Early-onset oromandibular-laryngeal dystonia and Charlot gait

New phenotype of DYT-KMT2B

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A 28-year-old man had spasmodic dysphonia due to focal oromandibular-laryngeal dystonia from the age of 6, evolving rostrocaudally into a particular gait dystonia resembling Charlie Chaplin's *Charlot* gait (video 1). Recently he developed a progressive worsening of bulbar symptoms and a severe dysarthrophonia. No pyramidal or parkinsonism signs were observed. No intellectual dysfunction was noted. Head MRI was normal. Secondary causes of dystonia were excluded. Dystonia next-generation sequencing panel (58 genes) was negative. The sequencing of *KMT2B* identified a heterozygous de novo variant c5198-4_5206del(p?-intron24/exon25, classified as pathogenic. This case expands DYT-KMT2B's clinical phenotype due to early oromandibular-laryngeal involvement and atypical gait dystonia.^{1,2}

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Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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Name	Location	Role	Contribution
Ana Brás, MD	Coimbra University Hospital Center	Author	Drafting of the manuscript, concept, design, and critical revision of manuscript for intellectual content
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