Teaching Neuroimages: An imaging clue in a boy with developmental delay

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Abstract

6 year old boy of non-consanguineous parents presented with global developmental delay since infancy with history of two 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 CK was 2138 IU/l.

MR brain showed developmental malformations highly suggestive of alpha dystroglycanopathy variant of congenital muscular dystrophy. (Figure 1, A-E). Next generation sequencing showed two compound heterozygous variants of unknown significance in POMGNT-1 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.

Appendix 1: Authors

<table>
<thead>
<tr>
<th>Name</th>
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<tbody>
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REFERENCES-


Figure 1. (Arrows added to mark all the salient MRI findings described in the figure legend)

Figure 1. MRI brain showing characteristic structural anomalies of congenital muscular dystrophy (POMGNT1 mutation)

A. T1 W axial images: Frontally predominant dysplastic cortex with poor grey-white matter differentiation with a cobblestone lissencephaly pattern (big arrow), islands of grey matter in white matter indicating heterotropia (small arrow)
B. T2 W axial images: Hyperintensities in central white matter (right arrows) with relatively normal subcortical white matter (left arrows).
C. T1 W sagittal images: Midbrain hypoplasia, fused colliculi with a thick tectum (big arrow) and relatively flat pons (small arrow)
D. T2 W axial Images: Lateral cerebellar cysts (arrows) frequently reported in POMGNT1 mutations.
E. T1 W axial images: Pontocerebellar hypoplasia (right big arrow) with poorly formed cerebellar foliation (left small arrows).
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