

Teaching NeuroImages: Cerebral adrenoleukodystrophy

A rare adult form

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A 42-year-old man with Addison disease presented to the emergency department with unsteady gait for 1 year. A half-brother had epilepsy and difficulty walking, and died at 6 years of age. Another half-brother has a long-standing gait disorder. Over 3 to 4 months, his gait has become worse with marked stiffness, ataxia, and dysarthria. Neurologic examination demonstrated saccadic pursuit, ocular flutter, severe spasticity throughout, peripheral neuropathy, hyperreflexia, clonus, with bilateral extensor plantar reflexes, marked dysmetria, and dysdiadochokinesia. He stands with a marked forward stoop and flexed knees. MRI is shown in figures 1 and 2. The plasma concentration of very-long-chain fatty acids, which is a general test for peroxisomal disorders, was elevated, confirming the diagnosis of adrenoleukodystrophy (ALD). Very-long-chain fatty acids were also elevated in the surviving brother's plasma.

Adult cerebral ALD is extremely rare. It is an X-linked peroxisomal disorder caused by mutation in the *ABCD1* gene located at Xq28 and involves the nervous system, adrenal cortex, and Leydig cells in the testes.^{1,2}

STUDY FUNDING

No targeted funding reported.

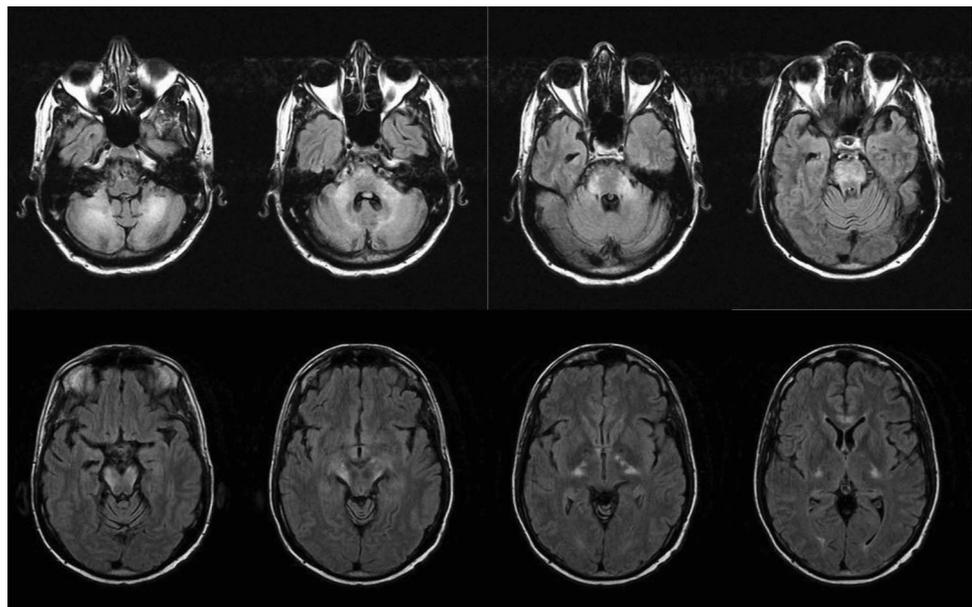
DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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Figure 1 Axial fluid-attenuated inversion recovery of brain MRI

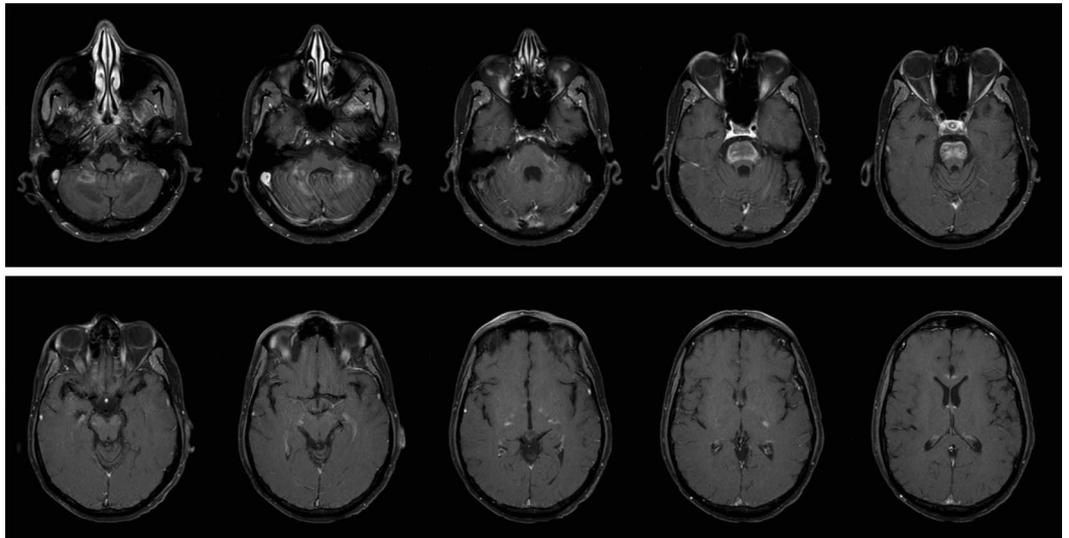


Hyperintensity in the cerebellum, middle cerebellar peduncles, pons, cerebral peduncle, internal capsule, and genu of corpus callosum.

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Figure 2 Brain MRI with gadolinium



Gadolinium enhancement of the cerebellum, middle cerebellar peduncles, pons, cerebral peduncle, internal capsule, and genu of corpus callosum.

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