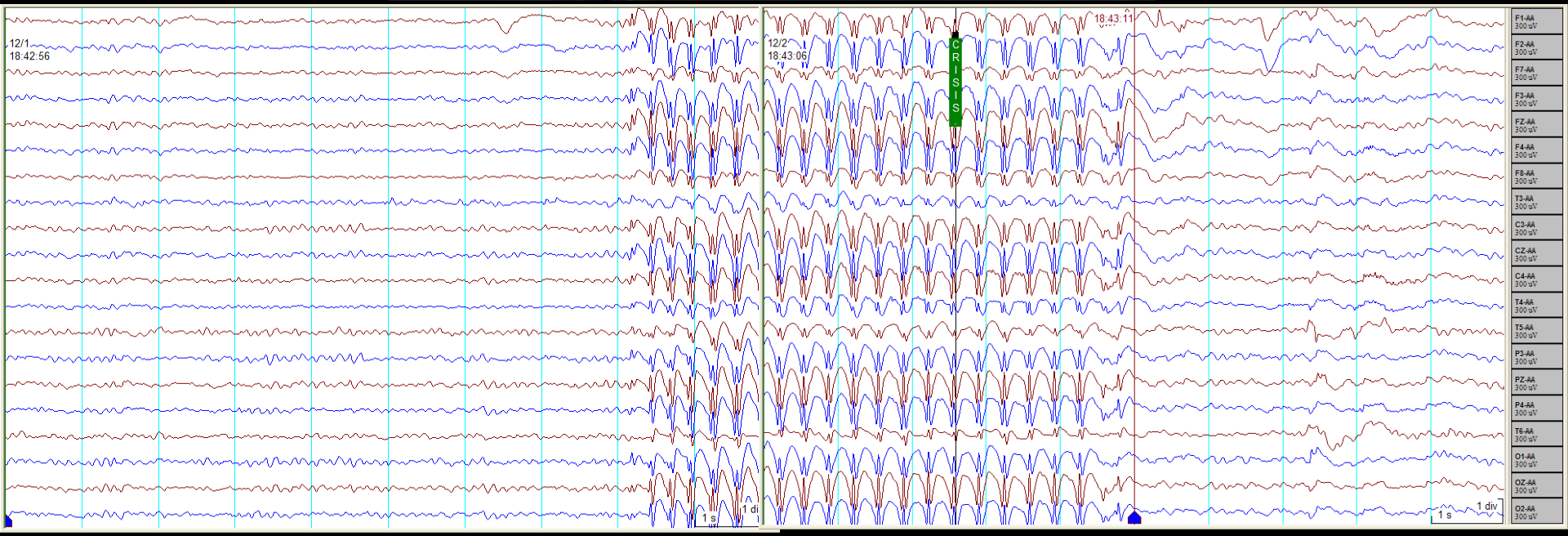




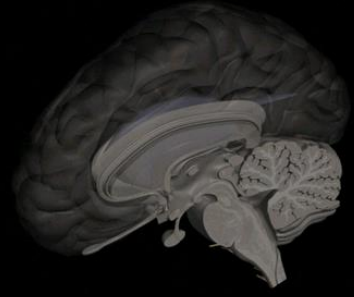
SÍNDROMES EPILÉPTICOS EN PEDIATRÍA

AUSENCIAS CLÁSICAS

-EPILEPSIA GENERALIZADA DE BASE GENÉTICA
-PREESCOLARES DE 2 A 12 AÑOS (MÁXIMA INCIDENCIA 5-6 AÑOS)
-DECENAS DE EPISODIOS DE DESCONECCIÓN DEL MEDIO
-CRISIS DE SEGUNDOS DE DURACIÓN DE COMPROMISO DE CONCIENCIA
-A VECES RELACIONADAS A PARPADEO Y/O AUTOMATISMOS FACIALES
-SIN CONCIENCIA PERSONAL DE SU OCURRENCIA
-MUCHAS VECES DETECTADAS POR PROFESORES
-HABITUALMENTE SE OBSERVAN EN NIÑOS SANOS Y CON DSM NORMAL
-PATRÓN EEG CARÁCTERÍSTICO
-DESENCADENADAS POR HIPERVENTILACIÓN
-PUEDEN RELACIONARSE A PROBLEMAS DE APRENDIZAJE-CONCENTRACIÓN
-SIN INDICACIÓN DE NEUROIMÁGENES



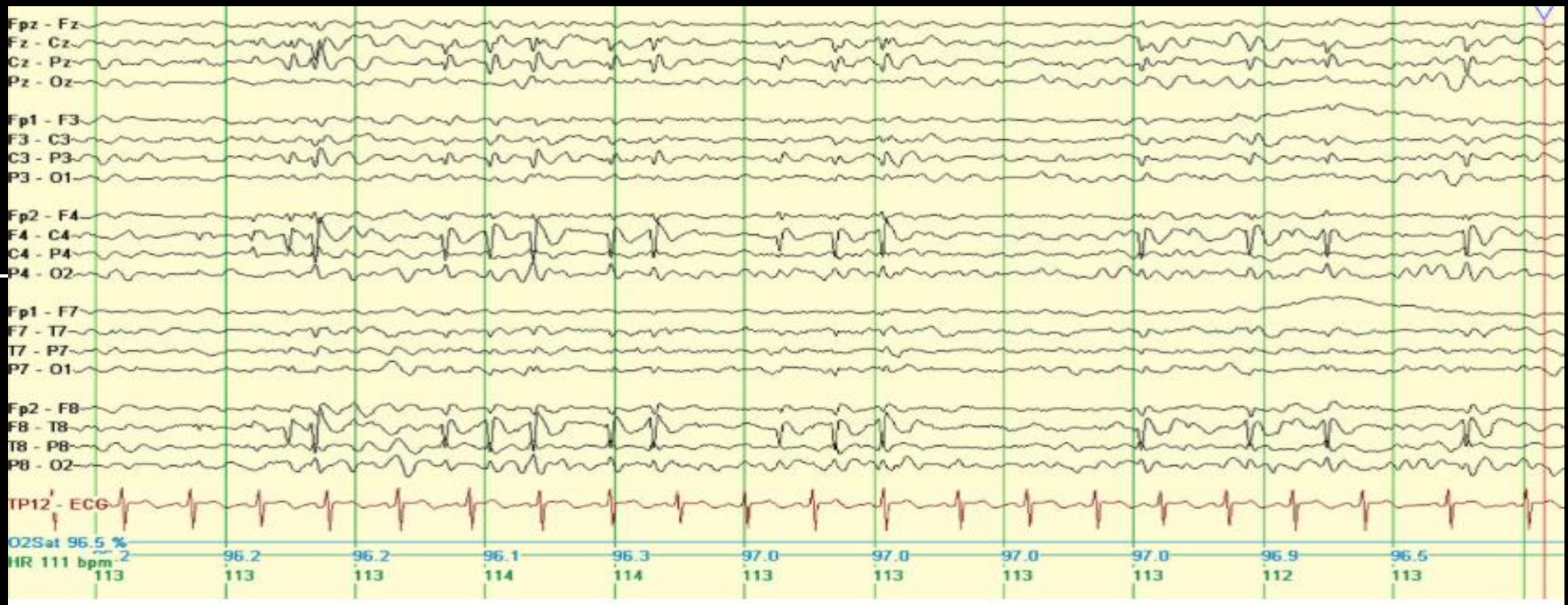
DESCARGAS ESPIGA-ONDA GENERALIZADAS A 3 HZ ASOCIADA A CLÍNICA DE AUSENCIAS

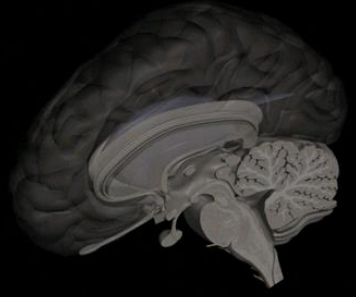


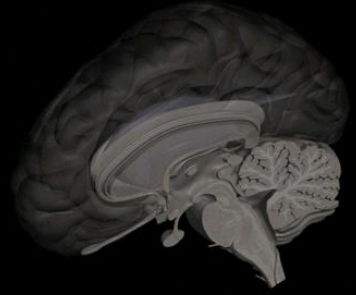
SÍNDROMES EPILÉPTICOS EN PEDIATRÍA

EPILEPSIA CON ESPIGAS CENTRO-TEMPORALES
(ROLÁNDICA O BENIGNA DE LA INFANCIA)

