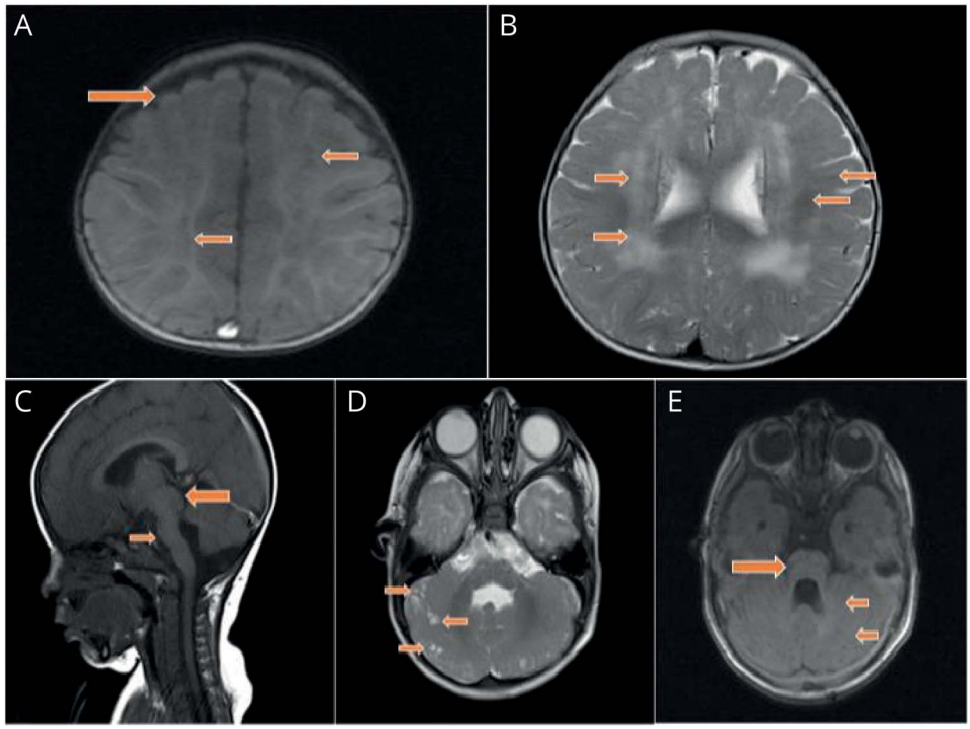
Teaching NeuroImages: An Imaging Clue in a Boy With Developmental Delay

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

Neurology® 2021;96:e1925-e1926. doi:10.1212/WNL.0000000000011286

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 heterotropia (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

MORE ONLINE

→Teaching slides
links.lww.com/WNL/B292

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

Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

[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.

Study Funding

No targeted funding reported.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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
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

References

1. Bonnemann CG, Wang CH, Quijano-Roy S, et al. Diagnostic approach to the congenital muscular dystrophies. Neuromuscul Disord 2014;24:289–311.
2. Biancheri R, Bertini E, Falace A, et al. POMGnT1 mutations in congenital muscular dystrophy: genotype-phenotype correlation and expanded clinical spectrum. Arch Neurol 2006;63:1491–1495.

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

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

Neurology 2021;96;e1925-e1926 Published Online before print December 4, 2020
DOI 10.1212/WNL.0000000000011286