

CASK: What Do We Know From Our Registry?

Registry overview



An increase in registry sign-ups has improved our understanding of the characteristics of people with CASK-related disorders

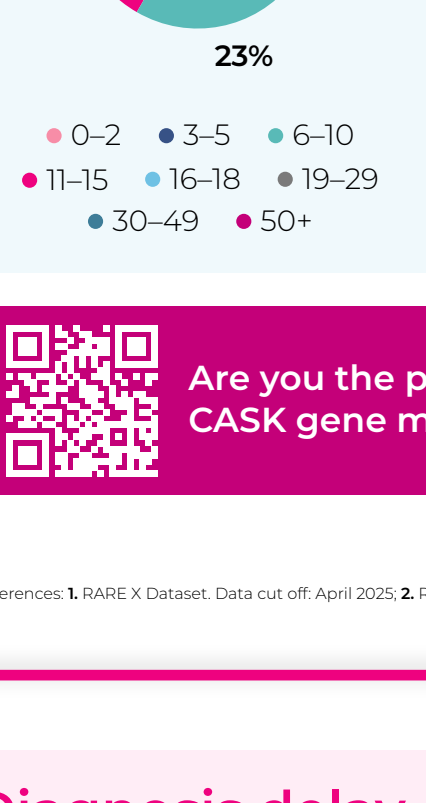
Increase in enrolment¹



Gender²



Age range (years)²

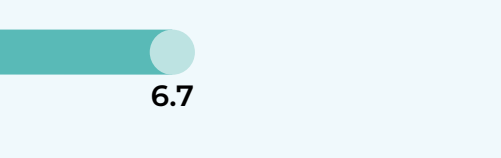


Worldwide spread²



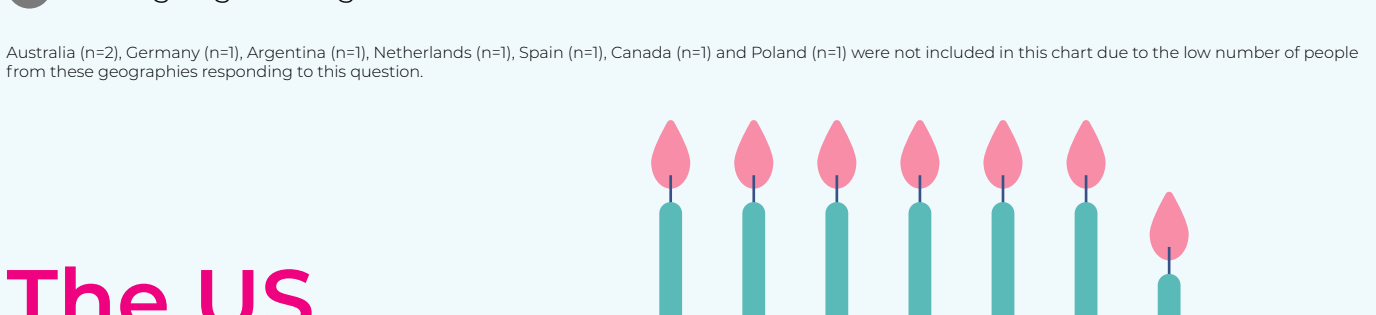
Are you the parent or guardian of someone diagnosed with a CASK gene mutation? Join the RARE-X registry here!

References: ¹ RARE X Dataset. Data cut off: April 2025. ² RARE X Dataset. Data cut off: June 2025.



