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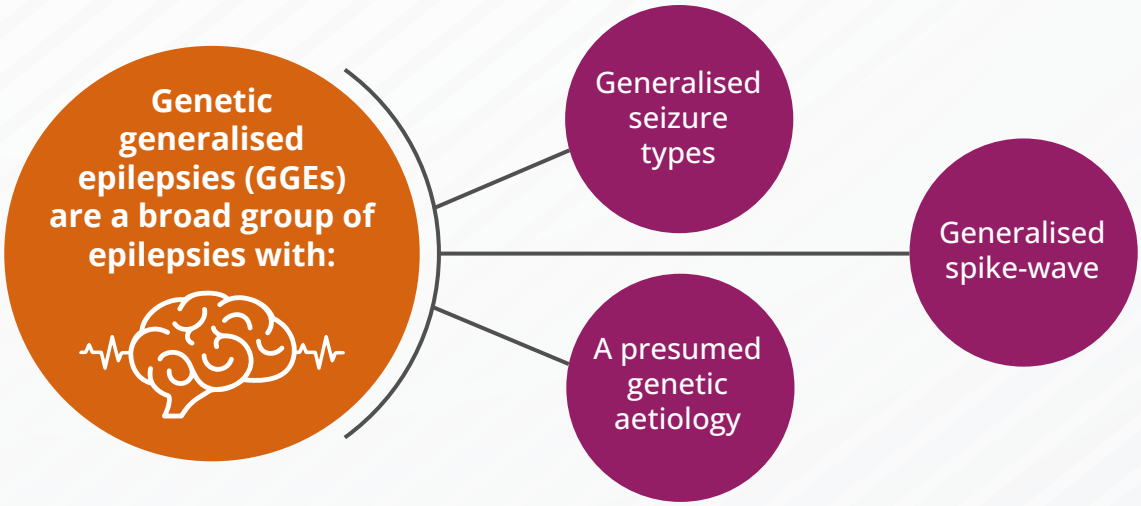
Idiopathic Generalised Epilepsy Syndromes

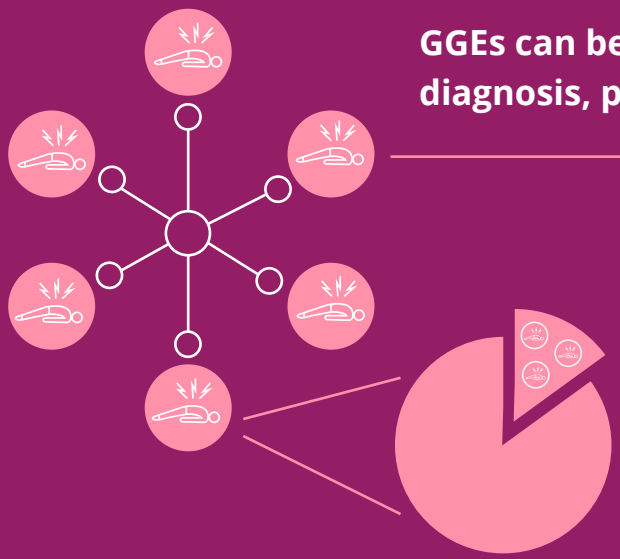
An updated classification and diagnostic criteria

Epilepsy is a neurological disorder associated with abnormal electrical activity in the brain and marked by sudden recurrent episodes of sensory disturbance, loss of consciousness, or convulsions



Based on advancements in:

- Genetics
- Imaging techniques
- EEG studies
- Existing classification of seizures and epilepsies



GGEs can be broken down into smaller subgroups for precise diagnosis, prognosis, and therapeutic decision-making

Of these, the distinct subgroup idiopathic generalised epilepsy (IGE) describes epilepsy syndromes which:

- Are seen in almost 15%–20% of people with epilepsy
- Overlap with some other non-IGE GGE syndromes, and with each other, clinically
- May evolve with age to another IGE, but not to an encephalopathy

To improve diagnosis, the ILAE's Nosology and Definitions Taskforce updated their classification for IGEs

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Idiopathic generalised epilepsies include:

- Childhood absence epilepsy (CAE)
- Juvenile absence epilepsy (JAE)
- Juvenile myoclonic epilepsy (JME)
- Generalised tonic-clonic seizures alone (GTCA)

