Adult-Onset Adrenoleukodystrophy Presenting With Atypical Location of White Matter Lesions

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49-year-old man, presented with progressive limb stiffness, slowness, cognitive impairment, and urinary disturbance over two years. Examination revealed skin hyperpigmentation, apathy, attention deficit, spastic dysarthria, symmetric upper limb rigidity, lower limb spasticity and, hyperreflexia. MRI brain is shown in figure 1. Serum cortisol levels confirmed adrenal insufficiency. Plasma very-long-chain-fatty acids were elevated. A 5’splice-site pathogenic variant in ABCD1(C.900+2T>A) confirmed Adrenoleukodystrophy(ALD).

80% patients with childhood cerebral-ALD present with posterior white matter hyperintensities1. Adult ALD involves atypical areas like frontal white matter, corticospinal projection fibers, and rarely cerebellum, underscoring its importance in differentials of adult white matter diseases.1,2
References


Figure Title: MRI Brain with atypical location of white matter lesions in adult onset Adrenoleukodystrophy

Figure legend
Figure 1: “Axial fluid-attenuated inversion recovery (FLAIR) weighted (a, b) images show bilateral symmetrical hyperintense signal changes in both cerebellar white matter (short arrows in a), middle cerebellar peduncles (long arrows in a), frontal lobe white matter (short arrows in b & d), genu of corpus callosum, internal-external capsules (long arrows in b & d) and peri atrial white matter(dotted arrows in b). These lesions are iso-hypointense on T1-WI (c) with moderate enhancement in post-gad T1-WI (d).
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