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Teaching Video NeuroImage: Mirror Movements in a 57-Year-Old Woman With KMT2B-Related Dystonia

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Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data

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A 57-year-old woman had bilateral mirror movements (MM) since birth, which were evident on finger tasks or utensil use. Mild blepharospasm was observed on neurologic examination. Other neurologic and laboratory examinations and brain magnetic resonance imaging were normal. Her 34-year-old daughter had adolescent-onset segmental dystonia affecting the cervical, shoulder, and laryngeal muscles combined with mild MM (Video 1). Whole exome sequencing detected no pathogenic variant in *DCC*, *NTN1*, *RAD51*, or other known culprit genes for congenital MM¹. A heterozygous mutation in *KMT2B* (c. 1439C>T) was identified in the patient and her daughter, which was classified as likely pathogenic according to the ACMG guidelines. Although mirror dystonia has been widely reported in focal hand dystonia, bilateral congenital MM are rarely reported in dysonia before². The congenital MM of this patient and her daughter might be related to the *KMT2B*-related dystonia and the findings suggested a shared pathophysiology of dystonia and MM.

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Video legend

Video title: Mirror movements and dystonia of the patient and her daughter

Segment 1. The patient showed bilateral mirror movements during finger tasks and utensil use.

Segment 2. The patient showed mild blepharospasm.

Segment 3. The patient's daughter showed mirror movements during utensil use.

Segment 4. The patient's daughter showed segmental dystonia affecting the cervical, shoulder, and laryngeal.



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