-NIÑOS DE 3-14 AÑOS (MÁXIMO 8-9 AÑOS) CRISIS HEMIFACIALES FRECUENTES EN EL DORMIR (A VECES CRISIS FOCALES O TCG NOCTURNAS)
-CLÁSICAMENTE DEFINIDA COMO AUTOLIITADA EN UNA EDAD ESPECÍFICA
-HABITUALMENTE OCURRE EN NIÑOS SANOS Y CON DSM NORMAL
-PUEDE RELACIONARSE A ALTERACIONES FUNCIONES EJECUTIVAS
-PATRÓN EEG CARÁCTERÍSTICO



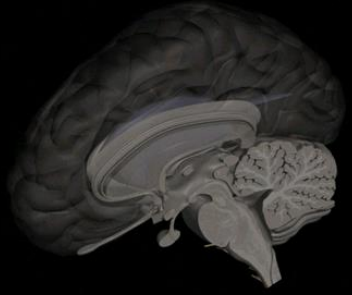


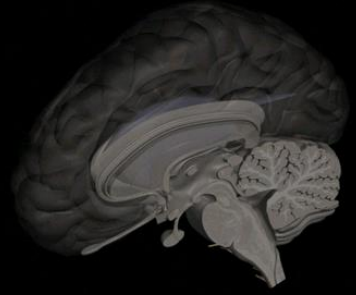


SÍNDROMES EPILÉPTICOS EN PEDIATRÍA

EPILEPSIA MIOCLÓNICA JUVENIL

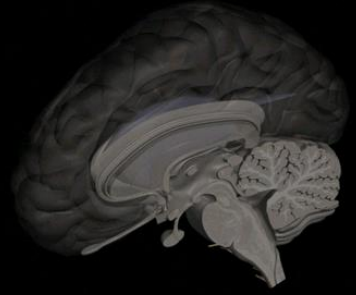
-UNA DE LAS EPILEPSIAS MÁS FRECUENTES EN JÓVENES Y ADULTOS
-DETERMINADAS GENÉTICAMENTE
-CON TENDENCIA A LA CRONICIDAD
-EL 5% TIENE EL ANTECEDENTE DE EPILEPSIA DE AUSENCIAS EN NIÑEZ
-CRISIS MIOCLÓNICAS Y GENERALIZADAS TÓNICO-CLÓNICAS, AUSENCIAS
-MIOCLONÍAS EVIDENCIADAS MÁS FRECUENTEMENTE AL DESPERTAR
-PATRÓN EEG CARACTERÍSTICO
-INICIO ENTRE LOS 8-15 AÑOS DE EDAD
-FOTOSENSIBILIDAD ES COMÚN
-SUJETOS SANOS, DSM Y NIVEL INTELECTUAL NORMALES
-EXACERBACIÓN POR CIERTOS FÁRMACOS ANTICONVULSIVANTES (CBZ)



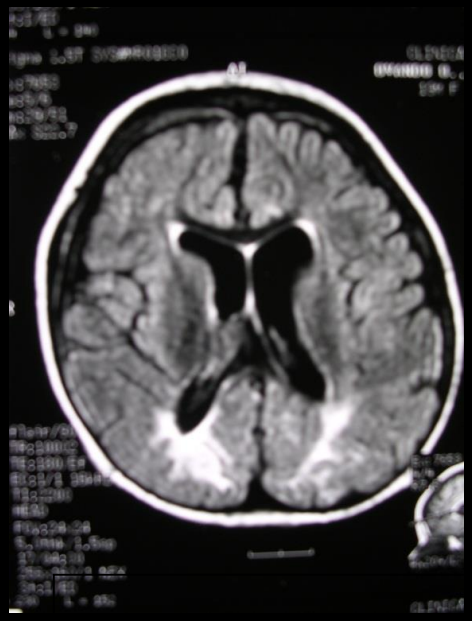


CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA – ESTUDIO

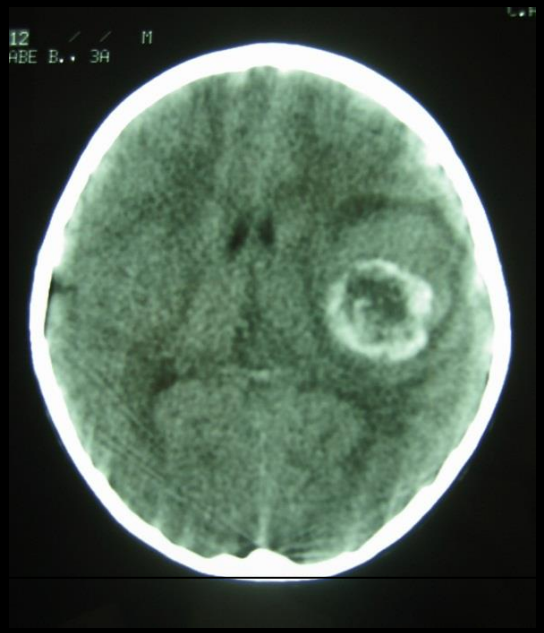
-HISTORIA CLÍNICA DETALLADA
-HISTORIA FAMILIAR
-ANTECEDENTES PERINATALES-DESARROLLO NEUROLÓGICO
-EEG ESTÁNDAR, CON PRIVACIÓN DE SUEÑO (PARCIAL O TOTAL)
-VIDEO-MONITOREO EEG
-HOLTER EEG
-RMN CEREBRAL-TAC CEREBRAL
-PET SCAN
-EVALUACIONES NEUROPSICOLÓGICAS (WISC-EMOCIONAL-PEDAGÓGICAS)
-EXÁMENES GENERALES
-ESTUDIOS GENÉTICOS
-ESTUDIOS METABÓLICOS (EIM)



