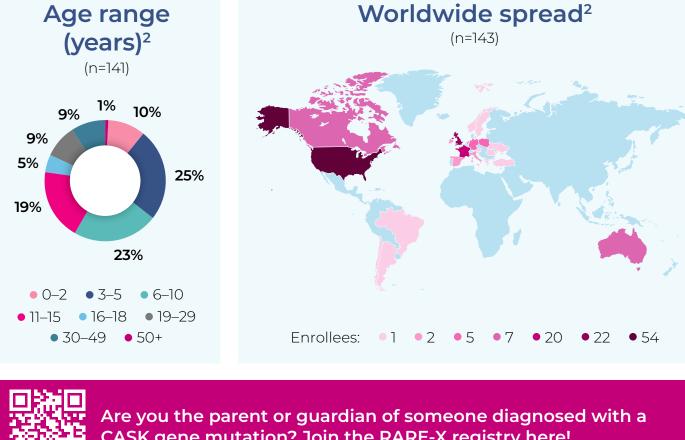
CASK: What Do We Know From Our Registry?

Registry overview





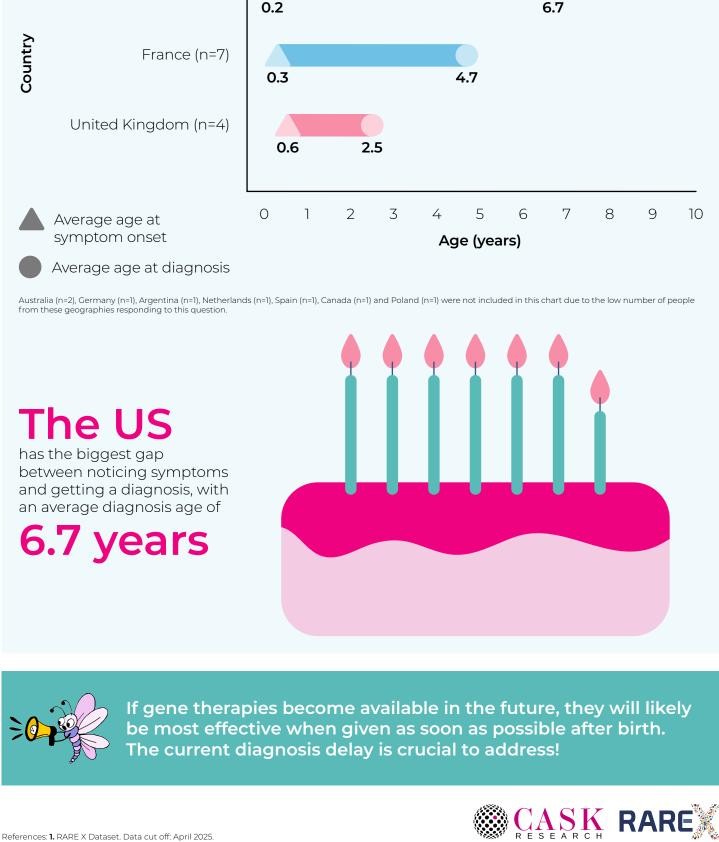




Diagnosis delay



United States (n=11)





Unsure

Number of people

References: 1. RARE X Dataset. Data cut off: April 2025.

reporting symptoms:

Unusual movements

The sample size for each survey used to determine symptom prevalence varied. Excludes 'abnormal muscle function'.

Symptoms

References: 1. RARE X Dataset. Data cut off: April 2025

Cognitive impairment Seizures **Coordination issues** Hypertonia Abnormal eye movement Constipation **Esophagus issues** Visual impairment Oral dysphagia Abnormal facial muscles

> Brain and nervous system
> Muscles
> Digestive
> Eyes and vision GrowthHead, face, and neckBehaviour survey

Hypotonia

Abnormal

electroencephalogram

Symptoms related to the brain and nervous system are the most common¹ Key symptoms occur relatively early in childhood¹ Cognitive impairment Seizures Abnormal EEG (n=51)(n=20)(n=19)0–3 months 4–7 months 8–11 months 1–2 years 3–5 years 6-10 years 11–15 years 16-18 years 19-29 years

