

Treatment Consultations

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Due diligence in research for updates in SCN8A-related clinical information is expected before changing treatment course. Never disregard professional medical advice because of something you have read within this reference guide.

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Discover clinical wisdom about
SCN8A Epilepsy from physicians with
experience treating this rare disorder.

Learn more about the resources available for
your patients with SCN8A Epilepsy.

SCN8A Epilepsy Clinician Information & Reference Guide

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General Information

SCN8A mutation pathophysiology

- Most concentrated in neural tissue but also found in cardiac tissue
- SCN8A mutations lead to altered persistent sodium current (most commonly **UP**-regulation of sodium within the channel) and neuronal excitability.¹

SCN8A mutation onset

- Age: mean 4 months (Range: birth to >10 years, N=127)²
- Seizure type varies
- EEG findings vary from normal or baseline slowing to infantile spasms with or without hypsarrhythmia

Treatment Plan

Drugs of choice

Sodium-channel blockers

- Use early and sometimes at higher-than-standard doses
- Monitor for drug-efficacy AND toxicity
- Treat aggressively to achieve seizure freedom when possible
- Seizure control may be related to prognosis
- Those with the best success rates at high dose:
 - Oxcarbazepine
 - Carbamazepine
 - Lamotrigine
 - Phenytoin
 - Lacosamide

Vitamin D supplementation

- Use for Osteopenia prevention

Drugs to avoid

Levetiracetam (Keppra) has been shown to increase seizure severity or has no effect in up to 90% of SCN8A patients. Failure rate = 82%*² (makes seizures worse, or no effect)
*N=83.

Comorbidities

Consider the following consultations/referrals:

- **Gastroenterology:**
G-tube dependency, Constipation
- **Pulmonary:**
Sleep apnea, Laryngomalacia, Pneumonia
- **Orthopedic:**
Scoliosis
- **Ophthalmology:**
Cortical Visual Impairment
- **Cardiology** (with experience in channelopathies):
Arrhythmias, Bradycardia
- **Physical/Occupational Therapy:**
Hypotonia, Ataxia, Dyskinesias/ Movement disorders
- **Speech and Language Pathology:**
Unsafe swallow, Non-verbal
- **Behavior/Neuropsychology:**
Autism Spectrum Disorder, ADHD

Monitoring

SUDEP

Genetic epilepsies have a higher risk of Sudden Unexpected/Unexplained Death in Epilepsy (SUDEP). SCN8A specifically may increase the risk of SUDEP possibly due to increased susceptibility of arrhythmias.³

- Consider pulse oximetry, video/audio monitoring, movement monitors, and anti-suffocation pillows in these patients due to potential for comorbid bradycardia and/or prolonged apneic events during seizures.
- Counsel parents to use on child during sleep.
- Encourage parents/caregivers to receive CPR certification

NOTE:

The presence of either sleep apnea or a bradycardia diagnosis (or both) will aid in insurance approval for pulse oximetry. Consider sleep study.

Genetic Testing

- Consider testing for genetic epilepsy (namely SCN8A) in any patients who have shown an adverse response to levetiracetam.
- Consider SCN8A testing for biological parents to determine *de novo* status versus possible inheritance patterns.
- Even in unaffected carriers of SCN8A genetic mutations or if a variant is considered non-pathogenic, consider cardiac work-up and regular follow-up.

Genetic Counseling

Most useful in the cases of inherited SCN8A mutations and/or when parents are considering future family planning.

Patients with SCN8A mutations are **NOT to be treated like those with SCN1A mutations or Dravet Syndrome**

References

1. American Epilepsy Society. "Mysteries of SCN8A mutation in epilepsy unraveled." ScienceDaily. ScienceDaily, 6 December 2015. <www.sciencedaily.com/releases/2015/12/151206164755.htm>
2. SCN8A Families Support Group. Survey of SCN8A parents. SCN8A Registry data. <www.scn8a.net>
3. American Epilepsy Society. "Unraveling the Genetic Basis of Sudden Unexpected Death in Epilepsy." https://www.aesnet.org/about_aes/press_releases/unravelinggeneticbasissudep