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# Rapid whole genome sequencing identifies a novel GABRA1 variant associated with West syndrome

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## Abstract

A nine month old infant was admitted with infantile spasms which improved on topiramate and steroids. He also had developmental delay, esotropia, hypsarrhythmia on interictal electroencephalogram (EEG), and normal brain magnetic resonance imaging (MRI). West syndrome is the triad of infantile spasms, interictal hypsarrhythmia, and mental retardation. Rapid trio whole genome sequencing (WGS) revealed a novel, likely pathogenic, de novo variant in the gene encoding  $\gamma$ -aminobutyric acid GABA type A receptor,  $\alpha 1$  polypeptide (GABRA1 c.789G>A, p.Met263Ile) in the proband. GABRA1 mutations have been associated early infantile epileptic encephalopathy type 19 (EIEE19). We suggest that GABRA1 p.Met263Ile is associated with a distinct, West syndrome phenotype.

## ACCEPTED MANUSCRIPT

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