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Epilepsy Care

for Rare and Not Rare

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Rare Disease Day Conference 2025

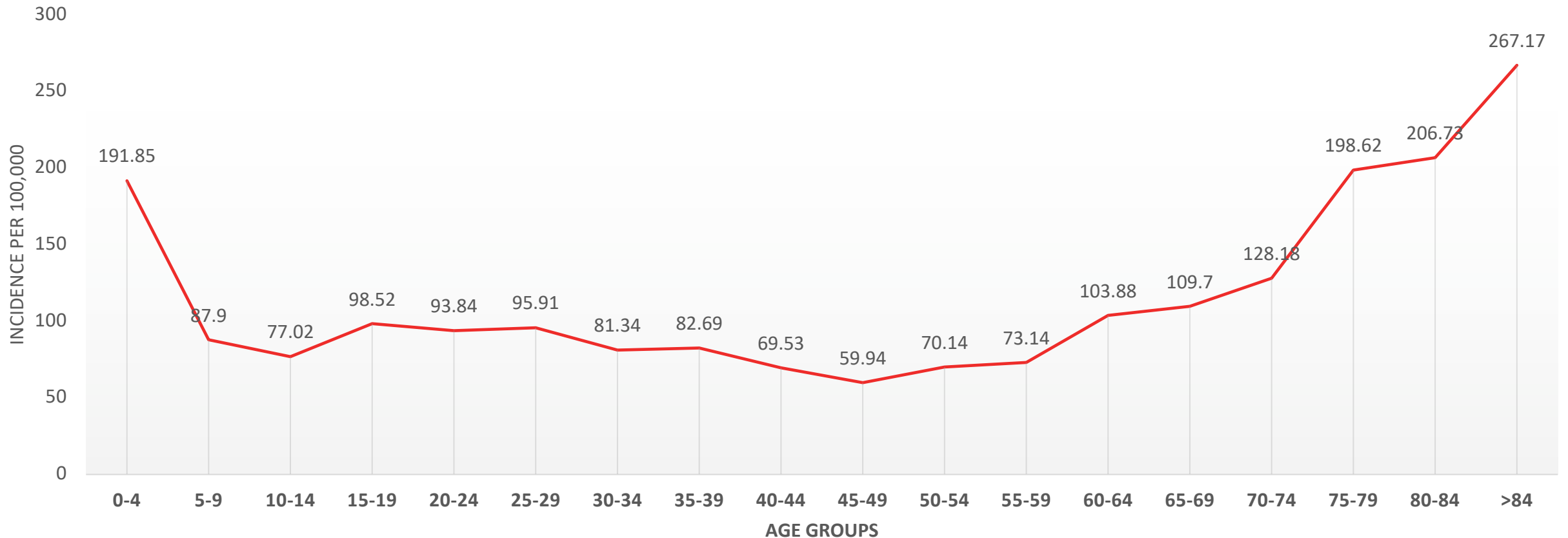
Key Facts

- 50 million people worldwide have epilepsy
- 5 million people are diagnosed with epilepsy each year
- One of the most common neurological diseases globally
- 80% of people with epilepsy (PwE) live in low- and middle-income countries
 - $\frac{3}{4}$ of PwE do not get the treatment they need
- Fear, misunderstanding, discrimination and social stigma



