

Teaching NeuroImages: Frontal lobe involvement in adult-onset cerebral X-linked adrenoleukodystrophy

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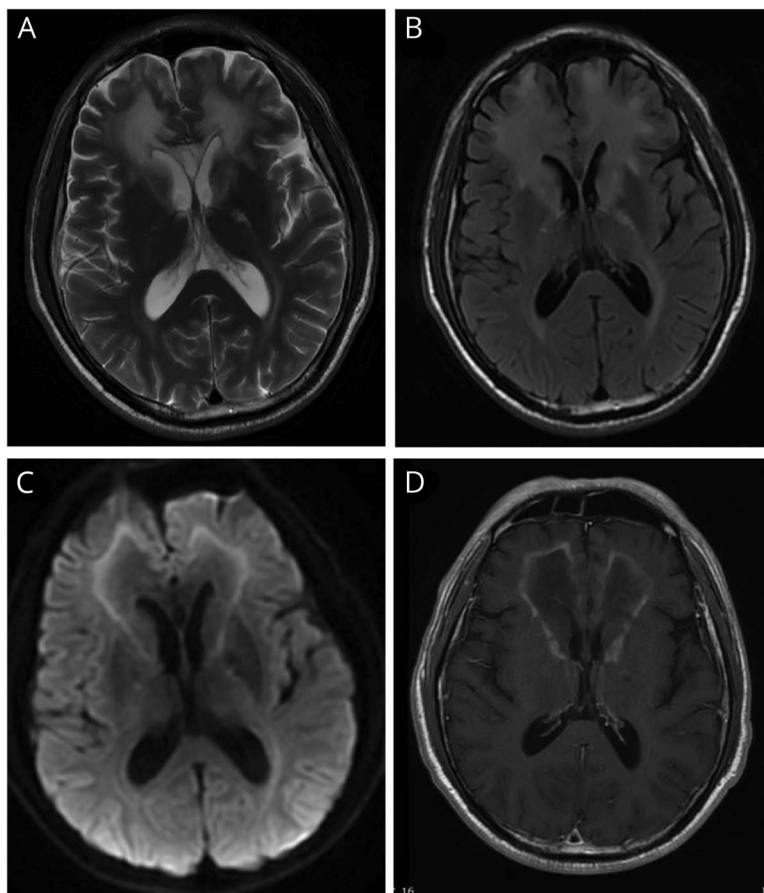
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A 42-year-old man presented with 1 year of progressive behavioral and personality changes. His antenatal, birth, and neurodevelopment were unremarkable. Examination showed hyperactive behavior, lower limbs paralysis (Medical Research Council grading 3) with pyramidal signs, and no skin hyperpigmentation or visual disturbance. Brain MRI revealed diffuse lesions in both frontal lobes, with peripheral rim restricted diffusion and enhancement (figure 1). Elevated plasma very-long-chain fatty acid level, p.Ser98Ter mutation in *ABCD1* gene, and his pedigree (figure 2) confirmed

Figure 1 Brain MRI



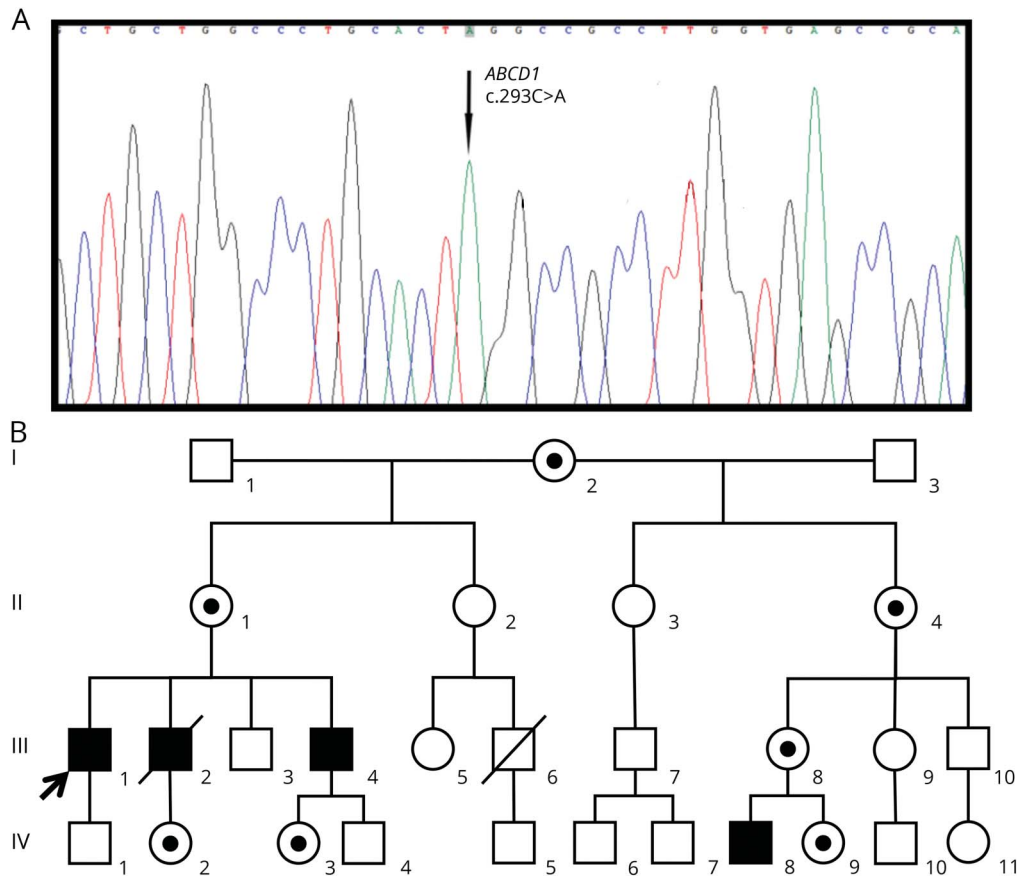
Axial T2-weighted (A) and fluid-attenuated inversion recovery (B) images show diffuse white matter hyperintensity in both frontal lobes. Diffusion-weighted imaging sequence reveals peripheral rim restricted diffusion in the white matter of the frontal lobes (C), with enhancement (D).

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Figure 2 Genetic testing and family tree



(A) Sequence determination of c.293C>A (p.Ser98Ter) mutation of the *ABCD1* gene. The arrow indicates the involved nucleotide. (B) Family tree: The presented case is marked with an arrow. III-2, III-4, and IV-8 had X-linked adrenoleukodystrophy; I-2, II-1, II-4, III-8, IV-2, IV-3, and IV-9 were female carriers.

the diagnosis of adult-onset cerebral X-linked adrenoleukodystrophy (X-ALD). X-ALD is an inborn error of metabolism predominantly within posterior involvement including parieto-occipital lobes and splenium of the corpus callosum; about 15% of patients have an anterior pattern of atypical involvement.^{1,2} Frontal involvement can be seen in X-ALD, and this imaging feature can expedite the diagnosis of this atypical X-ALD.

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Disclosure

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

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Name	Location	Role	Contribution
Chenchen Liu, MD	Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology	Author	Study concept and design, acquisition of data, analysis and interpretation of data

Appendix (continued)

Name	Location	Role	Contribution
Yayun Cao, MD	Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology	Author	Study concept and design, acquisition of data, analysis and interpretation of data
Bitao Bu, MD	Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology	Author	Study concept and design, critical revision of manuscript for intellectual content

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