Stereotyped Upper Limb Movement in MECP2 Duplication Syndrome

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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. However, the movement in MECP2 duplication syndrome is writhing the arms and hands\(^1\), unlike Rett syndrome\(^2\).

A 23-year-old man had epilepsy, mental retardation, and a stereotyped movement (Figure 1 and Video, http://links.lww.com/WNL/B400), which was exacerbated by his own emotion. No abnormality was detected in the electroencephalogram when he performed this movement. The array comparative genomic hybridization (array-CGH) showed a Xq28 duplication: arr[GRCh37]Xq28(153032004_153406233)x2 (Figure 2).

**REFERENCES**


**Figure legends**

**Figure 1**

The movement in MECP2 duplication syndrome is writhing the arms and hands.
Figure 2

Detection of the Xq28 duplication by using array-CGH. *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.
Video Legend

Stereotyped Upper Limb Movement of MECP2 Duplication Syndrome

This was like a choreiform movement, and intermittent, spontaneous writhing movement of the both-side arms and hands that at time also involved the upper body without movement of lower extremities at all during this movement.
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