# Teaching NeuroImages: X-linked adrenoleukodystrophy

# Spinocerebellar variant

Lisa W.C. Au, MD, Anne Y.Y. Chan, MD, and Vincent C.T. Mok, MD Neurology® 2019;93:e731-e732. doi:10.1212/WNL.0000000000007958

A 29-year-old previously healthy man presented with 2 months' history of progressive dysarthria and unsteady gait. Examination revealed dysmetria, lower limbs hyperreflexia, and ankle

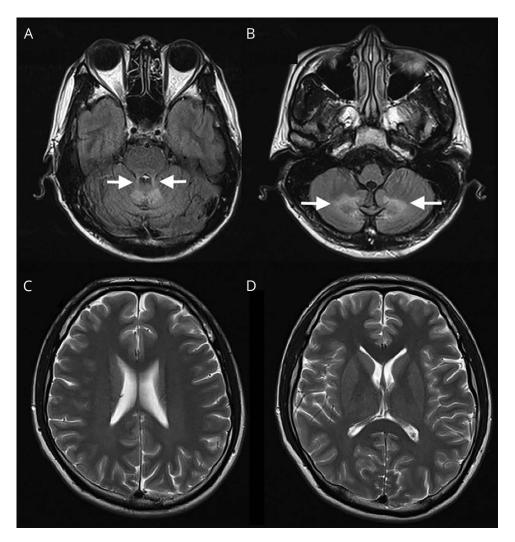
Figure MRI brain of the patient

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Axial fluid-attenuated inversion recovery images show hyperintensity over bilateral middle cerebellar penducles (A) and cerebellar white matter (B). The cerebral cortex is spared, as shown on axial T2 images (C, D).

From Lui Che Woo Institute of Innovative Medicine, Margaret K.L. Cheung Research Centre for Management of Parkinsonism, Chinese University of Hong Kong. Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article. clonus. MRI showed symmetrical T2/fluid-attenuated inversion recovery hyperintensity over bilateral middle cerebellar peduncles and cerebellar white matter but sparing the cerebral cortex (figure). Very long-chain fatty acids were detected in blood. Genetic testing showed hemizygous p.Arg554His mutation in the *ABCD1* gene, confirming the diagnosis of adrenoleukodystrophy (ALD). ALD is an X-linked peroxisomal disorder. Lesions are typically found in the cerebral white matter. The spinocerebellar variant is a rare phenotype and has been estimated to account for 1%–2% of ALD.<sup>2</sup>

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#### **Disclosure**

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

### **Appendix** Authors

Name	Location	Role	Contribution
Lisa W.C. Au, MD	Chinese University of Hong Kong, HKSAR	Author	Designed and conceptualized study, analyzed the data, drafted the manuscript for intellectual content
Anne Y.Y. Chan, MD	Chinese University of Hong Kong, HKSAR	Author	Major role in the acquisition of data
Vincent C.T. Mok, MD	Chinese University of Hong Kong, HKSAR	Author	Revised the manuscript for intellectual content

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