Newly diagnosed patients in Alberta

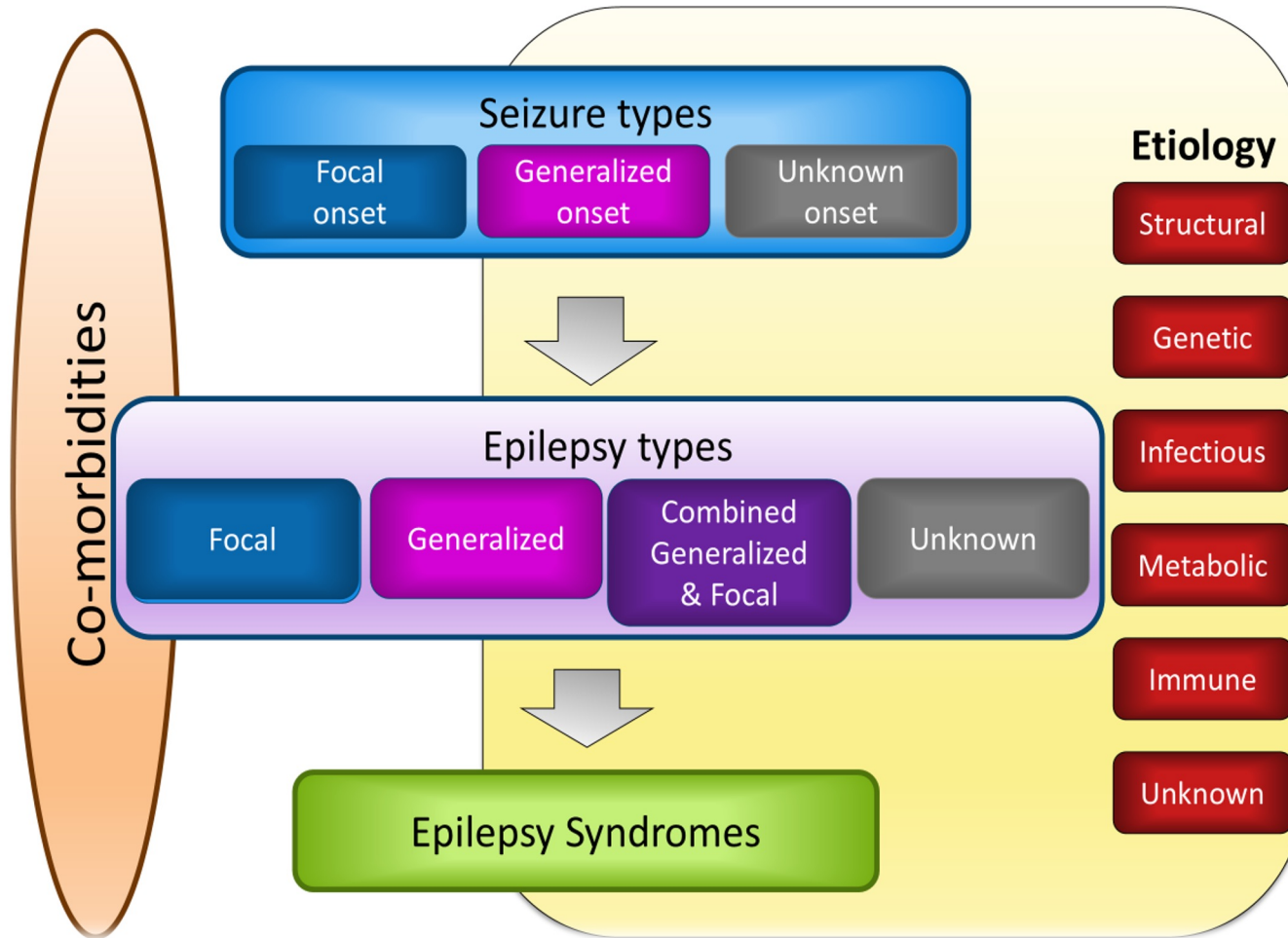
- Incidence in 2023: 99.93 per 100,000



What is Epilepsy?

- Epilepsy is the tendency to recurrent seizures
 - I. Two unprovoked seizures occurring >24 h apart
 - II. One unprovoked + probability of further seizures at least 60% in 10 years
 - III. Epilepsy syndrome
- Not a single diagnosis, but a symptom for many different causes
- > **140 rare diseases** may present with epilepsy as the major symptom

International League Against Epilepsy (ILAE)

