

Adult-onset cerebral X-linked adrenoleukodystrophy presenting as obsessive-compulsive disorder

Adrenoleucodistrofia ligada ao X forma cerebral do adulto manifestando como transtorno obsessivo-compulsivo

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A 35-year-old man presented with a five-year history of obsessive-compulsive disorder with excessive hand washing (Figure 1). Maternal family history disclosed a three-generational antecedent of childhood and adult-onset X-linked adrenoleukodystrophy (X-ALD). Examination showed global brisk tendon reflexes and bilateral Babinski sign. Neuroimaging studies showed periventricular white matter lesions (Figure 2). Plasma very-long chain fatty acid

concentrations were suggestive of X-ALD. Genetic analysis revealed a definite pathogenic variant (c.311G > A; p.Arg104His) in the *ABCD1* gene, confirming the diagnosis of adult-onset cerebral X-ALD.

Personality changes, psychotic symptoms and obsessive-compulsive disorder may be the presenting features of adult-onset cerebral X-ALD^{1,2,3}, preceding the onset of motor compromise^{1,2,4}.

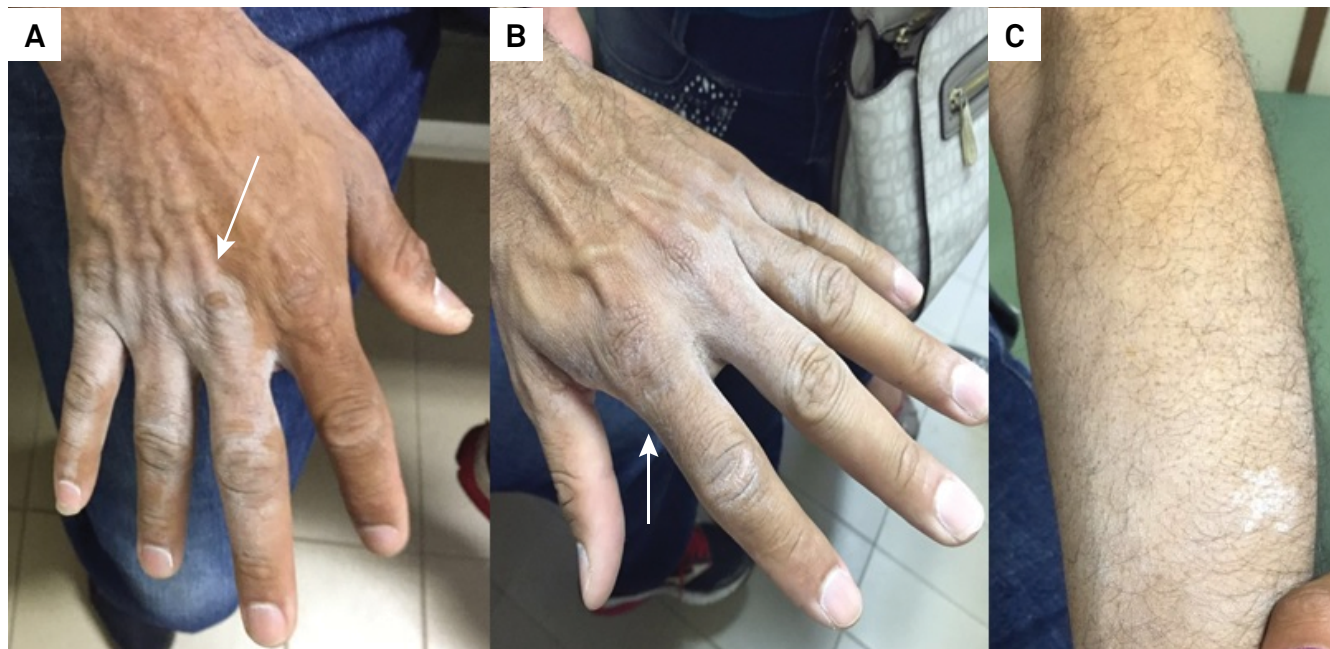






Figure 1. Examination findings in obsessive-compulsive behavior in adult-onset cerebral X-linked adrenoleukodystrophy. (A-C) Ectoscopic evaluation revealed the presence of excoriations and superficial traumatic exfoliations of the epidermis in the extensor surface of hands and forearms (white arrows).

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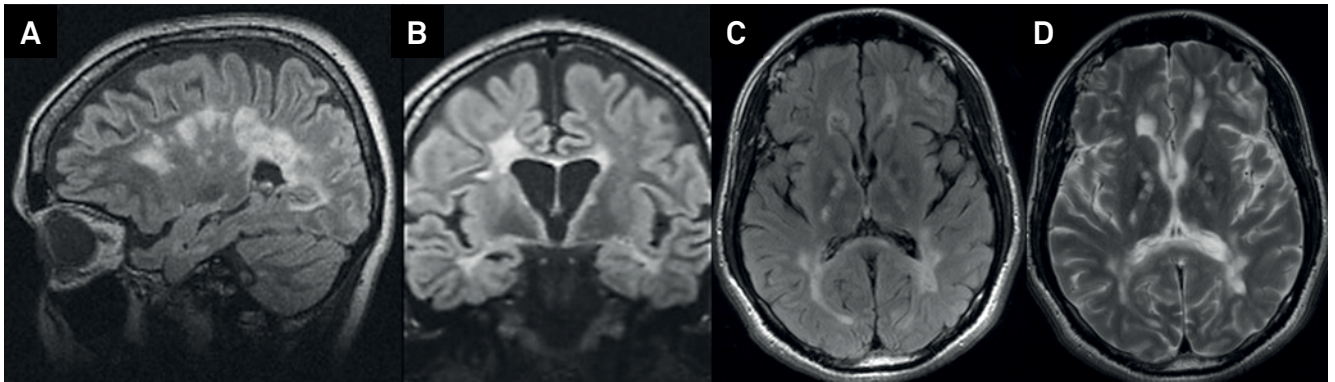


Figure 2. Neuroimaging studies in adult-onset cerebral X-linked adrenoleukodystrophy. (A) Sagittal and (B) coronal brain MR imaging showing confluent asymmetric periventricular hyperintense lesions in FLAIR sequence. Axial brain MR imaging also revealed the involvement of the corpus callosum and internal capsule in FLAIR (C) and T2-weighted images (D).

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