Diagnosis delay

How long does it take to get a diagnosis?¹



The US

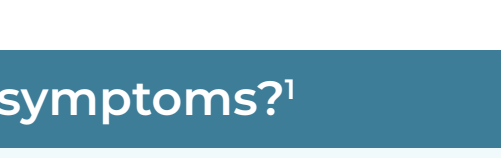
has the biggest gap between noticing symptoms and getting a diagnosis, with an average diagnosis age of

6.7 years



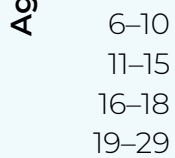
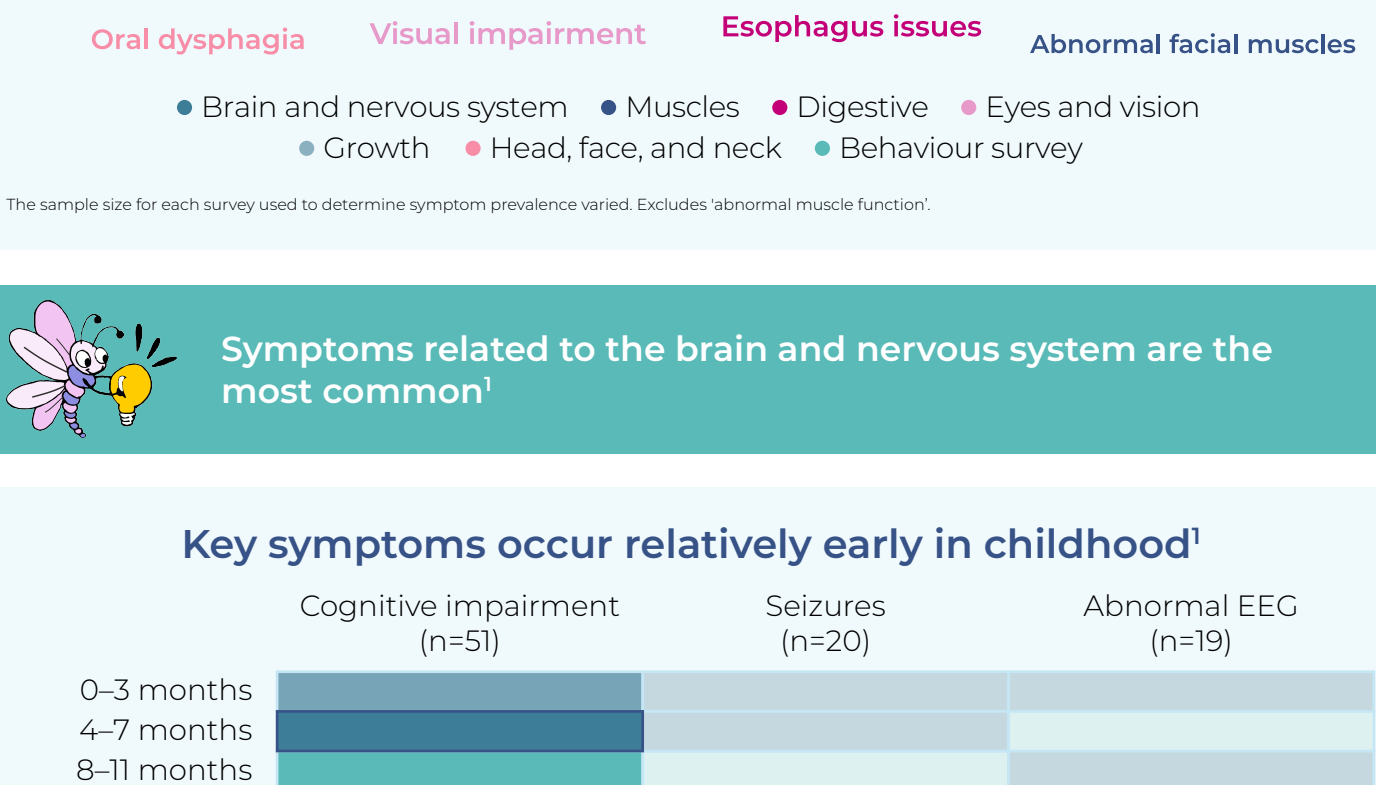
If gene therapies become available in the future, they will likely be most effective when given as soon as possible after birth. The current diagnosis delay is crucial to address!

