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 nonepileptic, 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 a 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 pathogenic variants in SCN8A, including both neonatal seizures and movement disorders.

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Appendix References


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Appendix (continued)

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