LEUKODYSTROPHY IN A MEXICAN WOMAN WITH SPINOCEREBELLAR ATAXIA TYPE 3

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Introduction: Machado-Joseph disease (MIM ID #109150), also known as spinocerebellar ataxia type 3 (SCA3) is a polyglutamine repeats disease with an autosomal dominant inheritance caused by the unstable (CAG)n trinucleotide repeat expansion in ataxin-3 gene (ATXN3; 607047) on chromosome 14q32.12 Clinically is characterized by ataxia, spasticity, and ocular movement abnormalities.

Objective: The aim of this report is to present the imaging findings in a patient affected by SCA3.

Case report: A woman aged 58 years-old with a familial history of ataxia was studied. The patient presented limb and truncal ataxia, dysmetria, disdiadochokinesis and gaze-evoked nystagmus.

Results: Molecular analysis revealed a number of expansion repeats of 21/65. Brain MRI scans revealed brain stem, midbrain, cortical, subcortical frontal lobe and cerebellar, atrophies; enlarged magna cistern; and bilateral leukoencephalopathy at the frontal lobe with periventricular white matter lesions.

Discussion: In previous studies used to characterize and compare the pattern of gray and white matter atrophy in patients with SCA1, 2 and 3 in India population they did not found significant areas of white matter atrophy in SCA3 patients compared to controls. The present case showed gray and white matter atrophy, but this is the first time that is described periventricular white matter lesions in a patient with SCA3.

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