Stereotyped Upper Limb Movement in *MECP2* Duplication Syndrome

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Figure 1 Writhing of Arms and Hands in *MECP2* Duplication Syndrome

A 23-year-old man had epilepsy, mental retardation, and a stereotyped movement (figure 1 and video1), which was exacerbated by emotion. No abnormality was detected in the EEG during this movement. Array comparative genomic hybridization showed a Xq28 duplication: arr [GRCh37]Xq28(153032004_153406233)x2 (figure 2).

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Video

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Figure 2 Detection of the Xq28 Duplication by Using Array Comparative Genomic Hybridization

MECP2, IRAK1, and L1CAM were included in the duplication with a size of approximately 0.4 Mb. Genomic positions are based on data from the human genome assembly GRCH37/hg19.

The methyl-CpG-binding protein 2 gene (*MECP2*) duplication syndrome is a rare genetic disease whose causative gene is the same as that for Rett syndrome. The characteristic symptom of both diseases is stereotyped movement. The movement in *MECP2* duplication syndrome is writhing arms and hands,¹ unlike Rett syndrome.²

Study Funding

No targeted funding reported.

Disclosure

The authors declare that there is no conflict of interest regarding the publication of this article. Go to Neurology.org/N for full disclosures.

Appendix Authors

Name	Location	Contribution
Tomohiro Wakabayashi, MD	Sapporo Medical University School of Medicine; Hakodate Municipal Hospital, Hokkaido, Japan	Designed and conceptualized study, major role in the acquisition of data, analyzed the data, drafted the manuscript for intellectual content

Appendix (continued)

Name	Location	Contribution
Shinobu Fukumura, MD, PhD	Sapporo Medical University School of Medicine, Hokkaido, Japan	Designed and conceptualized study, major role in the acquisition of data, revised the manuscript for intellectual content
Satoru Takahashi, MD, PhD	Asahikawa Medical University, School of Medicine, Hokkaido, Japan	Major role in the acquisition of data
Kenji Kurosawa, MD, PhD	Division of Medical Genetics, Kanagawa Children's Medical Center, Japan	Major role in the acquisition of data
Shuichi Miyamoto, MD, PhD	Hakodate Municipal Hospital, Hokkaido, Japan	Interpreted the data, revised the manuscript for intellectual content
Kosuke Tsuchida, MD	Sapporo Medical University School of Medicine, Hokkaido, Japan	Interpreted the data, revised the manuscript for intellectual content
Shinsuke Kato, MD	Sapporo Medical University School of Medicine, Hokkaido, Japan	Interpreted the data, revised the manuscript for intellectual content
Takeshi Tsugawa, MD, PhD	Sapporo Medical University School of Medicine, Hokkaido, Japan	Interpreted the data, revised the manuscript for intellectual content

Continued

Appendix (continued)

Name	Location	Contribution
Yoshiyuki Sakai, MD, PhD	Sapporo Medical University School of Medicine, Hokkaido, Japan	Interpreted the data, revised the manuscript for intellectual content
Yukihiko Kawasaki, MD, PhD	Sapporo Medical University School of Medicine, Hokkaido, Japan	Designed and conceptualized study, major role in the acquisition of data, revised the manuscript for intellectual content

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2. Temudo T, Oliveira P, Santos M, et al. Stereotypies in Rett syndrome: analysis of 83 patients with and without detected MECP2 mutations. *Neurology*. 2007;68:1183-1187.

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Stereotyped Upper Limb Movement in *MECP2* Duplication Syndrome

Neurology® 2022;98:384. doi:10.1212/WNL.0000000000012757

In the Video NeuroImage article “Stereotyped Upper Limb Movement in *MECP2* Duplication Syndrome” by Wakabayashi et al.,¹ the first sentence should read: “A 23-year-old man had epilepsy, intellectual disability, and a stereotyped movement (Figure 1 and Video 1), which was exacerbated by emotion.” The authors regret the error.

Reference

1. Wakabayashi T, Fukumura S, Takahashi S, et al. Stereotyped upper limb movement in *MECP2* duplication syndrome. *Neurology*. 2021; 97(2):92-94.

Targeted Next-Generation Sequencing Panels in the Diagnosis of Charcot-Marie-Tooth Disease

Neurology® 2022;98:384. doi:10.1212/WNL.0000000000010635

In the article “Targeted Next-Generation Sequencing Panels in the Diagnosis of Charcot-Marie-Tooth Disease” by Cortese et al.,¹ the gene named throughout the article should be *FGD4*. In the amended supplementary table e-2 (doi.org/10.5061/dryad.kp8pb51), the mutation in patient 111 should be *PMP22*: c.68G>C p.(Thr23Arg), and the mutation in patient 21 should be *GARS*: c.880G>C p.Gly294Arg. The authors regret the errors.

Reference

1. Cortese A, Wilcox JE, Polke JM, et al. Targeted next-generation sequencing panels in the diagnosis of Charcot-Marie-Tooth disease. *Neurology*. 2020;94(1):e51-e61.

CORRECTION & REPLACEMENT

What Is the Role of Stathmin-2 in Axonal Biology and Degeneration?

Neurology® 2022;98:384. doi:10.1212/WNL.0000000000013018

In the Basic Science in the Clinic article “What Is the Role of Stathmin-2 in Axonal Biology and Degeneration?” by Benarroch,¹ a reference to a study by Melamed et al.² was inadvertently omitted at the beginning of the article and miscited elsewhere. The updated references and citations have been added in the replacement version of the article.

In addition, statements regarding TDP-43 in the first paragraph under the “Relationship Among STMN2, TDP-43, and Axonal Degeneration” heading have been corrected in the replacement version of the article. The author agrees with a reader who pointed out that the original wording was not substantiated.

The author regrets the omissions and errors. The original version with the changes highlighted is available from a link in the corrected article.

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1. Benarroch E. What is the role of stathmin-2 in axonal biology and degeneration? *Neurology*. 2021;97(7):330-333.
2. Melamed Z, Lopez-Erauskin J, Baughn MW, et al. Premature polyadenylation-mediated loss of stathmin-2 is a hallmark of TDP-43-dependent neurodegeneration. *Nat Neurosci*. 2019;22(2):180-190.