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Cortico-subcortical white matter bundles alterations in cervical dystonia and blepharospasm

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It is now thought that dystonia is due to an involvement of a network including basal ganglia, cerebellum, thalamus, and sensorimotor cortices [1–3]. In line with this hypothesis, several studies have found abnormalities in the basal ganglia thalamo-cortical circuit in patients with dystonia [1,4–8]. We aimed to investigate in cervical dystonia (CD) and blepharospasm (BSP) possible microstructural changes of white matter (WM) bundles connecting pre-defined subcortical and cortical regions involved in the network underlying the pathophysiology of focal dystonia and possible correlations between WM microstructural damage and clinical features of dystonic patients. Thirty-five patients (17 with CD and 18 with BSP) and 17 healthy subjects underwent MRI, including diffusion tensor imaging (DTI). Probabilistic tractography (BedpostX) was performed to reconstruct WM tracts connecting globus pallidus, putamen, and thalamus, with primary motor, primary sensory, and supplementary motor cortices. WM tract integrity was evaluated by deriving their DTI metrics. Significant differences in mean, radial and axial diffusivity between CD and HS, and between BSP and HS were found in the majority of the reconstructed WM tracts, while no differences were found between the two groups of patients. We also found a significant correlation between the extent of WM damage and the clinical severity in patients with blepharospasm, but not in patients with CD. The observation of abnormalities in DTI metrics of specific WM tracts suggests a diffuse and extensive loss of WM integrity as a common feature of CD and BSP, converging to the increasing evidence of microstructural damage of several brain regions belonging to specific circuits, i.e., the basal ganglia-thalamo-cortical circuit, thus likely reflecting a common pathophysiological mechanism of focal dystonia.

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