Teaching Video NeuroImage: Dystonic Cataplexy in *KCNMA1* Paroxysmal Movement Disorder

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A 2-year-old girl with slight speech delay presented with spells of axial atonia, limb dystonia, and preserved consciousness. Spells could be triggered by strong emotions or without clear cause (Video 1). Attacks began at 8 months but peaked at 18 months, lasting 10–30 seconds and occurring 70 x/day. Neurologic examination between spells was unremarkable. EEG and brain MRI were normal. Whole-exome sequencing revealed a heterozygous pathogenic variant on *KCNMA1* (NM002247.4) c.2984A>G; p.N995S. Cataplexy, although typically associated with narcolepsy, has been reported in association with dystonia in patients with *KCNMA1* variants including the N995S variant (also called N999S, N1036S, or N1053S depending on the reference transcript).[1] Recognition is important because spells are a distinguishing feature and can improve with stimulant therapy.[2] Spells improved completely with lisdexamfetamine, but due to side effects the dose was reduced. Currently, she experiences 5 spells/day with improvements in language and development, which has been anecdotally reported.[2]

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