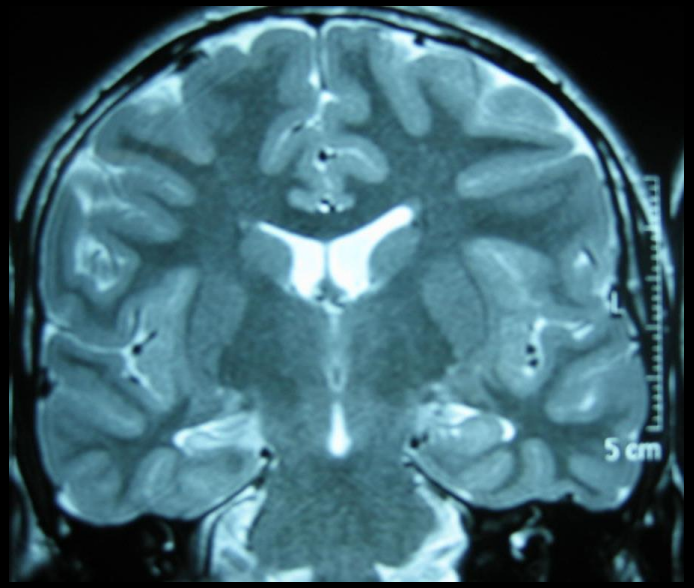
ESTUDIO NEUROIMÁGENES



GLIOSI$ SUSTANCIA BLANCA POSTERIOR

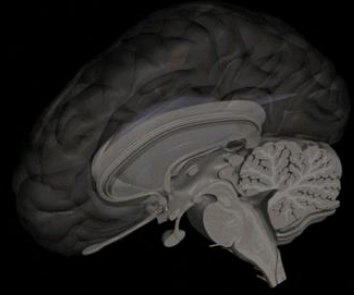


PROCESO EXPANSIVO HEMISFÉRICO



CORTES CORONALES EN T2 HIPOCAMPOS

ESTUDIO GENÉTICO EN EPILEPSIA



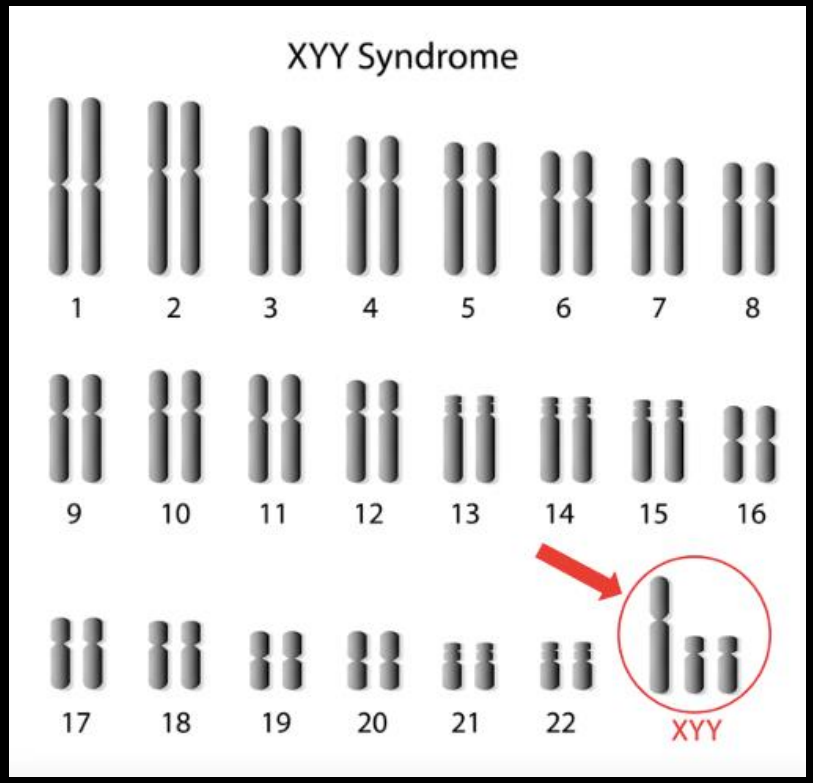
Thomas, R. H. & Berkovic, S. F. *Nat. Rev. Neurol.* advance online publication 15 April 2014;

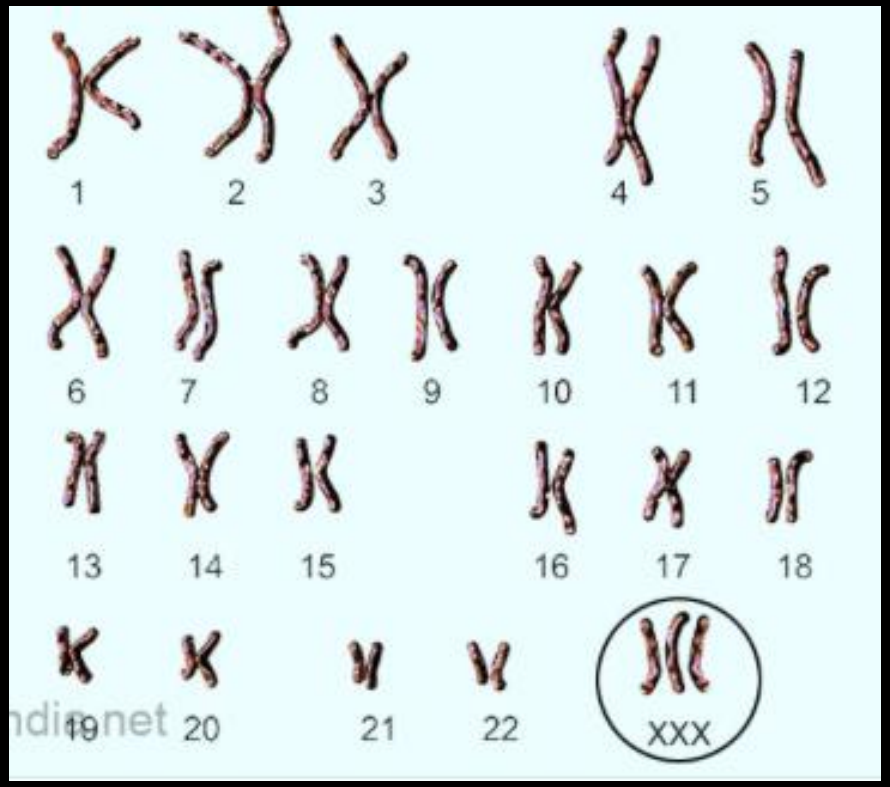
Table 1 | Genetic testing in patients with epilepsy

Genetic test	Detects	Indication	Consequence
Current relevance			
CGH array[42,43]	Copy number variation	Epilepsies with (comorbid) intellectual disability, autism or dysmorphic features	Counselling
Karyotyping[112,113]	Large-scale chromosomal abnormality	Suspected ring chromosome 20	Diagnosis
Gene panel[49,65]	Known mutations in (typically) 20–25 genes	Epileptic encephalopathies	Diagnosis
HLA typing[109]	Risk allele associated with hypersensitivity to carbamazepine	Probable Han Chinese or South Asian ethnic origin	AED choice
SCN1A[54,55]	Mutations in sodium channel type 1α	Dravet syndrome	Diagnosis AED choice
SLC2A1[103–106]	Mutations in GLUT1	GLUT1 encephalopathy Early-onset childhood absence epilepsy Paroxysmal exertional dyskinesia	Ketogenic diet Diagnosis
PCDH19[61,92]	Mutations in protocadherin 19	Female patients with clustered focal seizures (onset under 3 years of age) and often with intellectual disability	Counselling
Probable future relevance			
GRIN2A[73–75]	Mutations in NMDA receptor subunit ε1	Landau–Kleffner syndrome Epilepsy–aphasia spectrum disorders	Counselling
DEPDC5[69,70]	Mutations in DEP domain-containing 5	Familial focal epilepsy with variable foci Most common mutation in focal epilepsy	Counselling
KCNT1[76,77]	Mutations in potassium channel, subfamily T, member 1	Autosomal dominant nocturnal frontal lobe epilepsy Epilepsy in infancy with migrating focal seizures	Counselling

Seven widely available genetic tests have important management consequences that every neurologist should know about, and another three are likely to become of clinical importance soon. Abbreviations: AED, antiepileptic drug; CGH, comparative genomic hybridization; GLUT1, glucose transporter protein type 1; HLA, human leukocyte antigen; NMDA, *N*-methyl-D-aspartate receptor.

ESTUDIO GENÉTICO EN EPILEPSIA: CARIOGRAMA





ESTUDIO GENÉTICO EN EPILEPSIA: CARIOTIPO MOLECULAR

Indication: Developmental Delay, Seizure Disorder

ABNORMAL Microarray Result, Male
Pathogenic 1.24 Mb Deletion of 16p13.11
16p13.11 Microdeletion Syndrome

****FISH Testing Not Possible--See Comments Below****

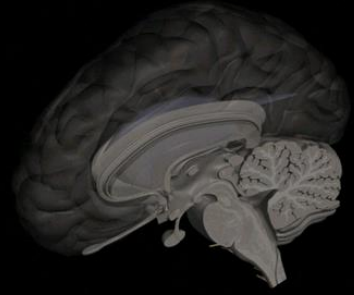
This analysis detected a deletion of the short arm of chromosome 16, approximately 1.24 Mb in size. This deletion contains at least 21 genes and is within the 16p13.11 Microdeletion syndrome region (see abnormality details below for complete list of genes). Deletions of this region are associated with a highly variable phenotype that may include intellectual disability, autism, multiple congenital anomalies, and epilepsy. Penetrance may not be complete as deletions have also been observed in apparently normal parents (see references below).

These microarray results provide information about the size and gene content of the copy change, but do not provide information about the chromosomal structure of the aberration. Although this alteration likely represents an interstitial deletion of 16p, FISH analysis is recommended to visualize this alteration and exclude the rare possibility that it is part of a more complex unbalanced rearrangement. Note that FISH is not possible on the EDTA blood specimen submitted. For FISH analysis at no additional charge to the patient, please submit a secondary peripheral blood specimen, collected in sodium heparin, to this laboratory.