6-8

9–10

Starts in the left and

right sides of the brain³

11–13

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14–16

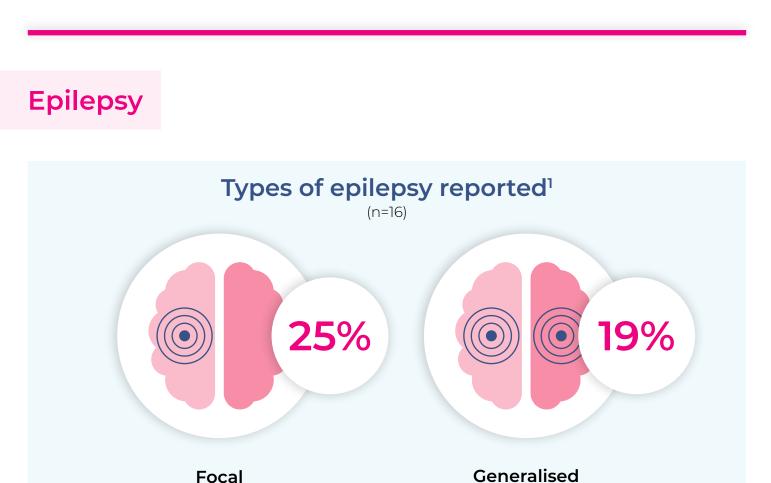
3–5

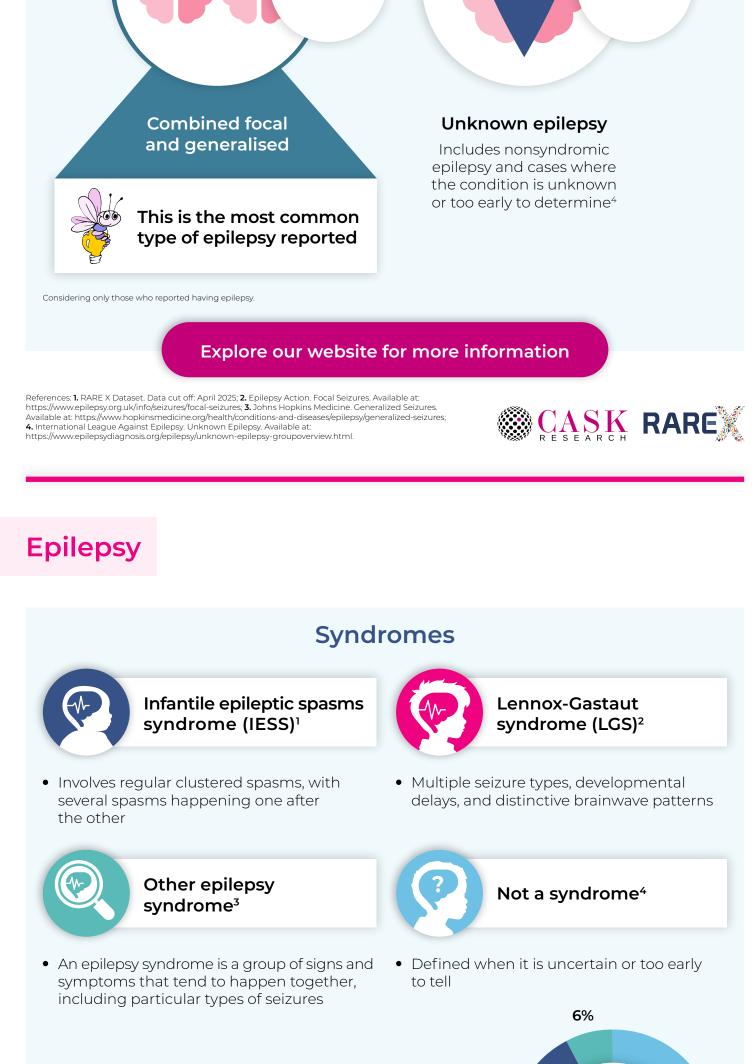
- Represents the most frequent age range at which each symptom appeared.

0–2

Starts in one part

of the brain²





24%

12%

52% of patients have treatment-resistant epilepsy (n=21)⁵

Epilepsy status

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59%

References: 1. Epilepsy Action. Infantile Epileptic Spasm Syndrome (West Syndrome). Available at: https://www.epilepsy.org.uk/info/syndromes/west-syndrome-infantile-spasms; 2. National Institute of Neurological Disorders and Stroke. Lennox-Gastaut Syndrome. Available at: https://www.ninds.nih.gov/health-infor-

6. RARE X Dataset, Data cut off; April 2025

Generalised

tonic-clonic seizures

Time since last seizure

Today

1-6 days ago

1-4 weeks ago

5-12 weeks ago

13-26 weeks ago

6-12 months ago

13-24 months ago

Considering only those who reported having seizures.

