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X-Linked ALD as an Etiological Cause of Progressive Spastic Paraplegia: A Case Report

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Abstract

X-linked adrenoleukodystrophy (X-ALD) is a rare peroxisomal disorder caused by mutations in ABCD1, thereby causing impairment of very long-chain fatty acid (VLCFA) β-oxidation. Its adult-onset form, namely, adrenomyeloneuropathy (AMN), often manifests as progressive spastic paraparesis that mimics hereditary spastic paraplegia (HSP). Brain and spinal magnetic resonance imaging (MRI) findings are often unremarkable, contributing to diagnostic delays and misdiagnosis. We report the case of a 60-year-old woman who presented with a 10-year history of progressive lower limb stiffness, weakness, and gait disturbance. She was initially diagnosed with HSP and sought a second opinion. Her family history was remarkable for her father, who experienced a chronic gait disturbance of unknown etiology but never received a specific diagnosis. Neurological examination revealed mild weakness and spasticity in the bilateral lower extremities and bladder dysfunction, but the MRI of the brain and spine was normal. Given the presence of a relevant family history and urinary symptoms, metabolic testing was performed. Elevated plasma VLCFA levels and a pathogenic variant in ABCD1 confirmed the diagnosis of X-ALD presenting as AMN. This case underscores the need to consider X-ALD in cases with progressive spastic paraparesis even in the absence of imaging abnormalities.