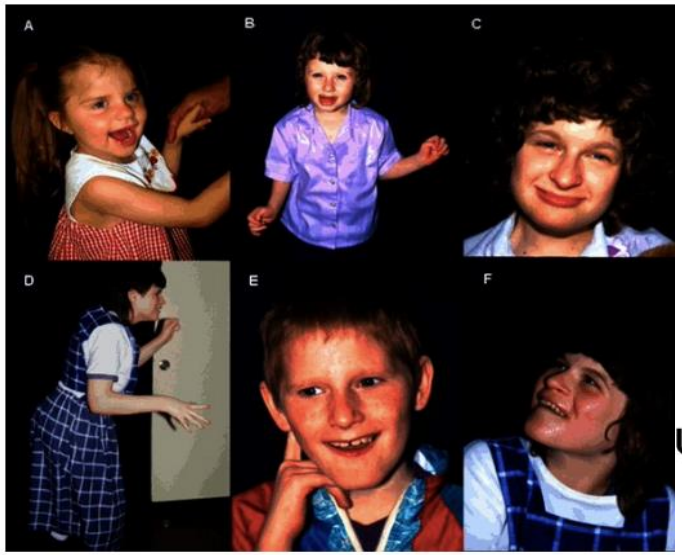
Clinical correlation is required. Genetic counseling and parental testing are recommended for this family.

Microarray Result: arr[GRCh37] 16p13.11(15049829_16287899)x1

ESTUDIO GENÉTICO EN EPILEPSIA: ESTUDIO DE UN GEN ESPECÍFICO

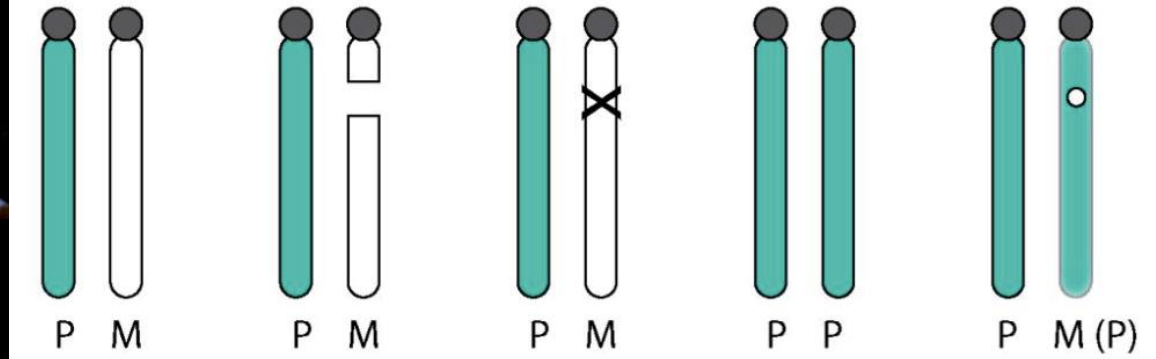


Individuals with Angelman syndrome

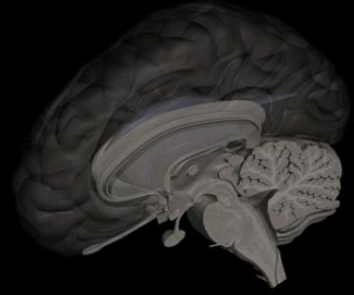


Credit: GeneReviews, © 1993–2020 University of Washington

Genetic Classes of AS



ESTUDIO GENÉTICO EN EPILEPSIA: PANEL DE EPILEPSIA



INVITAE

Name	DOB
Lucas Palacios	12.22.2016

Patient Name	DOB	Sex	MRN	Invitae #
	12.22.2016	Male		RQ306892

Clinical Team	Report Date	Sample Type	Sample Collection Date	Sample Accession Date
Genometrics Chile Jovanka Pavlov	03.08.2018	Saliva	02.14.2018	02.21.2018

Test Performed
Sequence analysis and deletion/duplication testing of the 189 genes listed in the results section below.

- Add-on FLNA Gene
- Add-on Genes for Glycine Encephalopathy
- Add-on PTEN Gene
- Add-on Preliminary-evidence Genes for Epilepsy
- Add-on RANBP2 Gene
- Invitae Epilepsy Panel

Reason for Testing
Diagnostic test for a personal history of disease

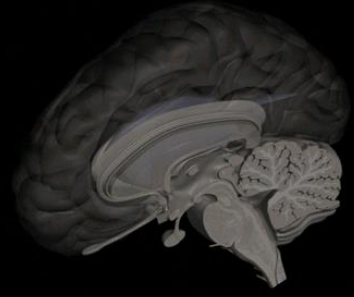
Summary

Positive result. Pathogenic variant identified in SCN1A.
Variants of Uncertain Significance identified in GLDC, KCNH2 and SCN5A.

Clinical Summary

- A Pathogenic variant, c.2593C>T (p.Arg865*), was identified in SCN1A.
  - The SCN1A gene is associated with a spectrum of autosomal dominant seizure disorders ranging from simple febrile seizures (MedGen UID: 338959) and genetic epilepsy with febrile seizures plus (GEFS+) (MedGen UID: 388117) to Dravet syndrome (MedGen UID: 148243) and intractable childhood epilepsy with generalized tonic-clonic seizures (ICE-GTC) (MedGen UID: 148243). Other SCN1A-related conditions have been reported (OMIM: 607208).
  - This result is consistent with a predisposition to, or diagnosis of, SCN1A-related conditions.
  - SCN1A-related conditions are seizure disorders with varying severity and early childhood onset. Febrile seizures are childhood seizures that occur with fever and often resolve by six years of age. GEFS+ is also characterized by febrile seizures; however with GEFS+, both febrile and afebrile seizures may continue throughout an affected individual's lifetime. Dravet syndrome is one of the most severe seizure disorders and is characterized by intractable seizures and usually associated with progressive dementia. ICE-GTC is considered a late-onset Dravet syndrome. Intrafamilial variability in seizure type, persistence, and response to treatment has been documented, as has reduced penetrance (http://www.orpha.net/data/patho/GB/uk-GEFS.pdf).
  - Close relatives (children, siblings, and parents) have up to a 50% chance of being a carrier of this variant. More distant relatives may also be carriers. Parental testing may clarify the inheritance of this variant and may inform recurrence risk and risk for other close relatives. Testing for this variant is available.

ESTUDIO GENÉTICO EN EPILEPSIA: EXOMA COMPLETO



CENTOGENE
THE RARE DISEASE COMPANY

CENTOGENE AG AmStrande 7 • 18055 Rostock • Germany

F. nacimiento: **04 abr. 2018**, Sexo: **femenino**, Ref. externa: **-**

Prueba(s) solicitada(s): CentoXome® Solo

INFORMACIÓN CLÍNICA

Anormalidad en el EEG; Atrofia cerebral; Convulsiones; Forma facial anormal; Microcefalia; Retardo global del desarrollo; Retraso del lenguaje y la comunicación; Talla baja
(Información clínica reportada atendiendo a la nomenclatura HPO.)

Pruebas realizadas anteriormente en CENTOGENE con resultados negativos: Análisis cromosómico de microarray (Nº pedido 62666323).

Historia familiar: Sí.
Madre: Aborto espontáneo Hermanos no afectados.
Padres consanguíneos: No.

Diagnóstico de sospecha: síndrome de Cornelia de Lange.

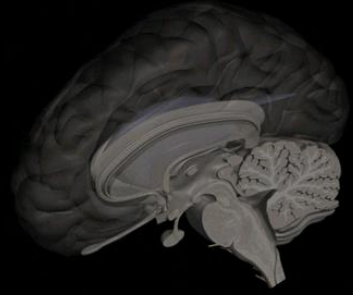
RESULTADO POSITIVO
Variante patogénica identificada

INTERPRETACIÓN

Se identificó una variante patogénica en heterocigosis en el gen DYRK1A. **El resultado confirma el diagnóstico genético del retraso mental autosómico dominante tipo 7.**

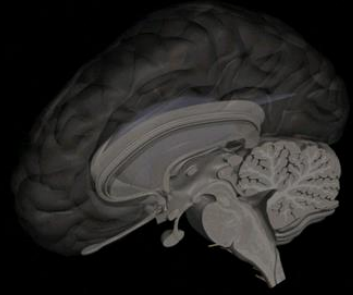
RECOMENDACIONES

- Se recomienda la prueba del portador a los padres para establecer si la variante detectada es heredada o *de novo*.
- Se recomienda asesoramiento genético.



CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA
ENFRENTAMIENTO CLÍNICO FRENTE A PRIMERA CRISIS CONVULSIVA

-HISTORIA CLÍNICA DETALLADA
-HISTORIA FAMILIAR
-ANTECEDENTES PERINATALES-DESARROLLO NEUROLÓGICO
-EEG ESTÁNDAR, CON PRIVACIÓN DE SUEÑO (PARCIAL O TOTAL)
-VIDEO-MONITOREO EEG
-HOLTER EEG
-RMN CEREBRAL-TAC CEREBRAL
-PET SCAN
-EVALUACIONES NEUROPSICOLÓGICAS (WISC-EMOCIONAL-PEDAGÓGICAS)
-EXÁMENES GENERALES
-ESTUDIOS GENÉTICOS
-ESTUDIOS METABÓLICOS (EIM)



CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA
ENFRENTAMIENTO CLÍNICO FRENTE A PRIMERA CRISIS CONVULSIVA

-PACIENTE PREVIAMENTE SANO, DSM (N), EXAMEN NEUROLÓGICO (N), EEG Y NEUROIMÁGENES NORMALES: OBSERVAR SIN FÁRMACOS, POSIBILIDAD DE RECURRENCIA DE CRISIS 50%
ENTRENAR A FAMILIA EN MANEJO DE CRISIS
EVITAR FACTORES DESENCADENANTES O PREDISPONENTES

-CLÍNICA ASUMIBLE DENTRO DEL CONTEXTO DE UN SÍNDROME EPILÉPTICO: TRATAMIENTO CON FÁRMACOS ANTI EPILÉPTICOS (FAE)

-PACIENTE EN QUE SE ASUME QUE POR SUS CARÁCTERÍSTICAS DE BASE O HALLAZGOS CLÍNICOS O DE LABORATORIO, TENGA UNA POSIBILIDAD MAYOR DE RECURRIR: INICIO DE TRATAMIENTO CON FAE

CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA

ENFRENTAMIENTO CLÍNICO FRENTE A PRIMERA CRISIS CONVULSIVA

-EXÁMENES GENERALES
-ESTUDIO EEG
-NEUROIMÁGENES → DECISIÓN DE INICIO DE TRATAMIENTO

↓

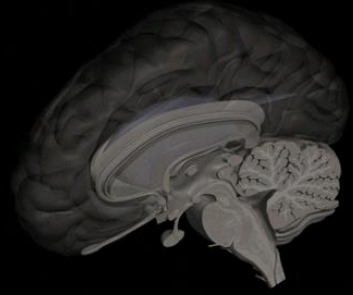
FÁRMACOS ANTIEPILÉPTICOS DOS AÑOS SIN CRISIS Y EEG(N):
DECISIÓN DE SUSPENSIÓN (2/3 BUENA EVOLUCIÓN)

↓

RECAÍDAS SE DAN MAYORMENTE EN LOS PRIMEROS 6 MESES POSTERIORES A SUSPENSIÓN DE FAE:
RIESGO ACUMULADO DE RECURRENCIA, PRIMER AÑO 25% -SEGUNDO AÑO 29%)

↓

CRITERIO DE EPILEPSIA CURADA: 10 AÑOS SIN CRISIS Y AL MENOS 5 SIN FAE



CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA

EPILEPSIA REFRACTARIA

-CUADRO EPILÉPTICO CON MANTENCIÓN DE CRISIS PESE AL USO CORRECTO DE DOS FÁRMACOS ANTIEPILÉPTICOS (FAE) DE DISTINTO MECANISMO DE ACCIÓN EN LAS DOSIS Y CON LAS INDICACIONES ADECUADAS PARA ESE PACIENTE EN PARTICULAR

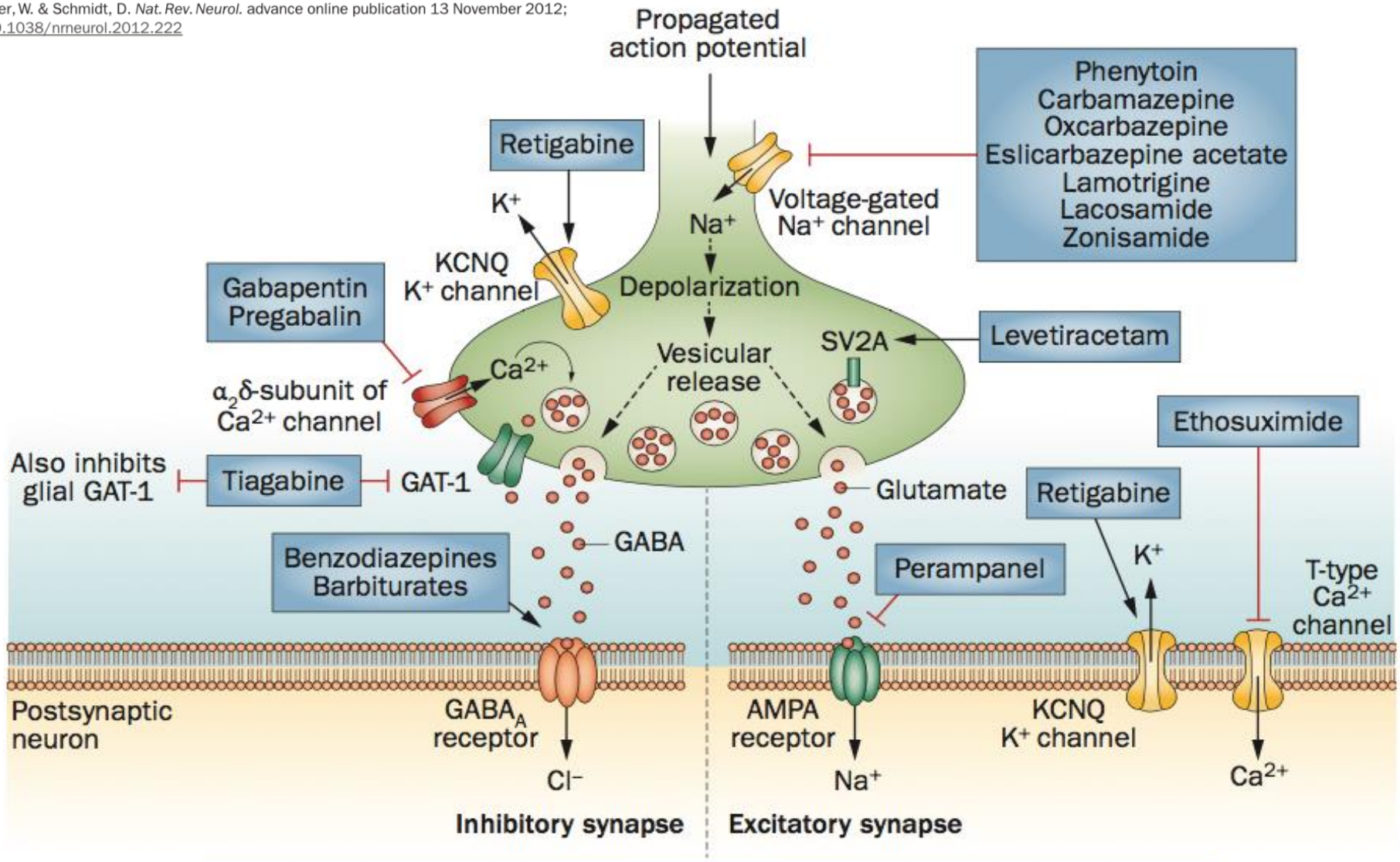
-REPRESENTA AL 20-25% DE LOS PACIENTES CON EPILEPSIA

OBSERVACIONES:

-CONSIDERAR SIEMPRE EN ESTOS CASOS QUE EL DIAGNÓSTICO DE EPILEPSIA NO SEA CORRECTO Y ESTEMOS TRATANDO CON FAE OTRO TIPO DE CUADRO

-REQUIERE DERIVACIÓN Y MANEJO EN CENTRO DE REFERENCIA EN EPILEPSIA

Löscher, W. & Schmidt, D. *Nat. Rev. Neurol.* advance online publication 13 November 2012; doi:10.1038/nrneurol.2012.222



Not illustrated:

- Vigabatrin → ↓ GABA degradation

and drugs with multiple mechanisms:

- Valproate → ↑ GABA turnover, ↓ Na^+ channels, ↓ NMDA receptors
- Topiramate → ↓ Na^+ channels, ↓ AMPA/kainate receptors, ↑ $GABA_A$ receptors
- Felbamate → ↓ Na^+ channels, ↑ $GABA_A$ receptors, ↓ NMDA receptors

