



Visual Diagnosis

Diagnostic Clue in Repetitive Hand Movements

Priyanka Madaan, DM, Bhanudeep Singanamala, MD, Lokesh Saini, DM*

Pediatric Neurology Unit, Department of Pediatrics, Postgraduate Institute of Medical Education and Research, Chandigarh, India



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This nine-year-old girl presented with progressive loss of developmental milestones. At age 18 months, she developed near-continuous mouthing and repetitive hand movements. These movements were present only while she was awake; they were restrainable, but she could not suppress them. The movements impaired the girl's ability to use objects. In addition, she had occasional generalized seizures and microcephaly. The physical examination revealed near-continuous complex mouthing and clapping-like hand stereotypies (video). *MECP2* gene sequencing revealed a pathogenic mutation suggestive of Rett syndrome.

Motor stereotypies are a common component of neurodevelopmental disorders. Rett syndrome and autism spectrum disorder are distinct disorders with several phenotypic commonalities. These disorders may be distinguished based on hand stereotypies' phenomenology.¹ Key descriptors of the stereotypies include the movements' pattern (clapping, mouthing, and washing), hand position (midline or away from the body), laterality (bilateral or unilateral), and intricacy (simple, involving one group of muscles versus complex, with clusters of several coordinated movements).^{1,2} With Rett syndrome, the occurrence of midline

stereotypies carries a worse prognosis.^{1,3} The pathophysiology of motor stereotypies is unclear. Proposed hypotheses include synaptopathies, basal ganglia circuitry abnormalities, and neurotransmitter abnormalities.⁴

Supplementary Data

The video related to this article can be found at <https://doi.org/10.1016/j.pediatrneurol.2019.03.012>.

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* Communications should be addressed to: Dr. Saini; Assistant Professor; Pediatric Neurology Unit; Department of Pediatrics; Postgraduate Institute of Medical Education and Research; Chandigarh, India-160012.

E-mail address: drlokeshsaini@gmail.com (L. Saini).