The four syndromes:

- Differ in their age of onset, predominant seizure type, seizure-specific medication, and remission
- Are all mostly responsive to drug therapy, but certain anti-seizure medications may exacerbate seizures in patients with IGEs

General characteristics of IGEs

Seizures:

One or more of the following generalised seizure types:

- Absence
- Myoclonic (may be focal or asymmetric)
- Tonic-clonic, which may be accompanied by focal or asymmetric features like
  - Eye deviation or version
  - Head deviation or version

Focal findings often shift sides from seizure to seizure

- Myoclonic–tonic–clonic
- Photosensitivity in some patients

EEG:

- Classically, generalised spike-wave discharges, typically 2.5–5.5 Hz are seen
- These mostly appear fragmented during sleep
- Can have focal features
- A photoparoxysmal response in cases with photosensitivity
- Normal background
- A sleep-deprived or prolonged EEG recording may display generalised spike-wave abnormalities in otherwise normal EEGs

Comorbidities:

- Have been associated with:
  - Mood disorders
  - Anxiety
  - ADHD and learning disorders

But further research in this field is required

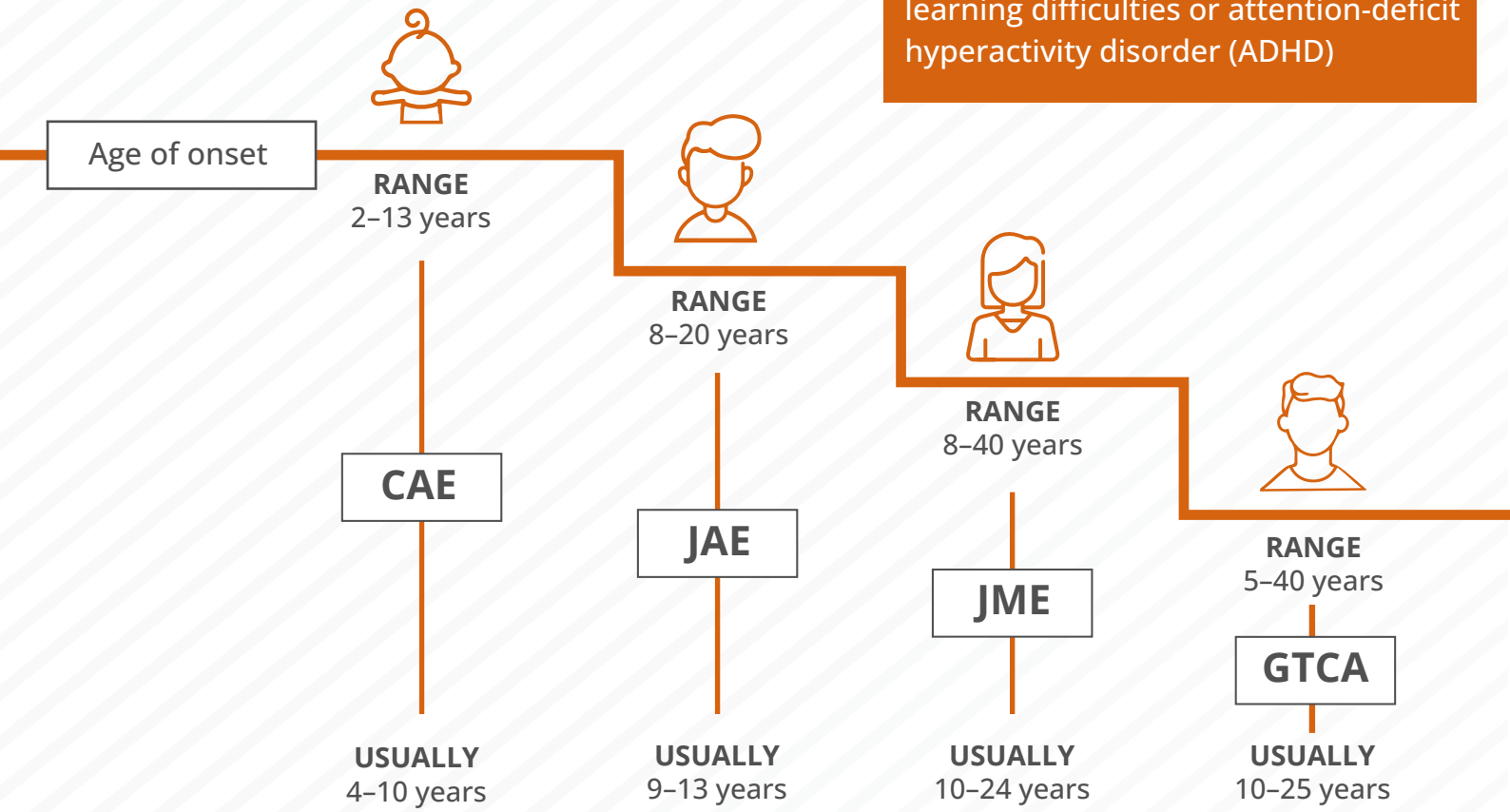
- Not associated with intellectual disability or developmental and epileptic encephalopathies
- Correlated with poorer long-term social outcomes like:
  - Lesser academic achievements
  - Increased risk of unplanned pregnancy
  - Psychiatric, emotional, and behavioural problems
  - Decreased social interaction