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



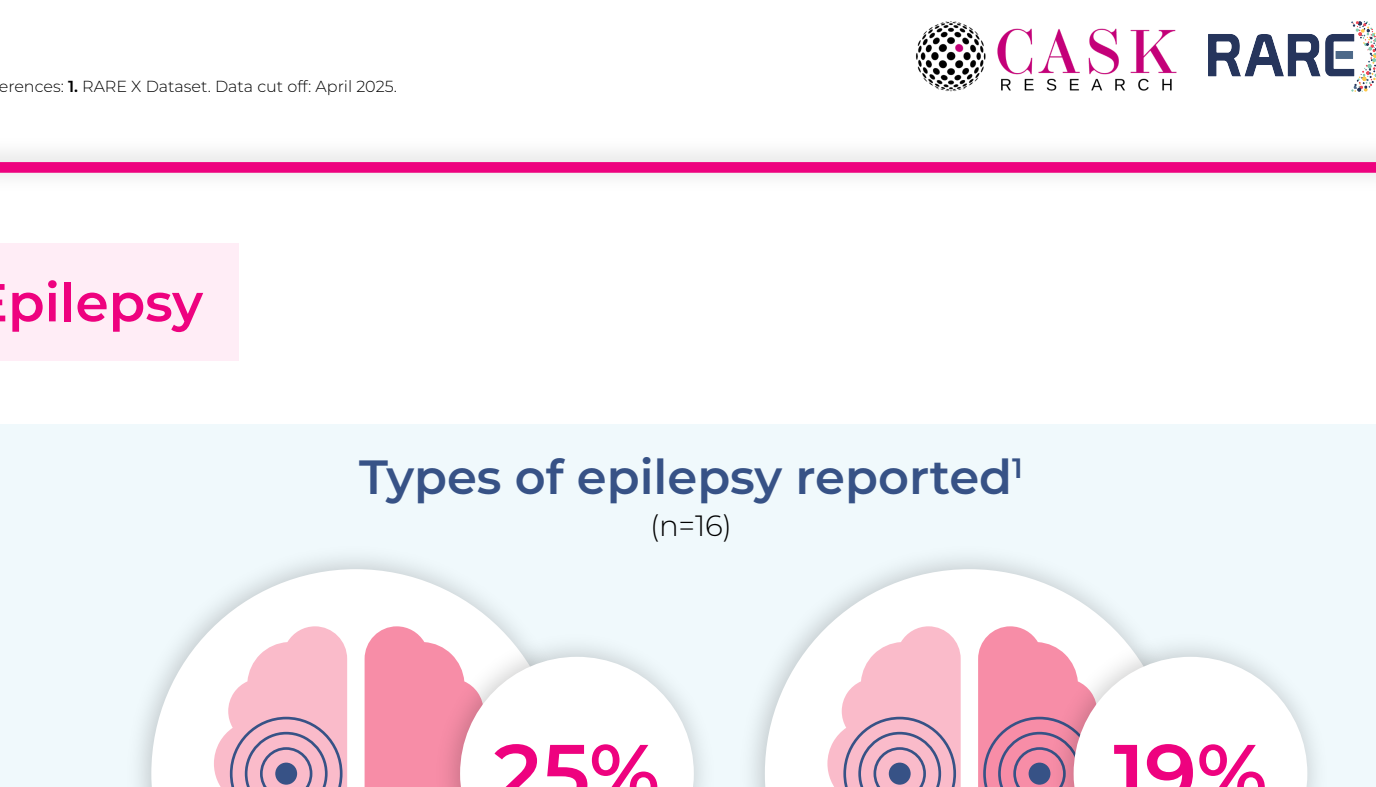
Symptoms

What are the most common symptoms?¹



Symptoms related to the brain and nervous system are the most common¹

