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). Recognition is important because 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.

**Acknowledgment**
The authors thank the patient’s family for gracefully sharing videos and providing consent for publication.

**Study Funding**
The authors report no targeted funding.

**Disclosure**
C.M. de Gusmao is a part-time employee at Mendelics Genomic Analysis and has received research grants from Ataxia-Telangiectasia Children’s Project and CureDRPLA and consulting fees from Ipsen therapeutics. L. Silveira-Moriyama has received speaker’s fee from Farmacimica and International Parkinson’s and Movement Disorders Society. The other authors report no relevant disclosures. Go to Neurology.org/N for full disclosures.

**Publication History**
Received by Neurology May 25, 2022. Accepted in final form August 12, 2022. Submitted and externally peer reviewed. The handling editor was Whitley Aamodt, MD, MPH.

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References

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Teaching Video NeuroImage: Dystonic Cataplexy in KCNMA1 Paroxysmal Movement Disorder
Neurology 2022;99;1010-1011 Published Online before print September 20, 2022
DOI 10.1212/WNL.0000000000201322

This information is current as of September 20, 2022