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
[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.

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**References**

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

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