

SCN2A-RELATED EPILEPSY

ASSOCIATED EPILEPSY SYNDROMES

- Self Limited Infantile Epilepsies (SeLIE)
- Early Infantile Epileptic Encephalopathy (type 11)
- Ohtahara Syndrome
- Infantile Epileptic Spasms Syndrome
- Lennox-Gastaut Syndrome (LGS)
- Generalized Epilepsy with Febrile Seizures
- Epilepsy of Infancy with Migrating Focal Seizures
- Later-onset epilepsy with ASD

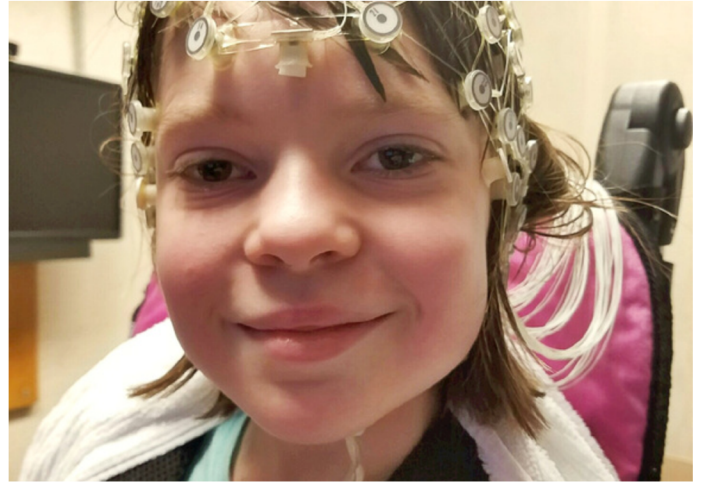
Seizures in SCN2A-related disorders vary and may change over time. Sudden unexpected death in epilepsy (SUDEP) is the sudden, unexplained death of someone with epilepsy. While risk is not elevated for all individuals with SCN2A, it is highest in those with early-onset, severe, or refractory epilepsy, particularly when convulsive seizures are present.



How Rare are SRDs?

SRDs affect an estimated 11 out of every 100,000 births, resulting in more than 400 children born each year in the United States. While loss-of-function (LoF) variants are predicted to outnumber gain-of-function (GoF) variants by approximately 7 to 1, GoF variants are often associated with earlier onset and more severe clinical presentations.

Source: George, Jr, A. L., Abbott, M., Bender, K. J., Brunklaus, A., Demarest, S., Egan, S., Haviland, I., Kearney, J. A., Myers, L. S., Olson, H. E., Sanders, S. J., SanInocencio, C., Symonds, J., & Thompson, C. H. (2024). SCN2A-Related Disorders. Elements in Genetics in Epilepsy. <https://doi.org/10.1017/9781009530361>



GAIN OF FUNCTION IN SCN2A & SODIUM CHANNEL BLOCKERS (SCB)

- There is a correlation between age at disease onset, response to SCBs and the functional properties of variants in children with SCN2A-related epilepsy.
- Variants associated with early infantile epilepsy tend to result in increased sodium channel activity with gain-of-function.
- SCBs were often associated with clinically relevant seizure reduction or seizure freedom in children with early infantile epilepsies (<3 months), whereas other anti-epileptic drugs were less effective.
- Clinical Trials are available for this subtype. Please visit [SCN2A.ORG](https://www.scn2a.org) for the latest therapeutic pipeline.

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EXAMPLES OF SODIUM CHANNEL BLOCKERS

phenytoin, carbamazepine, oxcarbamazepine, lacosamide, lamotrigine, zonisamide

**names may vary depending on country*