Teaching Video NeurolImage: 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. Neurological 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, was reported in association with dystonia in patients with KCNMA1 mutations including the N995S variant (also called N999S, N1036S or N1053S depending on the reference transcript). Recognition is important since spells are a distinguishing feature and can improve with stimulant therapy. 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. 

WNL-2022-201213_vid1 --- http://links.lww.com/WNL/C336
References


Video 1: Spells of dystonic cataplexy

Videos show four spells of loss of axial tone and behavioral arrest, as seen in cataplexy. However, in contrast to generalized loss of tone seen in typical cataplexy, there is dystonic posturing of the extremities. Some spells are triggered by excitement (playing with a ball, laughter) and end with yawning.
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