Key symptoms occur relatively early in childhood¹

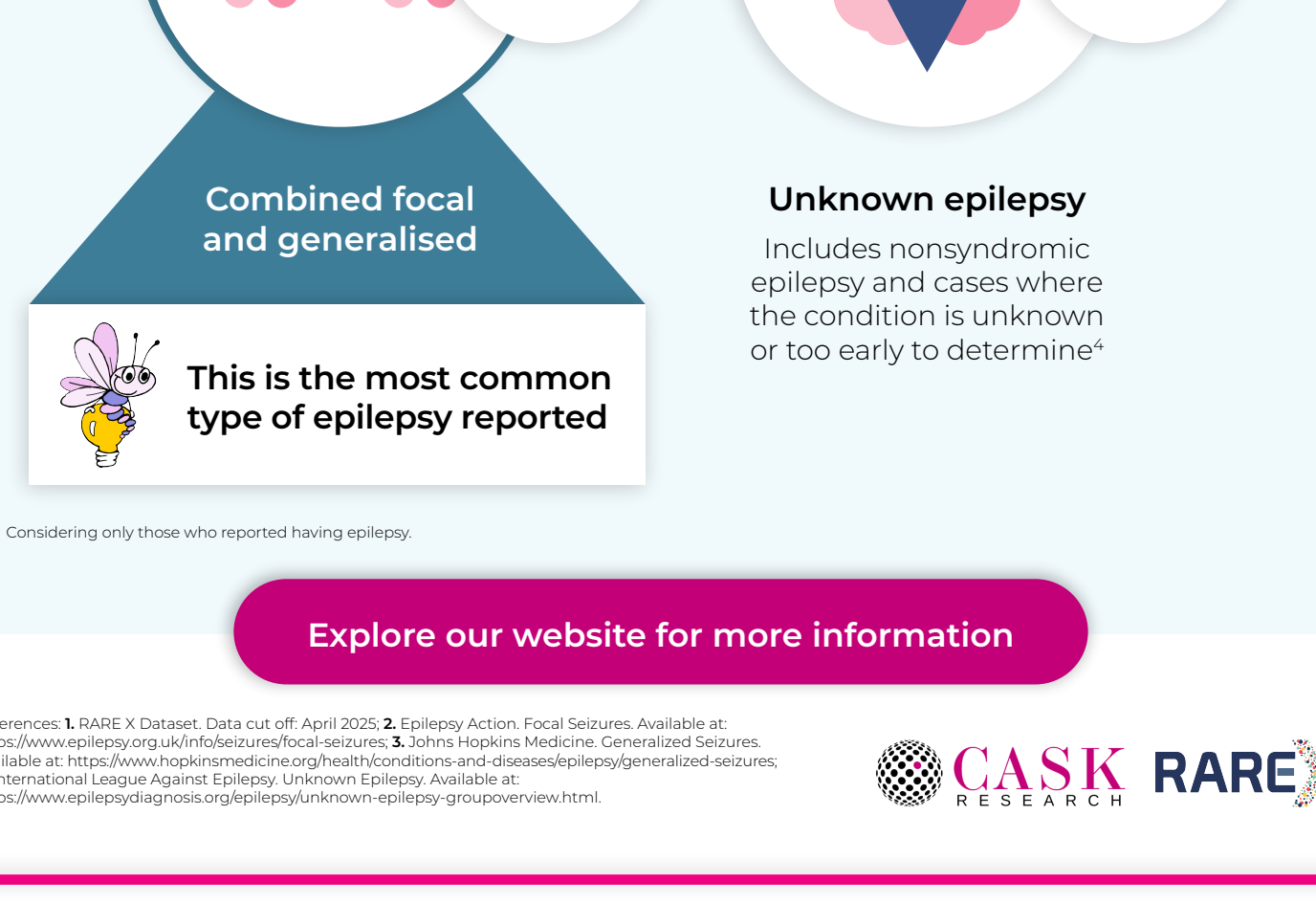


References: ¹ RARE X Dataset. Data cut off: April 2025.



Epilepsy

Types of epilepsy reported¹



Considering only those who reported having epilepsy.