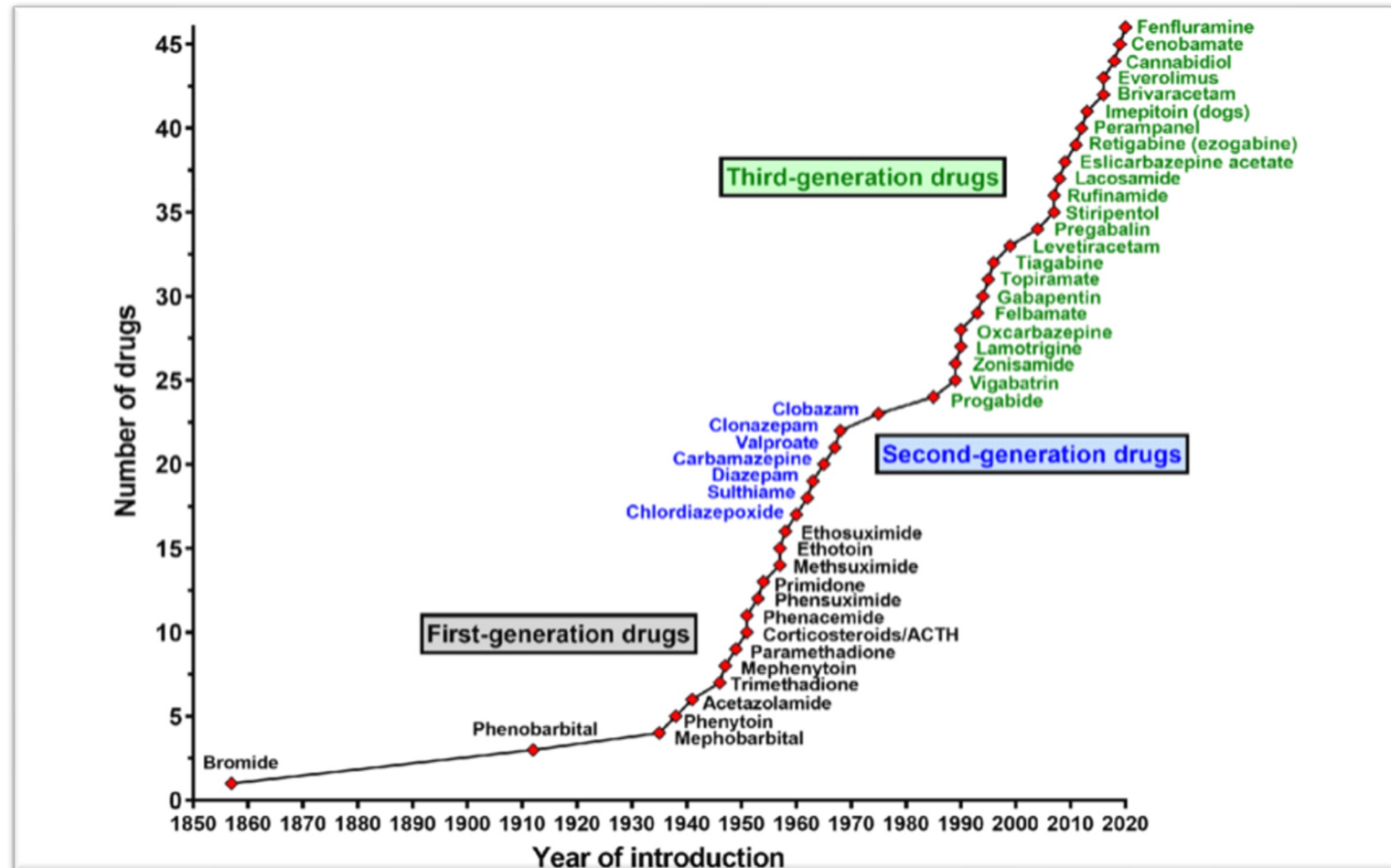


- Brain damage from prenatal or perinatal causes
- Meningitis, encephalitis or neurocysticercosis
- Stroke
- Brain tumours
- Autoimmune encephalitis
- Genetically-determined



Treatment approach

- Focal vs. generalized
- 60-70% of people living with epilepsy could become seizure free with appropriate use of antiseizure medicines
- Epilepsy surgery



