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A term female infant born after uncomplicated pregnancy presented on day of life 1 with near continuous abnormal movements (Video 1) and episodes of apnea concerning for seizures. Initial EEG captured movements, which were non-epileptic, and interictal EEG was normal. Repeat EEG several days later showed seizures characterized by apnea and bradycardia (Figure). Genetic testing revealed a de novo heterozygous pathogenic variant in SCN8A (c.3979A>G p.Ile1327Val), classically associated with epileptic encephalopathy. At 1 year follow-up, movements had decreased on oxcarbazepine and diazepam, but she had daily seizures and severe developmental delay. This case highlights the phenotypic variability of SCN8A mutations\(^1\), including both neonatal seizures and movement disorders\(^2\).

Video 1 Caption:

Infant displaying abnormal movements including eyelid myoclonus, head tremor, truncal titubation, and near continuous polymyoclonus of the distal extremities. Although not well appreciated in this video,
the patient was also noted to have intermittent saccadic intrusions. Movements were not suppressible, but ceased with sleep.

**Figure Caption:**

EEG showing patient’s seizure characterized electrographically by rhythmic low voltage alpha and beta frequency activity in the central head region with evolution to bilateral 2 Hz diffuse spikes. Clinically, the seizure was associated with apnea and bradycardia. Her interictal EEG was normal.

References


Teaching Video NeuroImage: Neonate With Complex Movement Disorder and Seizures
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