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

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Abstract:

Hereditary hyperekplexia is a rare neurological disorder characterized by an exaggerated startle response with profound muscle stiffness. 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. 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:

7-week-old full term girl with history of motor delay was referred for presumed daily tonic seizures since day 2 of life. Events were refractory to levetiracetam and phenobarbital. Family reported that episodes are triggered by tactile stimulation and resolve once the patient is held. We suspected hereditary hyperekplexia. On video EEG, four typical events were captured without epileptiform correlate (Video). 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).
**Video 1 Legend:**

Video EEG of stimulus induced hyperekplexia event triggered by touching patient’s abdomen. Clinical episode is observed as sustained full body tonic stiffening and bilateral limb extension. Event resolves once baby is held. No epileptiform correlate is observed on video EEG, only muscle artifact.

Video 1-[http://links.lww.com/WNL/B476](http://links.lww.com/WNL/B476)

Teaching Slides-[http://links.lww.com/WNL/B477](http://links.lww.com/WNL/B477)
References:


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