

Neurology®

The most widely read and highly cited peer-reviewed neurology journal
The Official Journal of the American Academy of Neurology



Neurology Publish Ahead of Print

DOI: 10.1212/WNL.0000000000207144

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

Junyu Lin, MD¹; Chunyu Li, MD¹; Qirui Jiang, MD¹; Huifang Shang, MD¹

Corresponding Author:

Huifang Shang, hfshang2002@126.com

1. Department of Neurology, Rare Disease Center, Laboratory of Neurodegenerative Disorders, West China Hospital, Sichuan University, Chengdu, Sichuan Province, China

Equal Author Contribution:

Junyu Lin and Chunyu Li contributed equally to this work.

Neurology® Published Ahead of Print articles have been peer reviewed and accepted for publication. This manuscript will be published in its final form after copyediting, page composition, and review of proofs. Errors that could affect the content may be corrected during these processes. Videos, if applicable, will be available when the article is published in its final form.

Contributions:

Junyu Lin: Drafting/revision of the manuscript for content, including medical writing for content;
Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data

Chunyu Li: Drafting/revision of the manuscript for content, including medical writing for content;
Analysis or interpretation of data

Qirui Jiang: Major role in the acquisition of data

Huifang Shang: Drafting/revision of the manuscript for content, including medical writing for content;
Major role in the acquisition of data; Analysis or interpretation of data

Figure Count: 0**Table Count:** 0**Search Terms:**

[162] Dystonia, mirror movements

Acknowledgment:

We thank the patient and her family for participating in this study.

Study Funding:

Sichuan Science and Technology Program (Grant No. 2022ZDZX0023)

Disclosures:

The authors report no relevant disclosures.

Preprint DOI:**Received Date:**

2022-09-16

Accepted Date:

2023-01-19

Handling Editor Statement:

Submitted and externally peer reviewed. The handling editor was Resident and Fellow Deputy Editor Katherine Fu, MD.

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 dystonia 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.

References

1. Galléa C, Popa T, Billot S, Méneret A, Depienne C, Roze EJJ. Congenital mirror movements: a clue to understanding bimanual motor control. 2011;258(11):1911-9. doi:10.1007/s00415-011-6107-9
2. Cox B, Cincotta M, Espay AJT, movements oh. Mirror movements in movement disorders: a review. 2012;2doi:10.7916/d8vq31dz

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.

Neurology®

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

Junyu Lin, Chunyu Li, Qirui Jiang, et al.
Neurology published online February 16, 2023
DOI 10.1212/WNL.0000000000207144

This information is current as of February 16, 2023

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/early/2023/02/16/WNL.0000000000207144.citation.full
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): Dystonia http://n.neurology.org/cgi/collection/dystonia
Permissions & Licensing	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2023 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