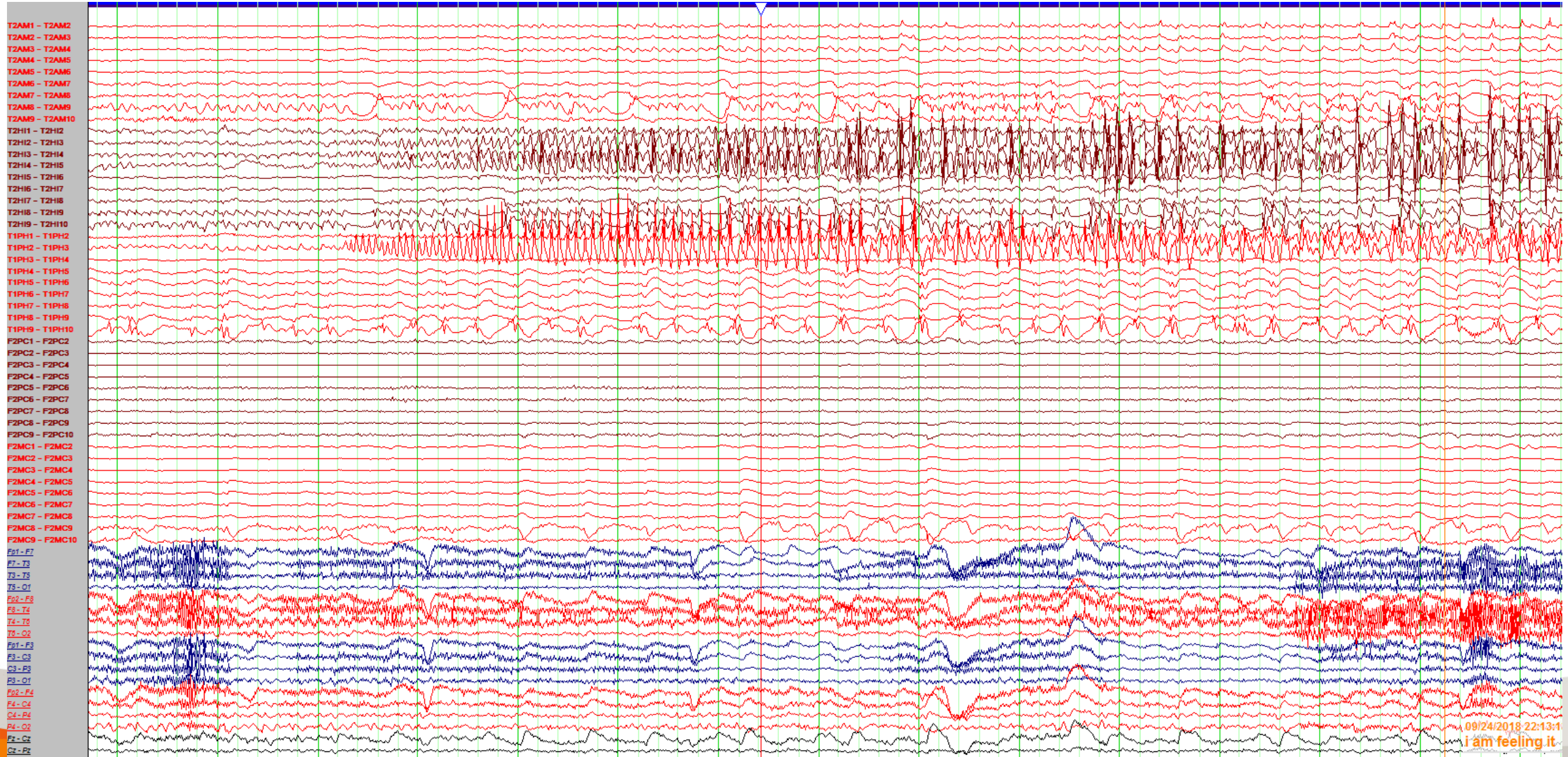
Epilepsy Surgery



StereoEEG

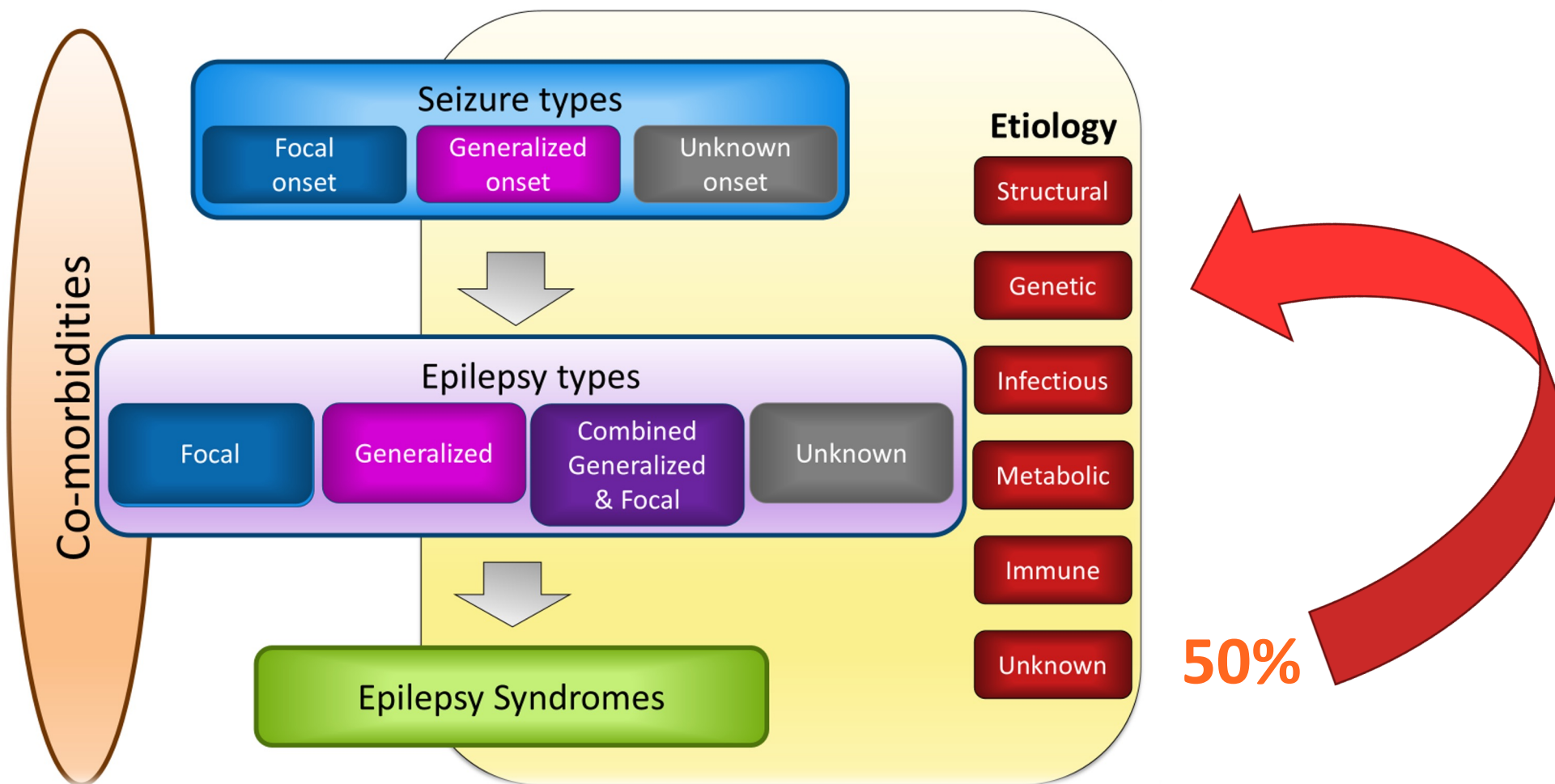


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09/24/2018 22:13:1
i am feeling it.

