Post-traumatic cervical dystonia in a patient with neurofibromatosis type 1

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A 6-year-old boy without any past medical history visited to our hospital, complaining of spasmodic torticollis. The symptom has come suddenly after he slipped down in bathroom. Although 3 months of treatment with soft collar apply and halter traction, the patient's symptom did not improve. On neurological examination, we couldn't find any focal neurological sign except spasmodic torticollis. Cervical spine x-ray and computed tomography scans revealed atlantoaxial rotatory subluxation. MRI of the cervical spine demonstrated extensive intramedullary hyperintensities without enhancement at the C1-C4 levels on sagittal T2-weighted image. Cerebrospinal fluid analysis was within normal range and anti-aquaporin-4 antibody test was also negative. Brain MRI demonstrated multifocal hyperintensities in the bilateral basal ganglia and left cerebellar peduncle on T2-weighted and fluid-attenuated inversion recovery images, suggesting neurofibromatosis-associated multiple gliomas. The patient has 7 cafe'-au-lait spots on his abdomen, back, and hip. In addition, NF1 gene mutation was found by genetic testing. Any abnormality or trauma of the cervical spine can present with spasmodic torticollis. Trauma, including minor trauma (sprains/strains), fractures, dislocations, and subluxations, often result in spasms of cervical musculature. It has been known that most cases of post-traumatic cervical dystonia are self-limited, and symptoms resolve in 1-2 weeks. However, our patient's symptom lasted more than 3 months without improvement. We assume that not only minor trauma but also NF contributed to the development of acute cervical dystonia in this case, since pathologic and neuroradiologic studies revealed focal lesions involving the basal ganglia, suggesting a pathogenic mechanism of focal dystonia.