DESARROLLO DE FÁRMACOS ANTICONVULSIVANTES

M.J. Brodie, G.J. Sills / Seizure 20 (2011) 369–375

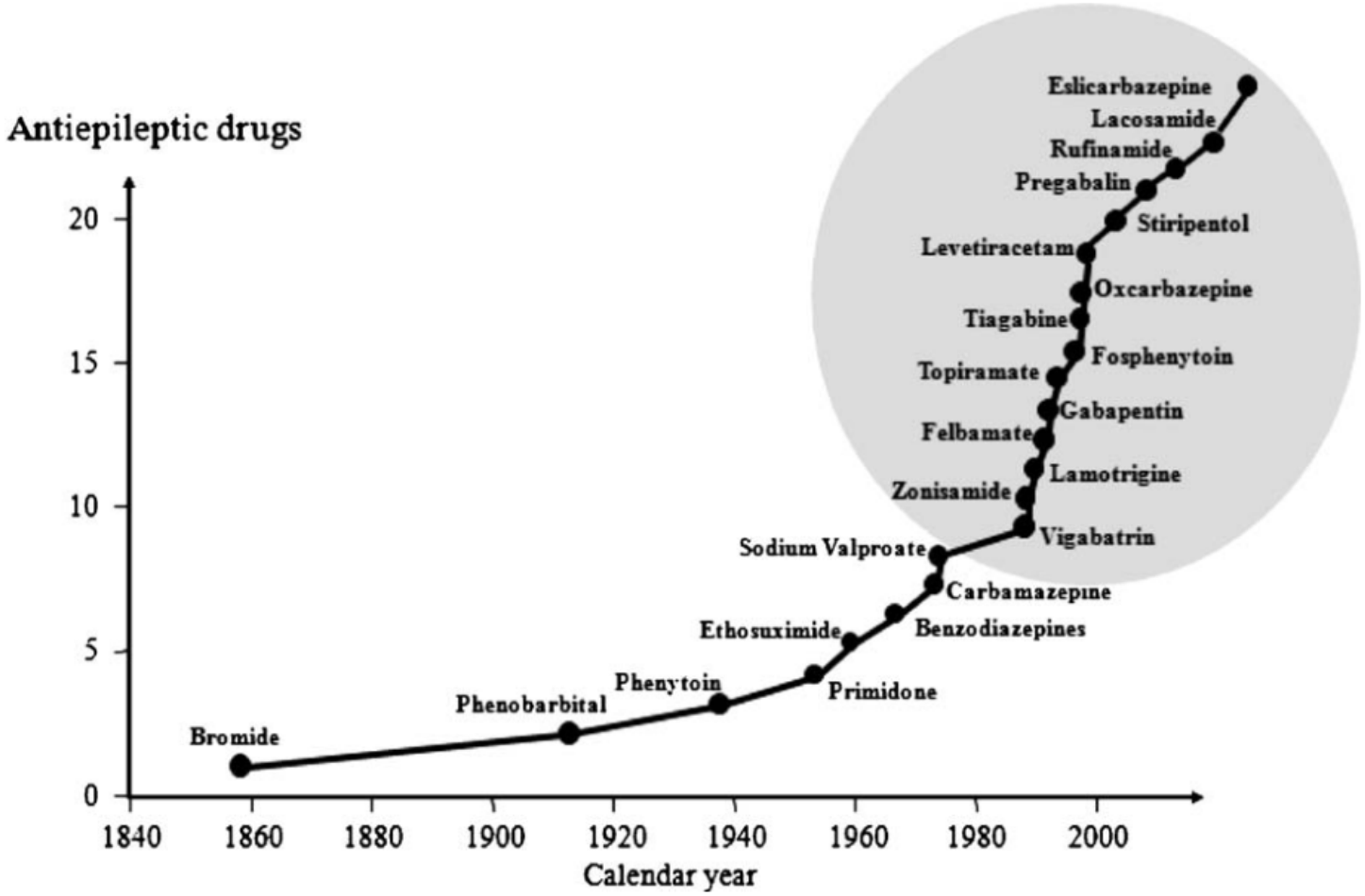
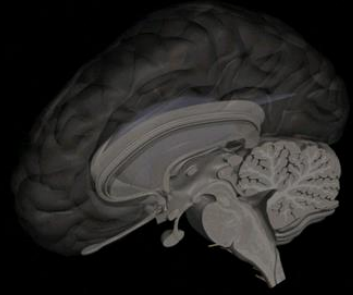


Fig. 1. Chronology of antiepileptic drug introduction over the past 150 years.

CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA: FÁRMACOS ANTIEPILÉPTICOS



-FENOBARBITAL (depresión respiratoria)
-FENITOINA (vo NO en Pediatría, arrítmias)
-CARBAMAZEPINA (agrava mioclonías, ↓Na)
-ÁCIDO VALPROICO (hepatotox, teratóg)
-PRIMIDONA
-ETOSUXIMIDA
-CLOBAZAM
-CLONACEPAM
-GABAPENTINA
-LAMOTRIGINA (alergia)
-OXCARBAMAZEPINA (idem CBZ)
-TOPIRAMATO (litiasis)
-LACOSAMIDA
-LEVETIRACETAM (psicosis, irritabilidad)
-VIGABATRINA
-ACTH
-PREGABALINA
-PERAMPANEL
-SULTIAME
-STIRIPENTOL
-RUFINAMIDA

CONCEPTOS GENERALES:
-INSTALACIÓN LENTA
-SUSPENSIÓN LENTA
-CONOCER EFECTOS ADVERSOS
-NIVELES PLASMÁTICOS
-MONITOREO CLÍNICO-LABORATORIO
-REACCIONES ADVERSAS
-RIESGOS TERAPIAS ASOCIADAS
-REACCIONES IDIOSINCRÁTICAS