Causes



Epilepsy Genetics Revolution



- Gene discoveries
- Diagnosis
- End of diagnostic odyssey
- Focus on pathophysiology
- Genetic counselling
- Target therapies (precision medicine)

Prof. Sam Berkovic & Prof. Ingrid Scheffer
University of Melbourne

De-novo mutations of the sodium channel gene SCN1A in alleged vaccine encephalopathy: a retrospective study

Samuel F Berkovic ¹, Louise Harkin, Jacinta M McMahon, James T Pelekanos, Sameer M Zuberi, Elaine C Wirrell, Deepak S Gill, Xenia Iona, John C Mulley, Ingrid E Scheffer

- 14 patients with alleged vaccine encephalopathy in whom the first seizure occurred within 72 h of vaccination
- 11 had *SCN1A* mutations
- Trigger for the encephalopathy, likely via fever or an immune mechanism

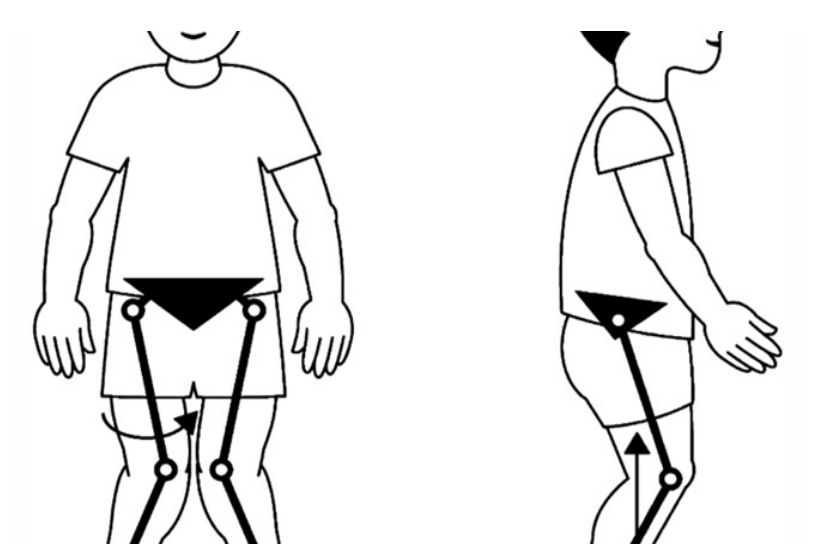
SCN1A spectrum

- From simple febrile seizures and GEFS+ at the mild end to **Dravet syndrome** and intractable childhood epilepsy GTC
- Autosomal dominant
- Myoclonic seizures, psychomotor regression
- Antiseizure agents to avoid: sodium channel blockers
- Status epilepticus
- SUDEP
- Precision medicine – CBD, Fenfluramine, and Stiripentol
- ASO - intratechal

Beyond seizures



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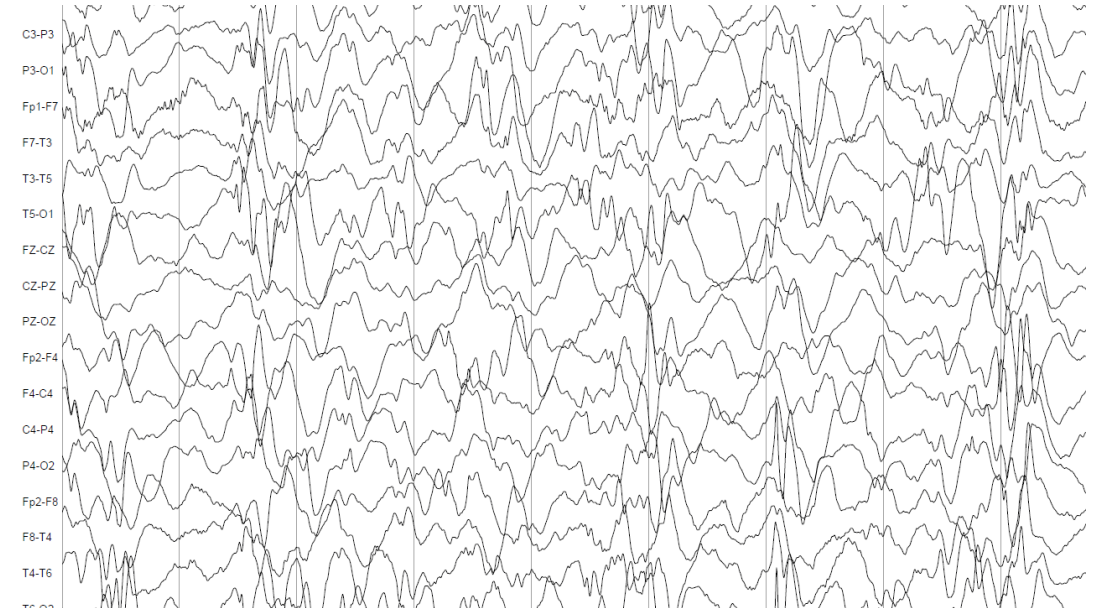


