A 20-year-old man presented with dizziness and gait disturbance for approximately 2 months. Neurologic examination revealed spontaneous downbeat nystagmus (DBN), limb ataxia, and abnormal tandem gait (video on the Neurology® Web site at Neurology.org). Video-oculography showed augmented DBN during position change (figure 1). MRI revealed diffuse atrophy and signal changes in the brainstem and cerebellum (figure 2). The plasma levels of very long-chain fatty acids (C26:0 concentration, C24:0/C22:0 ratio, and C26:0/C22:0 ratio) were elevated. Subsequent genetic testing revealed a missense mutation in the ABCD1 gene. The olivopontocerebellar form of X-linked adrenoleukodystrophy should be considered a rare but possible diagnosis in young men with dizziness and DBN.1,2

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Author contributions: S.-H. Kim conceived of the study, analyzed the data, reviewed the literature, drafted the manuscript, and approved the final version of the manuscript. S.-S. Kim interpreted the video-oculographic data, critically reviewed the manuscript, and approved the final version of the manuscript. H. Ha interpreted neuroimaging data, critically reviewed and edited the manuscript, and approved the final version of the manuscript. S.-H. Lee conceived of the study, interpreted the data, critically reviewed and edited the manuscript, and approved the final version of the manuscript.

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Brain MRI reveals atrophy and high signal intensity in the cerebellum, brainstem, and middle cerebellar peduncles on T2-weighted (A), fluid-attenuated inversion recovery (B), and gadolinium-enhanced T1 images (C).


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