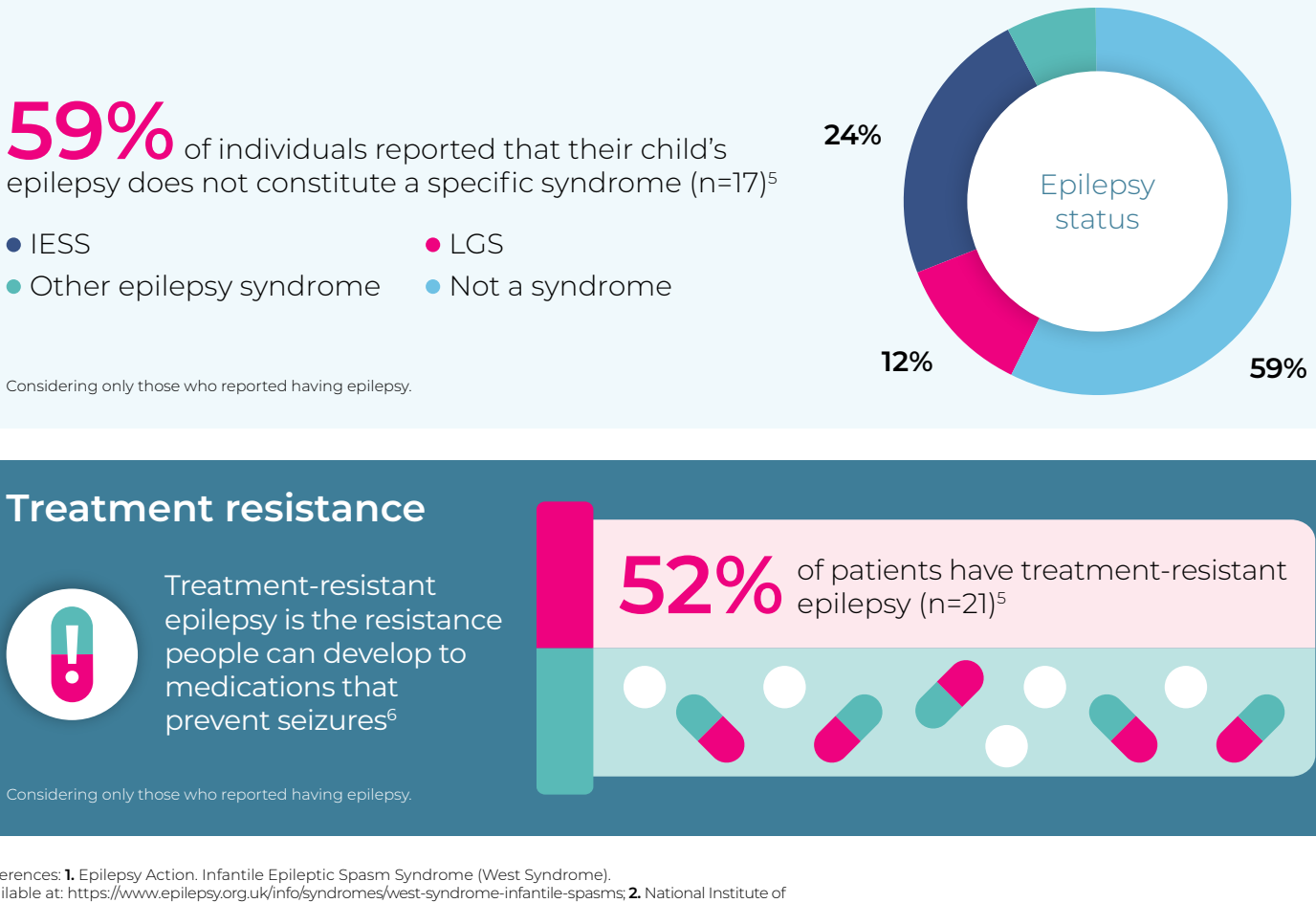
Explore our website for more information

