Teaching Video NeuroImage: Facial-Faucial-Finger Myoclonus in Kufor-Rakeb Syndrome

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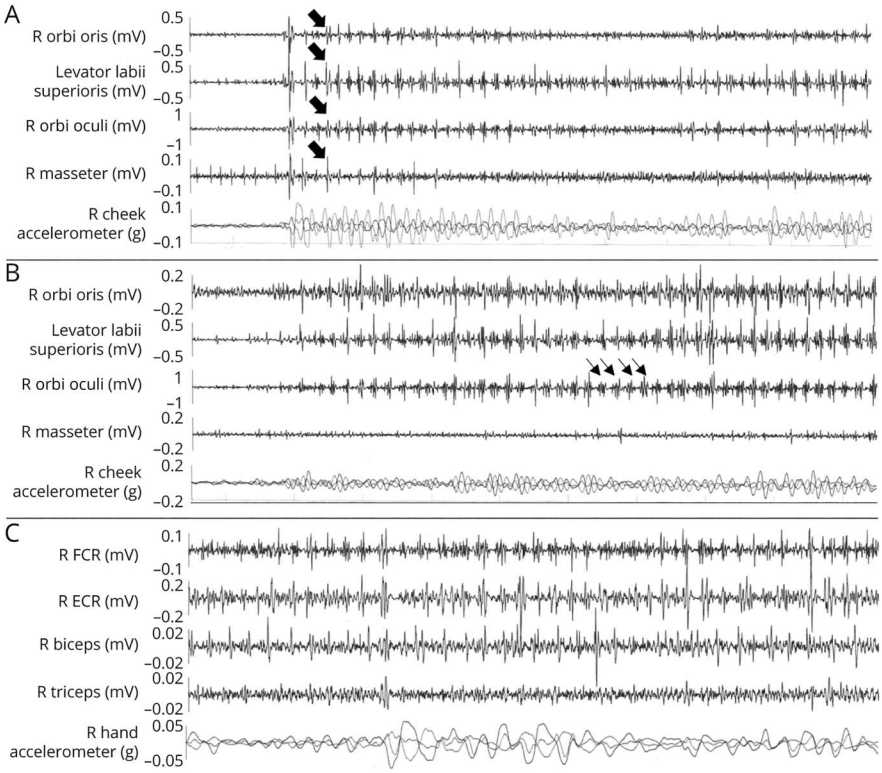
Figure 1 Surface EMG



Surface EMG showed both brief, synchronous <50 ms EMG bursting across facial muscles consistent with myoclonus (Panel A, wide arrows) and more sustained bursting typical of tremulous activity (Panel B, thin arrows). Upper limb surface EMG showed findings more consistent with a dystonic tremor, with continuous muscle activity present between discrete, tremulous EMG bursting (Panel C, arms extended). These neurophysiologic characteristics, to our knowledge not previously reported, demonstrate that what has been described phenomenologically as facial-faucial-finger myoclonus may have both myoclonic and tremulous features. ECR = extensor carpi radialis; FCR = flexor carpi radialis; orbi = orbicularis; R = right.

Two Chinese Australian siblings from nonconsanguineous parents presented with adolescent-onset dystonia-parkinsonism with prominent anxiety. Examination revealed eyelid, lower facial, and distal upper extremity myoclonus (Video 1), which was recorded using surface EMG (Figure 1). Compound heterozygous pathogenic variants in *ATP13A2* were identified, c.3176T>G (p.L1059R) and c.3253delC (p.L1088WfsX4), confirming the diagnosis of Kufor-Rakeb syndrome (KRS).[1] KRS classically presents as juvenile-onset, levodopa-responsive parkinsonism combined with pyramidal signs, upgaze palsy, cognitive decline, and, uniquely, facial-faucial-finger minimyoclonus.[2] This distinctive pattern of myoclonus is a useful clue to the diagnosis in affected individuals and distinguishes it from other forms of juvenile-onset parkinsonism.

MORE ONLINE

Video

Teaching slides
links.lww.com/WNL/C59

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Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

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2. Behrens MI, Brüggemann N, Chana P, et al. Clinical spectrum of Kufor-Rakeb Syndrome in the Chilean kindred with ATP13A2 mutations. *Mov Disord.* 2010; 25(12):1929-1937.

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