

# Teaching NeuroImages: Cerebral adrenoleukodystrophy

## A rare adult form

Rania A. Elenein, MD  
Sunil Naik, MD  
Stephanie Kim, MD  
Vineet Punia, MD  
Karin Jin, MD

Correspondence to  
Dr. Elenein:  
raniadml@yahoo.com or  
abouera@umdnj.edu.

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.<sup>1,2</sup>

### STUDY FUNDING

No targeted funding reported.

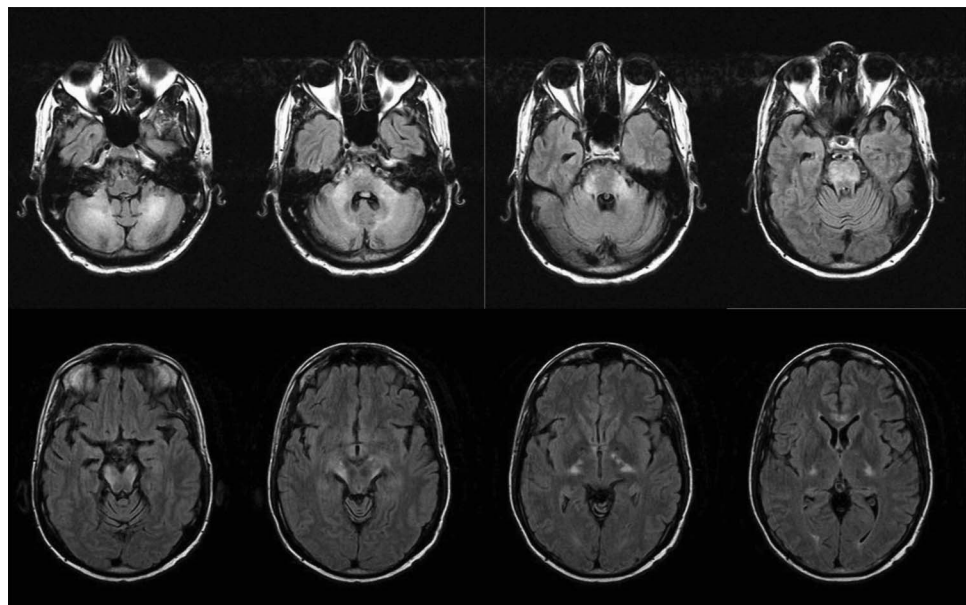
### DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to [Neurology.org](http://Neurology.org) for full disclosures.

### REFERENCES

1. Moser H, Dubey P, Fatemi A. Progress in X-linked adrenoleukodystrophy. *Curr Opin Neurol* 2004;17:263–269.
2. Moser HW, Raymond GV, Dubey P. Adrenoleukodystrophy: new approaches to a neurodegenerative disease. *JAMA* 2005; 294:3131–3134.

**Figure 1** Axial fluid-attenuated inversion recovery of brain MRI

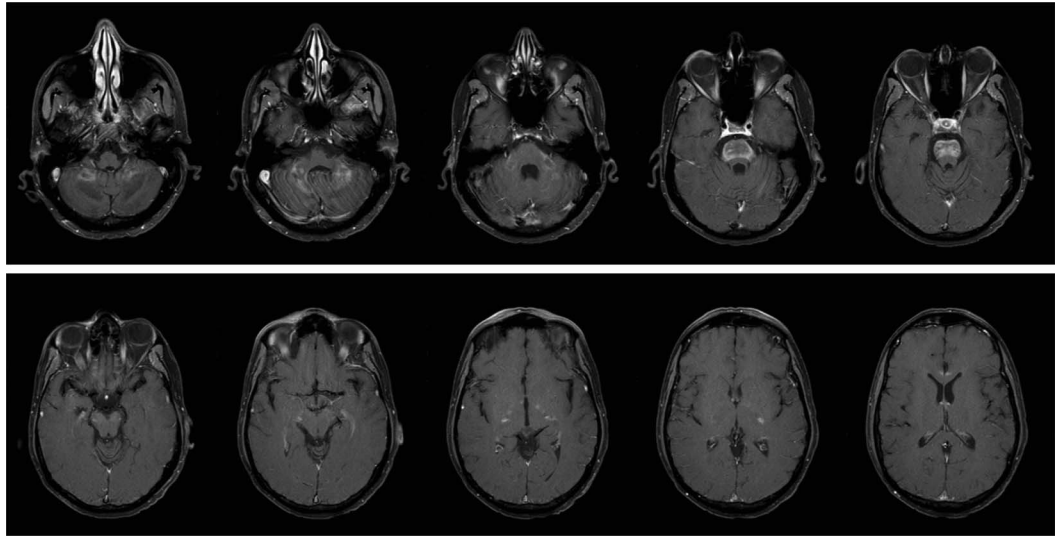


Hyperintensity in the cerebellum, middle cerebellar peduncles, pons, cerebral peduncle, internal capsule, and genu of corpus callosum.

From the UMDNJ–New Jersey Medical School, Newark, NJ.

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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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*Neurology* 2013;80:e69-e70

DOI 10.1212/WNL.0b013e3182815416

**This information is current as of February 4, 2013**

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