

X-linked adrenoleukodystrophy presenting as progressive ataxia and pure cerebellar involvement

Adrenoleucodistrofia ligada ao X apresentando-se como ataxia progressiva e envolvimento cerebelar isolado

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A 27-year-old man presented with a two-year history of progressive ataxia. Family history was unremarkable. Examination revealed ataxia and alopecia. Serum cortisol levels were low, suggesting adrenal insufficiency. Brain magnetic

resonance imaging (MRI) disclosed cerebellar white matter involvement (Figure 1). Exome sequencing showed homozygous mutations (c.268del p.Glu90Argfs*13) in the *ABCD1* gene and confirmed X-linked adrenoleukodystrophy (X-ALD).

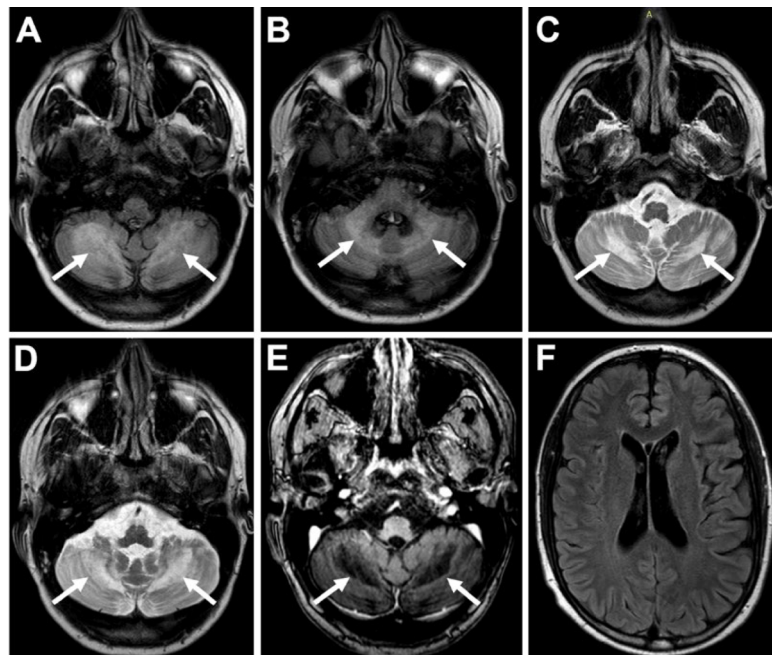


Figure 1. Patient with pure cerebellar ataxia related to X-linked adrenoleukodystrophy. Axial FLAIR-weighted brain MRI shows symmetrical cerebellar white matter and middle cerebellar peduncles hyperintense signal, besides pontocerebellar atrophy (A and B). Axial T2-weighted brain MRI discloses bilateral cerebellar white matter changes (C and D). Axial T1-weighted brain MRI reveals hypointense signal in cerebellar white matter (E). Axial FLAIR-weighted brain MRI shows no supratentorial abnormalities (F).

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Several forms of hereditary ataxias remain undetermined, despite being largely investigated. Whole-exome sequencing is a useful diagnostic approach for undetermined ataxias¹. Adult-onset

X-ALD usually presents with behavioral changes, pyramidal signs, and white matter changes. Pure cerebellar white matter changes with progressive cerebellar ataxia are uncommon in X-ALD².

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