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"Hot cross bun" sign resembling multiple system atrophy in a patient with Machado-Joseph disease

"Sinal da cruz" mimetizando atrofia de múltiplos sistemas em um paciente com doença de Machado-Joseph

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A 59-year-old woman presented with a 9-year history of progressive gait instability. She also had rapid-eye movement (REM) sleep behavior disorder and urinary incontinency. Neurological examination revealed ataxia and nystagmus. Brain magnetic resonance imaging showed the "hot cross bun" sign in the pons, as well as cerebellar and brainstem atrophy. Multiple system atrophy (MSA) was suspected, but family history was remarkable for an autosomal inherited ataxia. Genetic testing was positive for Machado-Joseph disease (MJD), which is the most common autosomal dominant spinocerebellar ataxia.

MJD may present with REM sleep behavior disorder and dysautonomia, similar to MSA, but to a lesser extent[1–3]. The "hot cross bun" sign is rarely found in MJD patients (1.3%), but it is a frequent brain imaging feature in MSA[4]. In this case, a detailed familial investigation was crucial to guide the diagnosis, as MJD is an autosomal dominant inherited ataxia, and MSA is sporadic.

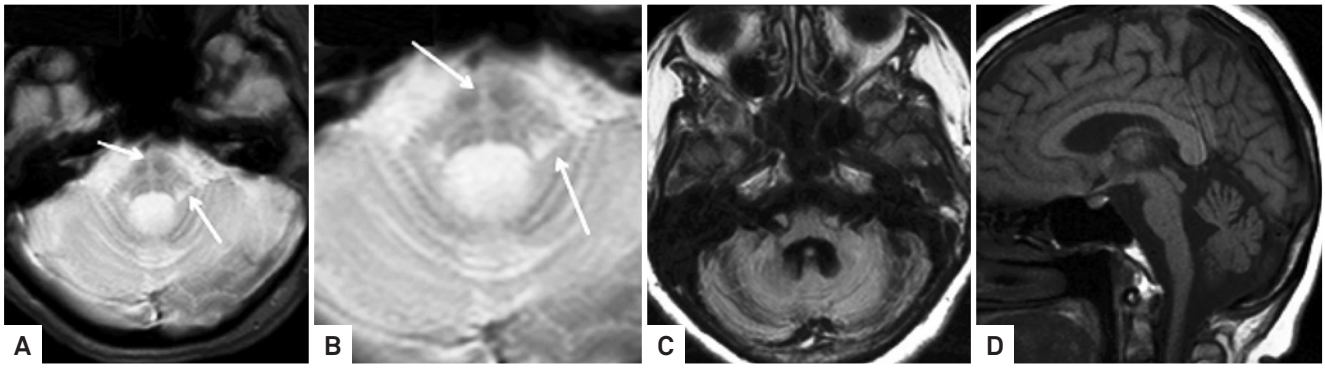

Figure. Axial T2* weighted brain MRI (A and B) showing the "hot cross bun" sign in the pons and cerebellar and brainstem atrophy. Wallerian degeneration was also observed, characterized by marked hyperintensity in the pontocerebellar fibers (arrow). Axial fluid-attenuated inversion recovery (FLAIR) (C) and sagittal T1 (D) reinforce olivopontocerebellar atrophy, but no hyperintensity was observed in the pontocerebellar fibers. Supratentorial images showed no atrophy or abnormal sign in the basal ganglia.

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