Teaching Neurorimage: ROBO3 Mutation Causing Horizontal Gaze Palsy and Brainstem Malformation

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Neurology® Published Ahead of Print articles have been peer reviewed and accepted for publication. This manuscript will be published in its final form after copyediting, page composition, and review of proofs. Errors that could affect the content may be corrected during these processes.

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A 10-month-old boy presented with motor developmental delay, torticollis, bilateral abduction restriction (incomplete horizontal gaze palsy), and left lower motor neuron facial palsy. His brain MRI demonstrated brainstem malformations, including absent facial colliculi (Figure, A), clefting of the medulla and pons (Figure, B), butterfly configuration of the medulla (Figure, C), and concave dorsal pontine border (Figure, D). Genetic testing revealed a homozygous missense mutation [c.437G>C (p.Arg146Pro)] in exon 2 of ROBO3 gene. Horizontal gaze palsy with progressive scoliosis (HGPPS1) results from axonal guidance signalling defects caused by ROBO3 mutations. The main symptoms include congenital horizontal gaze palsy, horizontal pendular nystagmus, and progressive scoliosis after two years of age. The radiological differential for this hindbrain malformation is ‘horizontal gaze palsy with progressive scoliosis-2’, caused by mutation in the DCC gene. Children with HGPPS2 also demonstrate intellectual impairment and agenesis of the corpus callosum.

http://links.lww.com/WNL/C576

References:


Teaching_NeuroImage: *ROBO3* Mutation Causing Horizontal Gaze Palsy and Brainstem Malformation
Geetha Chanda, Nihaal Reddy, Ramesh Konanki, et al.
*Neurology* published online December 23, 2022
DOI 10.1212/WNL.0000000000206821

This information is current as of December 23, 2022