References: ¹ RARE X Dataset. Data cut off: April 2025. ² Epilepsy Action. Focal Seizures. Available at: <https://www.epilepsy.org.uk/info/seizures/focal-seizures>. ³ Johns Hopkins Medicine. Generalized Seizures. Available at: <https://www.hopkinsmedicine.org/health/conditions-and-diseases/epilepsy/generalized-seizures>. ⁴ American Society of Gene + Cell therapy. Sali For Epilepsy. Part 2: An Overview of Syndromic and Monogenic Non-syndromic Epilepsies. Available at: <https://www.asgct.org/publications/news/may-2023/sali-for-epilepsy-part-2-5>. ⁵ Epilepsy Foundation. Drug Resistant Epilepsy. Available at: <https://www.epilepsy.com/treatment/medicines/drug-resistant-epilepsy>. ⁶ RARE X Dataset. Data cut off: April 2025.



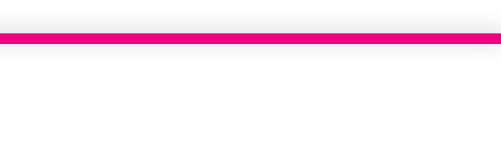
Epilepsy

Syndromes



Considering only those who reported having epilepsy.

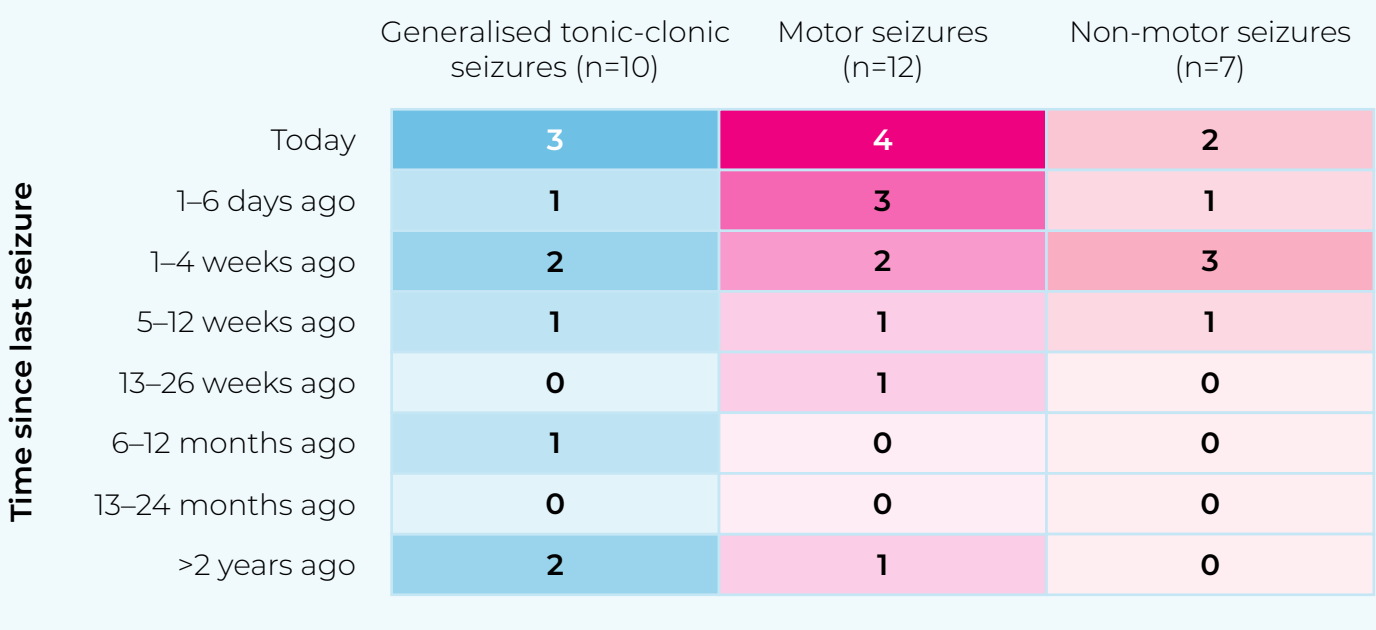
References: ¹ Epilepsy Action. Infantile Epileptic Spasm Syndrome (West Syndrome). Available at: <https://www.citizenshospitals.com/blog/difference-between-convulsions-seizures-and-epilepsy>. ² National Institute of Neurological Disorders and Stroke. Lennox-Gastaut Syndrome. Available at: <https://www.ninds.nih.gov/health-information/disorders/lennox-gastaut-syndrome>. 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. Sali For Epilepsy. Part 2: An Overview of Syndromic and Monogenic Non-syndromic Epilepsies. Available at: <https://www.asgct.org/publications/news/may-2023/sali-for-epilepsy-part-2-5>. ⁵ Epilepsy Foundation. Drug Resistant Epilepsy. Available at: <https://www.epilepsy.com/treatment/medicines/drug-resistant-epilepsy>. ⁶ RARE X Dataset. Data cut off: April 2025.