PRIMER FAE: CONTROL CRISIS 50%
SEGUNDO FAE: CONTROL CRISIS 63%
TERCER-CUARTO FAE: PLUS 3-4%

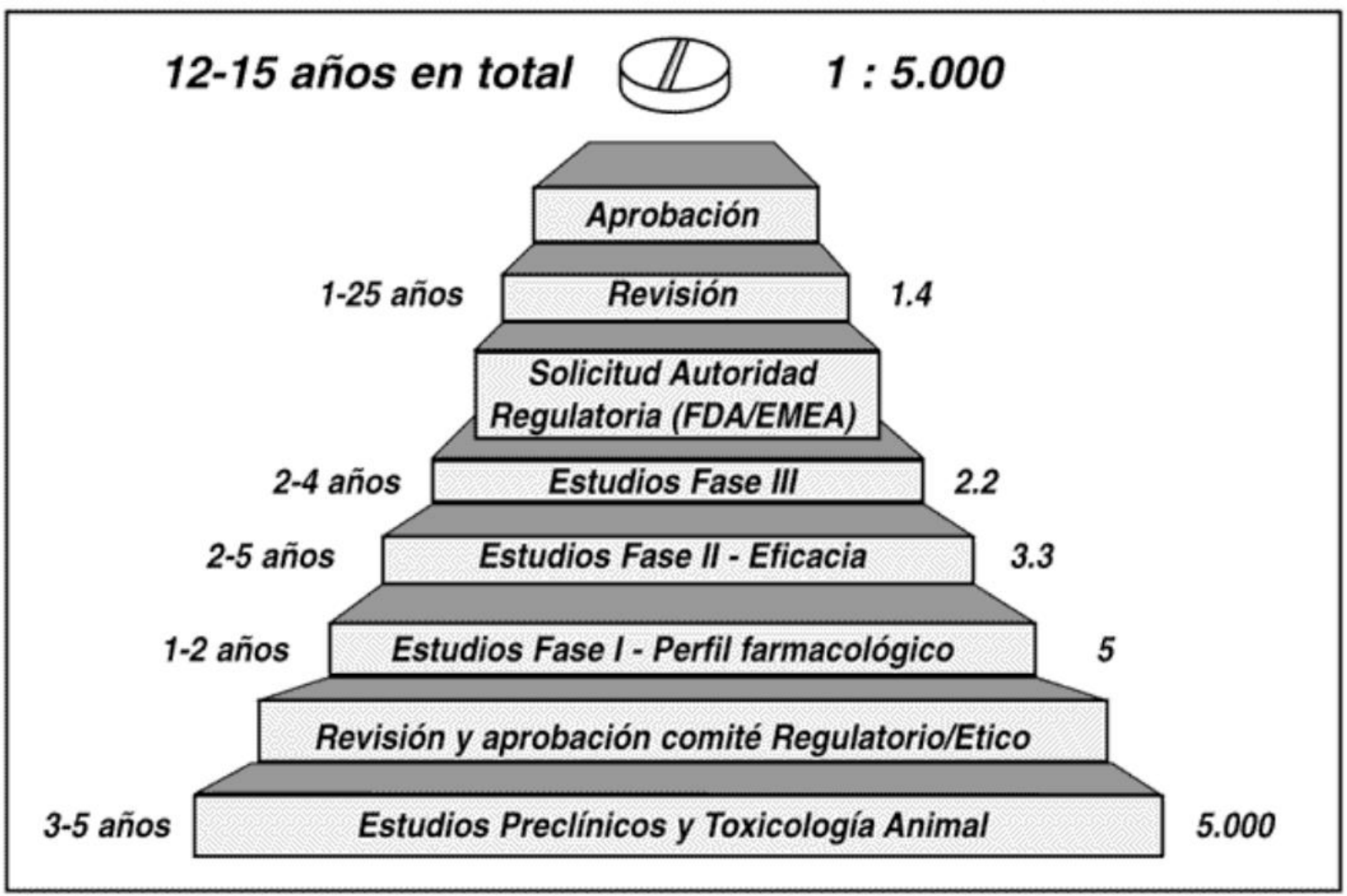
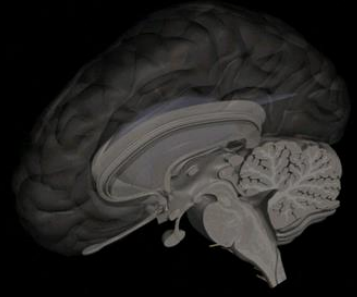
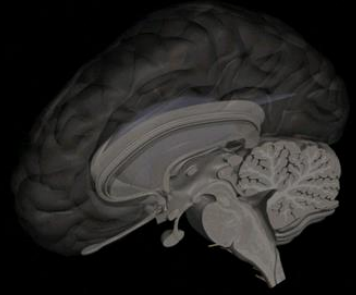


FIGURA 2. Proceso de descubrimiento, desarrollo y aprobación de drogas.

TRATAMIENTOS DISPONIBLES EPILEPSIA 2022



a. Acetazolamide (Diamox, Diamox Sequels)
b. ACTH
c. Brivaracetam (Briviact)
d. Cannabidiol (Epidiolex)
e. Carbamazepine (Carbatrol, Epitol, Equetro, Tegretol, Tegretol XR)
f. Clobazam (Onfi, Frisium)
g. Clonazepam (KlonoPIN)
h. Eslicarbazepine acetate (Aptiom)
i. Ethosuximide (Zarontin)
j. Felbamate (Felbatol)
k. Immunoglobulins
l. Lacosamide (Vimpat)
m. Lamotrigine (Lamictal)
n. Levetiracetam (Keppra, Roweepra, Spritam)
o. Nitrazepam
p. Oxcarbazepine (Oxtellar XR, Trileptal)
q. Perampanel (Fycompa)
r. Phenobarbital (Solfoton, Luminal)
s. Phenytoin (Dilantin, Phenytek)
t. Prednisone (Rayos, Sterapred, Deltasone)
u. Pyridoxine (Vitamin B6, Vitelle Nestrex, Pyridoxal 5'-Phosphate)
v. Retigabine (Potiga)
w. Rufinamide (Banzel)
x. Steroids
y. Stiripentol (Diacomit)
z. Sulthiame
aa. Topiramate (Topamax, Qudexy XR Sprinkle, Topamax Sprinkle, Trokendi XR, Topiragen)
bb. Valproate
cc. Vigabatrin (Sabril, Vigadrone)
dd. Zonisamide (Zonegran)
ee. Atkins diet
ff. Casein free diet
gg. Gluten free diet
hh. Ketogenic diet
ii. Corpus callosotomy
jj. Gamma knife radiosurgery
kk. Hemispherectomy
ll. Lobectomy
mm. Resection surgery
nn. Subpial transection
oo. Vagal nerve stimulator (VNS) surgery



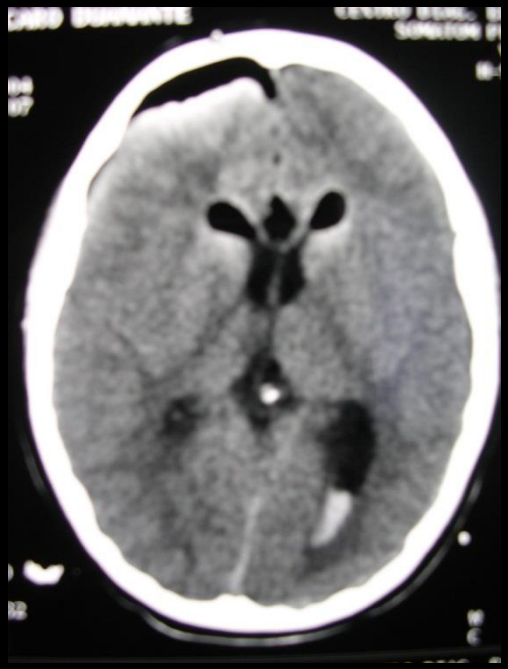
CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA
TERAPIAS NO FARMACOLÓGICAS

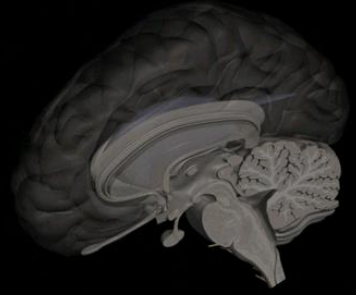
-DIETA CETOGÉNICA

-CIRUGÍA DE LA EPILEPSIA

-ESTIMULADOR VAGAL

-BIO-FEEDBACK





CRISIS CONVULSIVAS EN LA INFANCIA
EPILEPSIA
SECUELAS

-MUERTE

-SUDEP

-DAÑO NEUROLÓGICO

-DETERIORO INTELECTUAL

-ESTIGMA SOCIAL

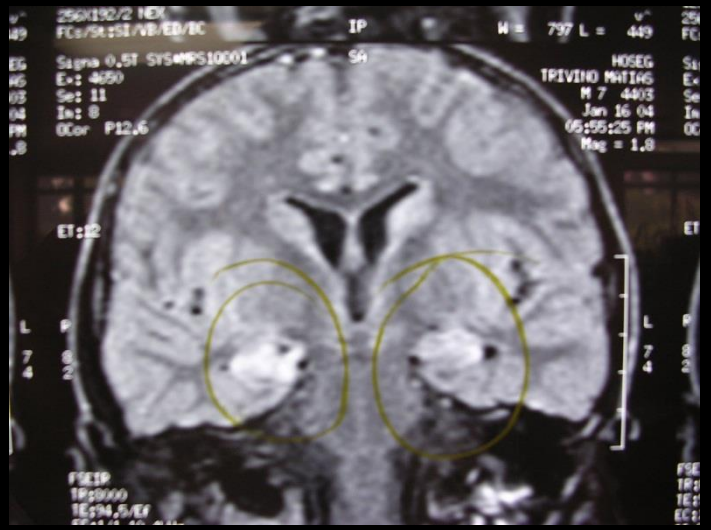
-COMPROMISO AUTOESTIMA-VIVENCIA

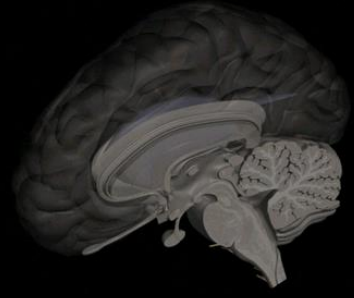
-”MALCRIANZA”

-COMPROMISO CAPACIDADES APRENDIZAJE

-PSICOPATOLOGÍA

-SOCIO-ECONÓMICAS





- Overview
  - Video Help
  - Give Feedback
- Seizure Classification
  - Generalized seizures
  - Focal seizures
  - Focal/Generalized
- Epilepsy syndromes
  - Neonatal/Infantile
  - Childhood
  - Adolescent/Adult
  - Variable Age
- Epilepsies by Etiology
  - Genetic
  - Structural
  - Metabolic
  - Immune
  - Infectious
  - Unknown
- Unclassified epilepsies
- Epilepsy imitators

EpilepsyDiagnosis.org

The ILAE Commission on Classification and Terminology welcomes you to EpilepsyDiagnosis.org, a cutting edge online diagnostic manual of the epilepsies.