>2 years ago

IESS

Other epilepsy syndrome

Considering only those who reported having epilepsy.

Treatment resistance

Treatment-resistant

epilepsy is the resistance people can develop to medications that prevent seizures⁶



behaviour, movements and consciousness¹

mation/disorders/lennox-gastaut-syndrome; **3.** Epilepsy Foundation. What Is an Epilepsy Syndrome? Available at: https://www.epilepsy.com/what-is-epilepsy/syndromes; **4.** American Society of Gene + Cell therapy. Sail For Epilepsy, Part 2: An Overview of Syndromic and Monogenic Non-syndromic Epilepsies.

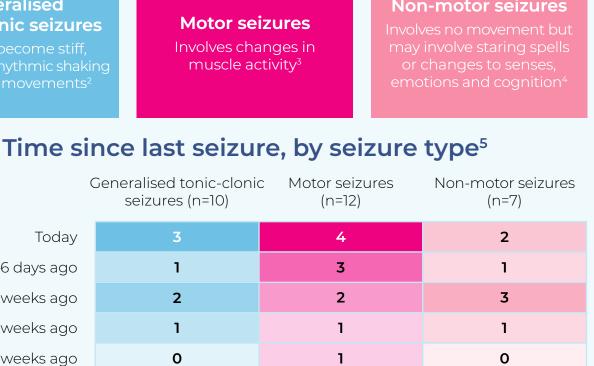
Available at: https://www.asgct.org/publications/news/may-2022/sail-for-epilepsy-part-2; 5. Epilepsy Foundation.

Drug Resistant Epilepsy. Available at: https://www.epilepsy.com/treatment/medicines/drug-resistant-epilepsy;

59% of individuals reported that their child's

epilepsy does not constitute a specific syndrome (n=17)⁵

Not a syndrome



0

0

0

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0

0

1

Sleep

0 of children fall asleep within 20 minutes of going to bed $(n=64)^1$

seizures (n=10)

1

2

1

0

0

2

relatively regular pattern⁵

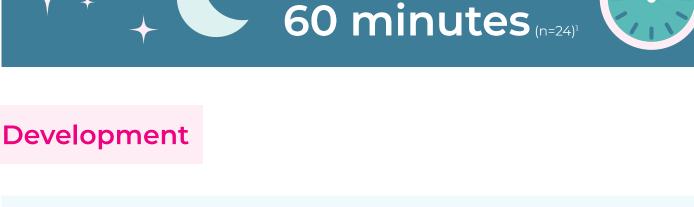
References: 1. Citizens Specialty Hospital. Difference between convulsions, seizures and epilepsy. Available at: https://www.citizenshospitals.com/blogs/difference-between-convulsions-seizures-and-epilepsy;

2. Epilepsy Society. Epileptic Seizures. Available at: https://epilepsysociety.org.uk/about-epilepsy/epileptic-seizures;

3. International League Against Epilepsy. Motor Seizure. Available at: https://www.epilepsydiagnosis.org/seizure/motor-overview.html; 4. Epilepsy Foundation. Types of Seizures. Available at: https://www.epilepsy.com/what-is-epilepsy/seizure-types; 5. RARE X Dataset. Data cut off: April 2025.

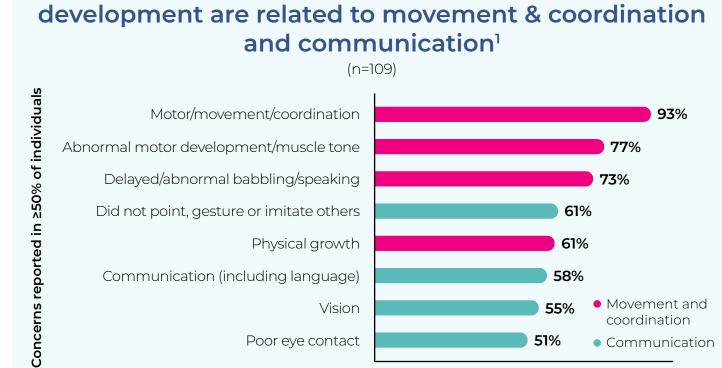
Among those who experience seizures, these events

generally occur on at least a monthly basis, suggesting a



The most commonly reported number of minutes awake during the night was

Most of the concerns expressed regarding



Physical growth

Poor eye contact

by signing up today

Vision

0

Communication (including language)

Abnormal motor development/muscle tone refers to participants who are unusually stiff or floppy

20

61%

Movement and

coordination

80

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Communication

100

58%

55%

51%

60

40