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

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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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[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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References

Appendix

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<tr>
<th>Name</th>
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<th>Contribution</th>
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Appendix Authors

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