Goal

The goal of ***epilepsydiagnosis.org*** is to make available, in an easy to understand form, latest concepts relating to seizures and the epilepsies. The principle goal is to assist clinicians who look after people with epilepsy anywhere in the world to diagnose seizure type(s), diagnose epilepsy syndromes and define the etiology of the epilepsy. The site is principally designed for clinicians in primary and secondary care settings caring for people with epilepsy and we hope will also serve as a useful teaching aid.

Structure

The structure of this site reflects the importance of seizure type, syndrome, and etiology in clinical practice. On this website, you will find current classification concepts for seizures, with their clinical features, video examples, EEG correlate, differential diagnosis and related epilepsy syndromes. Epilepsy syndromes are detailed by their clinical features, seizure types, EEG, imaging and genetic correlates and differential diagnoses. The site includes sections on etiologies of epilepsies and epilepsy imitators with cross-referencing between these sections and seizure and syndrome sections.

Definition of epilepsy

Epilepsy is a disease of the brain defined by any of the following conditions:

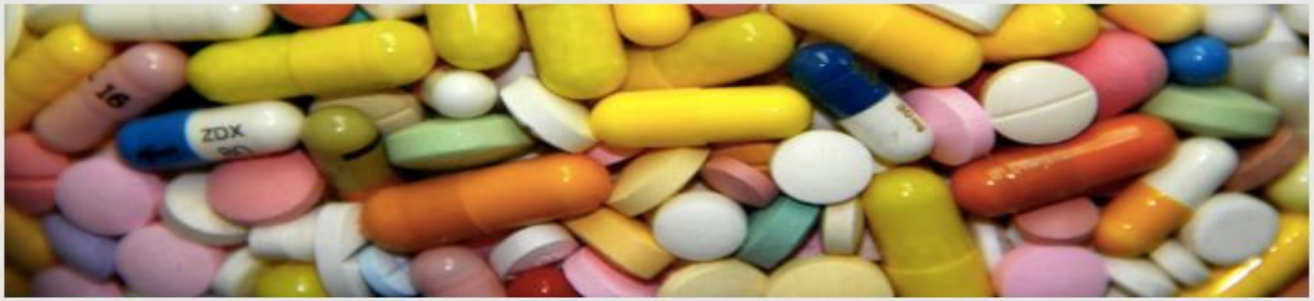
- At least two unprovoked (or reflex) seizures occurring more than 24 hours apart
- One unprovoked (or reflex) seizure and a probability of further seizures similar to the general recurrence risk (at least 60%) after two unprovoked seizures, occurring over the next 10 years
- Diagnosis of an epilepsy syndrome

LISTA DE MEDICAMENTOS
- CONSULTA DE MEDICAMENTOS
- AGOTADOS, DISCONTINUADOS
- INCORPORADOS

MEDICAMENTOS SUJETOS A CONTROL DE SALDO >
CÓDIGOS DE PSICOTRÓPICOS
SOLICITUD DE INSCRIPCIÓN MÉDICA >
SOLICITUD DE RECETARIOS >
ENLACES MÉDICOS >
EVENTOS CIENTÍFICOS >
PLAN ESTRATÉGICO DE ACCIÓN DE EPILEPSIA >
CENTRO DE DOCUMENTACIÓN >

Info Médicos

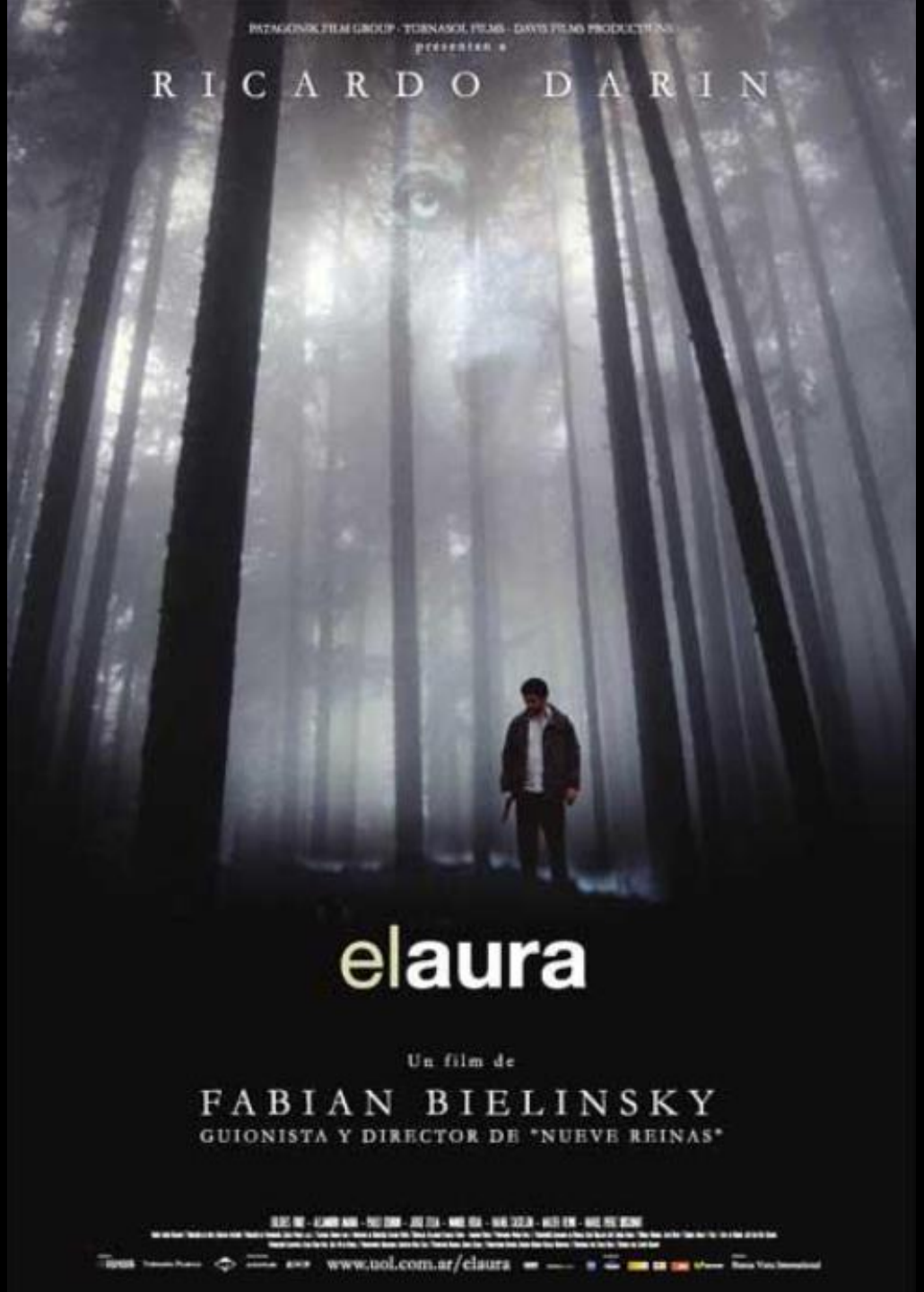
Lista de Medicamentos



La Liga Chilena contra la Epilepsia cuenta con la mayoría de los antiepilépticos existentes en el país y otros medicamentos de uso neurológico y psiquiátrico para las personas con epilepsia y la comunidad en general.

Busque su medicamento aquí.

Para **mayor información** llame al teléfono (56) 226967281 o al 600 300 1515 (desde Regiones).


PATAGONIK FILM GROUP - TORNASOL FILMS - DAVIS FILMS PRODUCTIONS
presentan a
RICARDO DARIN
elaura
Un film de
FABIAN BIELINSKY
GUIONISTA Y DIRECTOR DE "NUEVE REINAS"
www.uol.com.ar/elaura