Antisaccades in Spinocerebellar Ataxia Type 17 With Middle Cerebellar Peduncle Hyperintensities Without Hot-Cross-Bun Sign

Dear Editor,

Spinocerebellar ataxia type 17 (SCA17) is a rare genetic disorder affecting the nervous system that is characterized by ataxia, chorea, dystonia, and dementia.1 Brain magnetic resonance imaging (MRI) reportedly indicates variable atrophy of the cerebrum, brainstem, and cerebellum.1 Here we report a case of SCA17 with a high signal intensity in the bilateral middle cerebellar peduncle (MCP) in T2-weighted brain MRI.

A 54-year-old female presented with a 6-month history of dizziness and imbalance. Her symptoms had started to worsen 3 months previously, to the extent that she could hardly rotate, which prevented her from performing rotation exercises for Pungmulnori, a traditional Korean art form. Before the symptoms appeared 6 months previously, she had experienced no problems with performing the daily activities of living or in cognition. She denied symptoms of rapid eye movement (REM) sleep behavior disorder, which is characterized by abnormal and often violent behaviors during the REM stage of sleep. Moreover, she had no significant family history of cerebellar dysfunction, but her mother, who died of rectal cancer, had complained of dizziness. The patient was taking antihypertensive medication and had previously undergone surgery for right knee osteoarthritis and lumbar spinal stenosis. She had been diagnosed with osteoarthritis and was being followed up by a rheumatology department. She had no recent weight loss, but gained approximately 8 kg during the previous year. Her vital signs were stable. While she did not notice dysarthria or dysphagia, a neurological examination revealed titubation of the head and trunk and slurred speech without dysphagia. Video-oculography showed no spontaneous or gaze-evoked nystagmus, decreased smooth-pursuit gain, or prominent antisaccade hypometria compared to prosaccades (Fig. 1A and B). Although directional errors were not observed during the antisaccade test, the occurrences of multiple-step saccades and the saccade duration both increased (Supplementary Fig. 1 in the online-only Data Supplement). The results for the video head impulse test, bithermal caloric test, and ocular/cervical vestibular evoked myogenic potential were unremarkable (Supplementary Fig. 2 in the online-only Data Supplement). Motor power and the deep tendon reflex were preserved. Vibration sense was decreased by approximately 30% in the lower extremities, but pain and temperature senses were normal. Limb ataxia was more prominent in the left extremity. Moreover, gait ataxia was observed. However, the Romberg sign was negative, and no pyramidal or extrapyramidal sign (including chorea) was observed. She scored 9 out of 40 on the Korean version of the Scale for the Assessment and Rating of Ataxia. Serological tests revealed that rheumatoid factor was elevated at 77.7 IU/mL (normal range: 0–18 IU/mL) with no other abnormal rheumatological laboratory findings. Paraneoplastic antibody tests (Hu, Ri, and Yo) and antiganglioside antibodies were all negative. A nerve conduction study and electromyography revealed median neuropathy...
at the wrist. Triplet-repeat primed polymerase-chain-reaction fragment analysis for SCA17 revealed 38 CAG/CAA repeats in the TATA-box protein gene on 1 allele and 43 on the other. Other genetic tests, including of SCA1, -2, -3, -6, -7, and -8, produced negative findings. Brain MRI revealed cerebellar atrophy with a T2-weighted hyperintensity in the MCP without a hot-cross-bun sign, as well as prominent brainstem atrophy (Fig. 1C). The autonomic function test showed transient delayed orthostatic hypotension of a nonneurogenic type without dysautonomic symptoms (Supplementary Fig. 3 in the online-only Data Supplement).

This is the first report of neurotological findings in SCA17 with bilateral MCP abnormalities in MRI. The MCP, which is also called the brachium pontis, is the most-extensive afferent system of the cerebellum. It contains pontocerebellar tract fibers from the contralateral pontine nuclei. Bilateral hyperintensities on T2-weighted images of the MCPs have recently been reported, with the “MCP sign” occurring in different diseases, including neuromyelitis optica, Wallerian degeneration, toxic encephalopathy, and lymphoma in the primary central nervous system. Additionally, the MCP sign can also appear in other conditions that cause degenerative cerebellar ataxia, including multiple-system atrophy—cerebellar type (MSA-C) and fragile X-associated tremor/ataxia syndrome (FXTAS). However, the present study did not produce genetic findings of FXTAS. Previous imaging data inferred that the MCP sign could be accompanied by a hot-cross-bun sign in MSA-C and corpus callosum thinning in FXTAS. In our case, the MCP sign without the hot-cross-bun sign could explain both the saccade velocity and the range of eye motion being preserved in patients with SCA17, indicating the relative preservation of pontine nuclei. Our patient exhibited the interesting ability in the antisaccade test of initiating a voluntary saccade after suppressing a reflexive one. This test is a useful tool for investigating cortical and subcortical function. Previous pathological studies of SCA17 showed cerebellar atrophy as the predominant feature, moderate atrophy in the cerebral cortex, mild atrophy in the midbrain, and spared pontine nuclei, which support the preserved saccade velocity and prominent saccade hypometria during antisaccades in our patient. However, the repeat size and disease duration have been reported to differ, with saccade velocities having decreased in some recent SCA17 cases.9,10

Supplementary Materials
The online-only Data Supplement is available with this article at https://doi.org/10.3988/jcn.2023.0397.

Ethics Statement
This study was conducted retrospectively, and informed consent was waived.

Availability of Data and Material
The datasets generated or analyzed during the study are available from the corresponding author on reasonable request.

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REFERENCES