Teaching Video NeuroImage: Hereditary Hyperekplexia Mimicking Tonic Seizures in an Infant

Derek G. Neupert, MD, Kevin M. Rathke, MD, and Mohamad A. Mikati, MD

Abstract

Hereditary hyperekplexia is a rare neurologic disorder characterized by an exaggerated startle response with profound muscle stiffness.1,2 Given the nature of the spells, this condition is often misdiagnosed as epilepsy. Mutations in glycine receptors and transporters are the primary cause of this syndrome.1 We present an example of stimulus-induced hyperekplexia captured on video EEG in a 7-week-old girl with compound heterozygous variants in the presynaptic glycine transporter gene SLC6A5.

Case Summary

A 7-week-old full-term girl with a history of motor delay was referred for presumed daily tonic seizures since day 2 of life. Events were refractory to levetiracetam and phenobarbital. Her family reported that episodes are triggered by tactile stimulation and resolve once the patient is held. We suspected hereditary hyperekplexia. On video EEG, 4 typical events were captured without an epileptiform correlate (Video 1). She was transitioned to clonazepam with remarkable improvement. Genetic testing confirmed the diagnosis (compound heterozygous in the glycine transporter gene SLC6A5). Both variants have been reported previously in patients with hereditary hyperekplexia (pathologic variant: c.679+1G>T, IVS3+1G>T; and likely pathologic variant: c.1640T>C, p.F547S).

Study Funding

The authors report no targeted funding.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.
References

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Neurology 2021;97:e2248-e2249 Published Online before print July 15, 2021
DOI 10.1212/WNL.00000000000012538

This information is current as of July 15, 2021

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