Genetics:

- IGEs follow a complex polygenic inheritance
- Environmental factors may play a role in their development
- Links between IGEs and certain pathogenic genetic variants have been reported. These include:
  - GABA receptor subunit genes *(GABRG2, GABRA1)*
  - Glucose transporter-1 encoding gene *(SLC2A1)*
- These pathogenic variants could arise due to inherited or newly acquired mutations

Clinical features of IGEs

Development:
Typically normal, but may have learning difficulties or attention-deficit hyperactivity disorder (ADHD)



FEATURES:				
Seizure presentation	• Daily absence seizures of 3–20 seconds duration (median: 10s) with complete loss of awareness • Staring and loss of facial expression, mostly with oral or manual automatisms • Immediate return to normal activity, sometimes with momentary disorientation • History of febrile or generalised tonic-clonic seizures may be present	• Less frequent absence seizures of 5–30 seconds duration with less complete impairment of awareness • History of febrile or generalised tonic-clonic seizures may be present	• Myoclonic seizures, seen predominantly on awakening • Generalised tonic-clonic seizures seen in >90%, often preceded by myoclonic jerks • Febrile and infrequent absence seizures in 15% and 33% of cases, respectively • Seizures triggered by sleep deprivation and stimulation with light	• Generalised tonic-clonic seizures typically within 2 hours of awakening • Febrile seizures seen in 15% of cases • Seizures triggered by sleep deprivation

Diagnostic features of IGE syndromes

	CAE	JAE	JME	GTCA
EEG	Paroxysms of 2.5–4 Hz generalised spike wave	Paroxysms of 3–5.5 Hz generalised spike wave	>3–5.5 Hz generalised spike-wave or polyspike wave	
EEG background	ORIDA (occipital intermittent rhythmic delta activity) in 21%-30%	Normal		
Epileptiform abnormalities	**While awake:** Regular 3 Hz generalised spike-wave seen at the start of the seizure, however: - 21% of patients may have absences starting at 2.5 Hz spike wave - 43% may have absences starting at 4 Hz	3–5.5 Hz generalised spike-wave	Generalised 3–5.5 Hz spike-wave and polyspike–and–wave that may fragment in sleep	
	While asleep: Polyspike–and–wave may be seen in drowsiness and sleep only	Polyspike–and–wave may be seen in drowsiness and sleep only		
Irregular generalised spike-wave	Uncommon	More common than CAE Abnormalities are more frequent than in CAE	Irregular; seen in all states	Seen only in sleep
Photoparoxysmal response Rare intermittent photic stimulation (IPS) triggers generalised spike-wave without inducing seizures	In 15%	In 25%	Seen in 30%-90% and may trigger myoclonic jerks or generalised myolonic-tonic-clonic seizures	May be seen

Reference:
Hirsch, E., French, J., Scheffer, I.E., Bogacz A., Alsaadi, T., Sperling, M., … & Wirrell, E. (2022). ILAE definition of the idiopathic generalized epilepsy syndromes: position statement by the ILAE Task Force on Nosology and Definitions. *Epilepsia,* 1–25, https://doi.org/10.1111/epi.17236.

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