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Main Text
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, Surface EMG). 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.

Video 1 Legend:
Patient 1 demonstrated action-induced eyelid, facial, tongue and distal upper extremity myoclonus with vertical supranuclear gaze palsy and parkinsonism. “Faucial” myoclonus affecting the base of the tongue and palate was also present but not demonstrated on this video. Patient 2 had similar findings of parkinsonism, prominent facial and tongue myoclonus, and myoclonus affecting the fingers, particularly with action, and confirmed with surface EMG recording.

Figure 1, Surface EMG Legend:
Surface EMG showed both brief, synchronous <50ms 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 neurophysiological 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. R = right; Orbi = orbicularis; FCR = flexor carpi radialis; ECR = extensor carpi radialis

References:


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