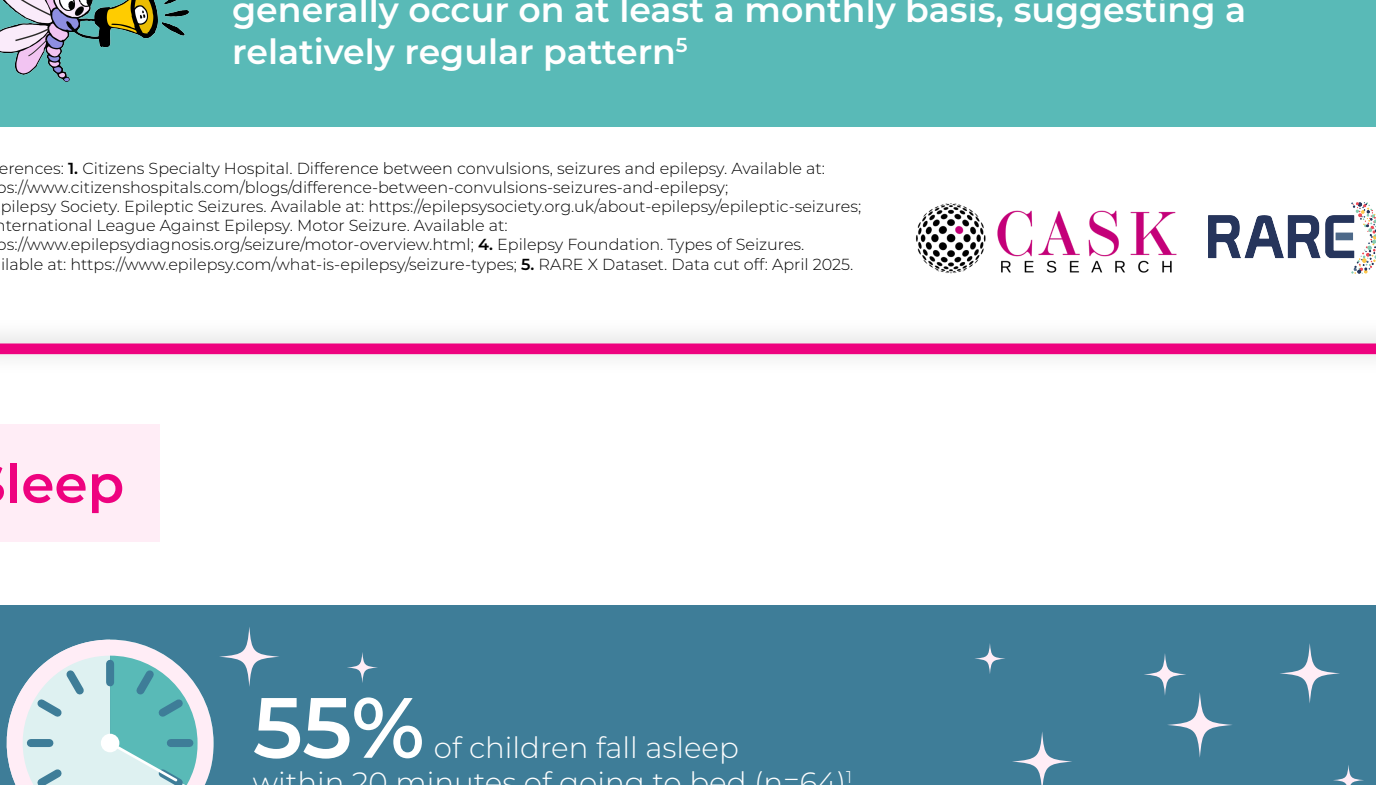
Seizures

Epilepsy involves a pattern of repeated seizures that are not caused by identifiable factors like injury or infection¹

A seizure is a sudden surge of electrical activity in the brain that can affect behaviour, movements and consciousness¹



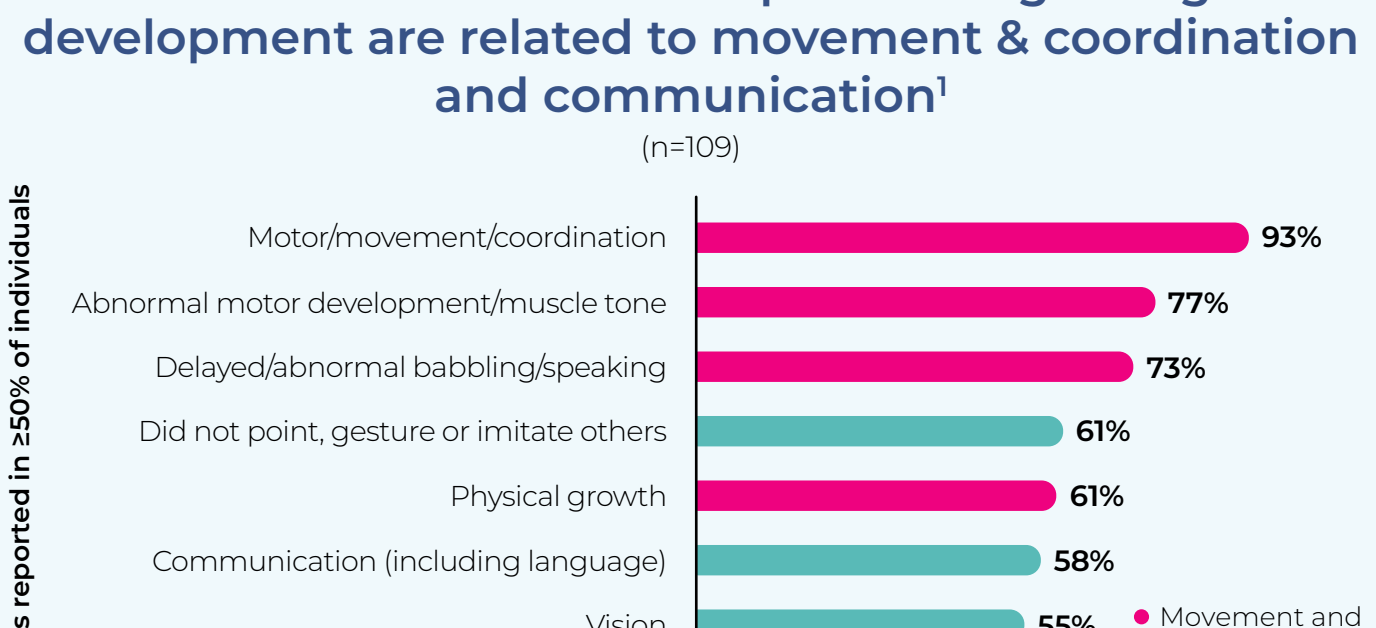
Time since last seizure, by seizure type⁵



Considering only those who reported having seizures.

Among those who experience seizures, these events generally occur on at least a monthly basis, suggesting a relatively regular pattern⁶

Sleep



Development

Most of the concerns expressed regarding development are related to movement & coordination and communication¹



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

Help us transform outcomes by signing up today

