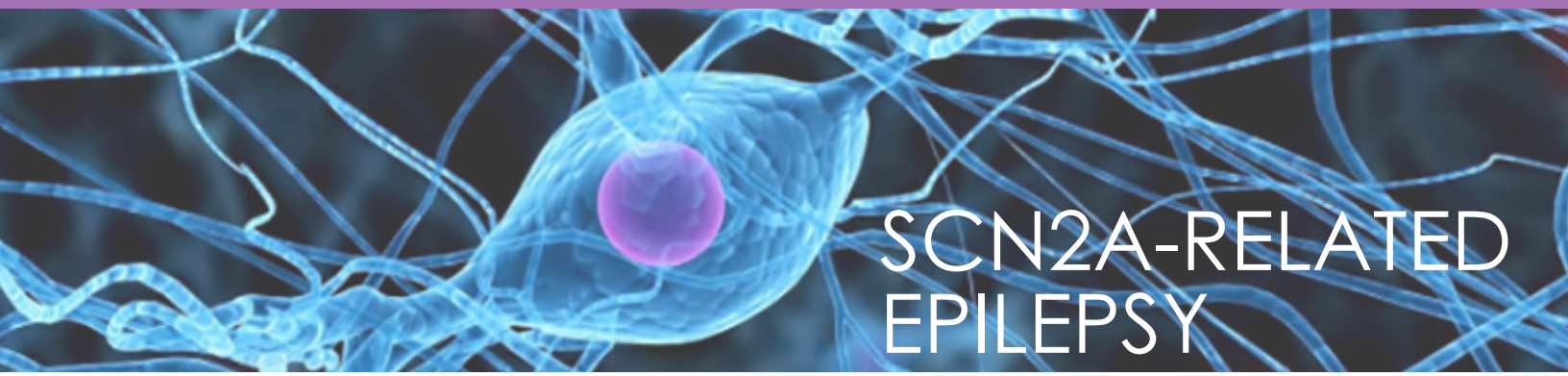
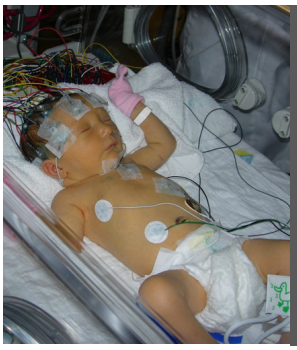


SCN2A-RELATED EPILEPSY

QUICK FACT SHEET

SCN2A ASSOCIATED EPILEPSY SYNDROMES

- Benign Familial Infantile Seizures (type 3)
- Early Infantile Epileptic Encephalopathy (type 11)
- Ohtahara Syndrome
- West Syndrome
- Lennox-Gastaut
- Generalized Epilepsy with Febrile Seizures
- Migrating Partial Epilepsy of Infancy
- Infantile Spasms
- Later-onset epilepsy with ASD



Seizure types vary from child to child and can change throughout different phases of growth and development. As in other severe epilepsies, there is an increased risk of sudden unexpected death in epilepsy, known as SUDEP.

HOW RARE IS SCN2A?

It is estimated that there will be approximately 11 SCN2A-related cases per 100,000 births.

Over 400 SCN2A-mediated disorders children will be born each year in the United States alone.

Source: Sanders, et al. (2018). Progress in understanding and treating SCN2A-mediated disorders. Trends in Neuroscience, 41(7):442-456.

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

- Correlation between age at disease onset, response to SCBs and the functional properties of mutations in children with SCN2A-related epilepsy.
- Mutations 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.
- SCBs were rarely effective in epilepsies with later onset (>3 months) and sometimes induced seizure worsening.

Source: Wolff, et al. (2017). Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 140(5):1316-1336.

Examples of Sodium Channel Blockers

phenytoin, carbamazepine, oxcarbamazepine, lacosamide, lamotrigine, zonisamide

SCN2A-RELATED AUTISM & EPILEPSY
FAMILIESCN2A® FOUNDATION
WWW.SCN2A.ORG