

Teaching NeuroImages: An Imaging Clue in a Boy With Developmental Delay

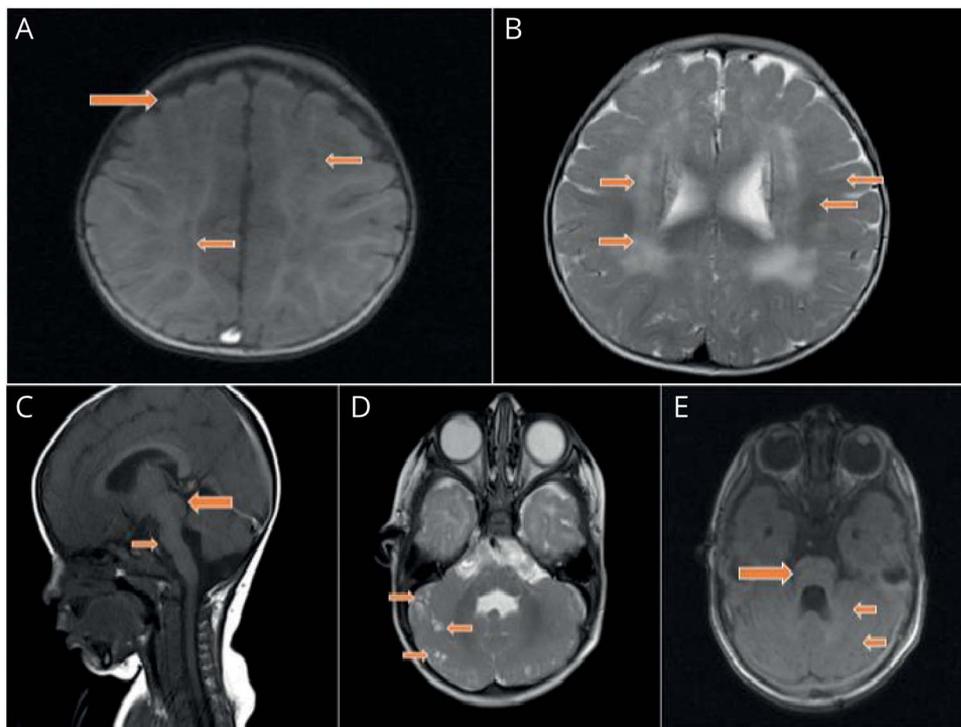
A.S. Jyotsna, DNB, K.P. Vinayan, DM, and Arun Grace Roy, DM

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Correspondence

Dr. K.P. Vinayan
vinayankp@aims.amrita.edu

Figure MRI Brain Showing Characteristic Structural Anomalies of Congenital Muscular Dystrophy (POMGNT1 Mutation)



(A) T1-weighted axial image: frontally predominant dysplastic cortex with poor gray-white matter differentiation with a cobblestone lissencephaly pattern (large arrow), islands of gray matter in white matter indicating heterotopia (small arrows). (B) T2-weighted axial image: hyperintensities in central white matter (right arrows) with relatively normal subcortical white matter (left arrows). (C) T1-weighted sagittal image: midbrain hypoplasia, fused colliculi with a thick tectum (large arrow) and relatively flat pons (small arrow). (D) T2-weighted axial image: lateral cerebellar cysts (arrows) frequently reported in *POMGNT1* mutations. (E) T1-weighted axial image: pontocerebellar hypoplasia (right large arrow) with poorly formed cerebellar foliation (left small arrows).

A 6-year-old boy of nonconsanguineous parents presented with global developmental delay since infancy with history of 2 episodes of seizures in early childhood. Examination showed hyperactivity, subnormal cognition, tight heel cords, weakness of limbs along with hypotonia, and sluggish tendon reflexes. Serum creatine kinase was 2,138 IU/L.

MR brain showed developmental malformations highly suggestive of α -dystroglycanopathy variant of congenital muscular dystrophy (figure, A–E). Next generation sequencing showed 2 compound heterozygous variants of unknown significance in the *POMGNT1* gene (Exon17

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From the Department of Pediatric Neurology, Amrita Institute of Medical Sciences, Cochin, Kerala, India.

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[chr1:46657796C>T] and Exon4 [chr1:46662444A>G]), in silico predictions of which were probably damaging by PolyPhen-2 and damaging by Mutation Taster 2.

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Disclosure

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Appendix Authors

Name	Location	Contribution
A.S. Jyotsna, DNB	Amrita Institute of Medical Sciences, Cochin, Kerala, India	Collected the clinical information and prepared the initial draft

Appendix *(continued)*

Name	Location	Contribution
K.P. Vinayan, DM	Amrita Institute of Medical Sciences, Cochin, Kerala, India	Crafted the manuscript, revised it critically for the intellectual content, responsible for correspondence
Arun Grace Roy, DM	Amrita Institute of Medical Sciences, Cochin, Kerala, India	Analyzed data and approved the final version to be published

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