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Functional magnetic resonance imaging and diffusion tensor imaging findings in a patient with ROBO3-related horizontal gaze palsy with progressive scoliosis

Achados de functional magnetic resonance imaging e diffusion tensor imaging em um paciente com paralisia da mirada horizontal com progressiva escoliose, relacionado ao gene ROBO3

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A 24-year-old-woman woman presented with birth-onset horizontal gaze ophthalmoplegia. Past medical history was unremarkable. Her brother had similar findings with severe progressive scoliosis, requiring surgery. After 12 years of follow-up, the patient developed mild head tremor which did not bother her and nightmare disorders that was successfully treated with nortriptyline. Spine X-rays revealed mild scoliosis. Whole exome analysis revealed c.906-17G>A mutation in ***ROBO3*** splicing site, confirming horizontal gaze palsy with progressive scoliosis (HGPPS). Diffusion tensor imaging (DTI) tractography revealed parallel non-decussating corticospinal tracts (Figure 1A), which markedly diverges from healthy controls (Figure 1B). Motor task functional magnetic resonance imaging (fMRI) revealed ipsilateral activation of the primary motor cortex (Figure 2).

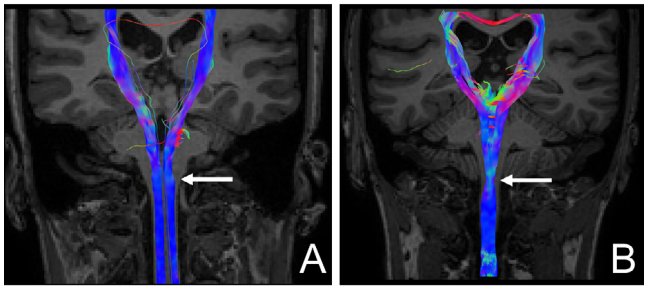

Figure 1. Diffusion tensor imaging tractography with adjusted parameters to evaluate the brainstem and the upper portion of the spinal cord. (A) Arrow indicates the absence of corticospinal tracts decussation in a patient with ROBO3-related horizontal gaze palsy with progressive scoliosis. (B) Diffusion tensor imaging tractography obtained from a healthy subject at Universidade Estadual de Campinas (Neuroimaging Laboratory database). Arrow highlights normal appearance of pyramidal decussation.

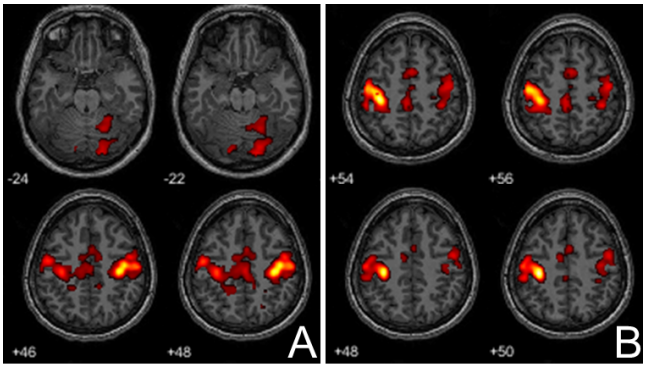

Figure 2. Functional magnetic resonance imaging images obtained from a patient with ROBO3-related horizontal gaze palsy with progressive scoliosis (images printed in neurological convention). (A) Predominant right primary motor cortex activation (yellow) while performing right-hand motor task. It is also noticeable that cerebellar activation (red) is also ipsilateral, confirming it was not due to technical error. (B) Predominant left primary motor cortex activation (yellow) while performing left-hand motor task.

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