Genton P et al. *Epilepsia* 2011
Rodda JM et al. *Arch Neurol* 2012
Fasano A et al. *Neurology* 2014

Early recognition matters!



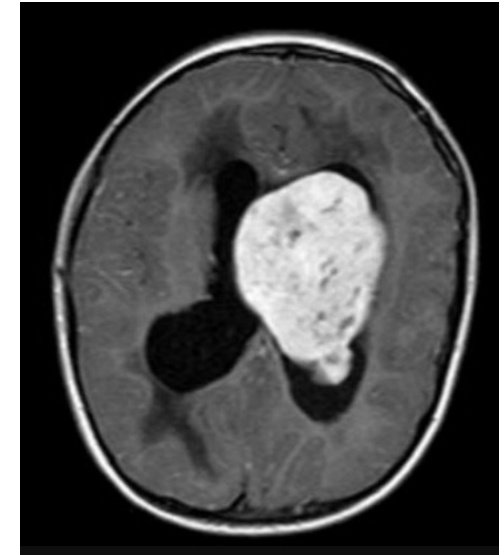
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Treatment options:

- ACTH
- Prednisolone
- Vigabatrin

Early recognition matters!



- Infantile spasms, cardiac rhabdomyoma, facial angiofibromas, kidney angio myolipomas (AMLs), retinal hamartomas, pulmonary lymphangioleiomyomatosis (LAM)
- Vigabatrin
- Epilepsy surgery
- Everolimus

TRANSITION TO ADULT CARE IN ADOLESCENTS WITH EPILEPSY



Transition of Care in Epilepsy

- ↑ Life expectancy of children with serious conditions
- Chronic neurological diseases beginning in childhood and persisting into adulthood
- Youth with chronic disease are at risk of adverse adult medical and social outcomes
 - Congenital heart disease, cancer, renal failure, cystic fibrosis, inflammatory bowel disease, etc..



Transition of Care in Epilepsy

**Epilepsy remission
did NOT protect against poor
social outcome**

The neurologist's role in supporting transition to adult health care

A consensus statement

Lawrence W. Brown, MD
Peter Camfield, MD,
FRCPC
Melissa Capers, MA
Greg Cascino, MD
Mary Ciccarelli, MD
Claudio M. de Gusmao,
MD
Stephen M. Downs, MD
Annette Majnemer, PhD,
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Amy Brin Miller, MSN
Christina SanInocencio,
MS
Rebecca Schultz, PhD
Anne Tilton, MD
Annick Winokur, BS
Mary Zupanc, MD

ABSTRACT

The child neurologist has a critical role in supporting the pediatric to adult health care transition, appropriately planning a youth's educational, vocational, and community needs. Youth with neurologic conditions who experience delayed entry into the adult care system require additional support during this transition. Eight common principles for the transition process have been outlined by a task force. Recommendations are introduced and described in the current paucity of evidence for consideration. *Neurology*® 2016;87

The child neurologist has a critical role in supporting youth with neurologic conditions. Recommendations for leadership and planning of a youth

CRITICAL REVIEW AND INVITED COMMENTARY



Epilepsy: Transition from pediatric to adult care. Recommendations of the Ontario epilepsy implementation task force

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Transition of Care in Epilepsy

Age and Time ↓	Provider	Parent/Family → Youth	
	Major responsibility	Provides care	Receives care
	Support to Parent/family & child/youth	Manages	Participates
	Consultant	Supervisor	Manager
	Resource	Consultant	Supervisor/CEO



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Thank you for listening