X-linked adrenoleukodystrophy presenting with positional downbeat nystagmus

Figure 1 Video-oculographic recording of vertical eye movements

Video-oculography shows spontaneous downbeat nystagmus (1°/s) augmented by position change (head bending, 10°/s; straight head hanging, 24°/s).

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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Supplemental data
at Neurology.org

Figure 2 Brain MRI with contrast enhancement

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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