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Mini-relato de caso

Neuroimaging in Kearns-Sayre syndrome

Neuroimagem na síndrome de Kearns-Sayre

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Kearns-Sayre Syndrome (KSS) is a multisystemic slowly progressive, and rare mitochondriopathy caused by mtDNA deletions. KSS manifests by a clinical triad: ophthalmoplegia, pigmentary retinopathy, and onset before the age of 20 years[1,2]. Neuroimaging shows cerebral and cerebellar atrophy; hyperintensities in basal ganglia, brainstem and cerebral and cerebellar white matter (WM); bilateral T2 hyperintensities in subcortical WM, thalami, brainstem and cerebellum[2]. KSS diagnosis is based on the clinical triad, but neuroimaging and striated muscle biopsy (SMB) might help[1,3].

The present case is about a 12-year-old female with ophthalmoplegia, bilateral eyelid ptosis and globally abolished tendon reflexes. On fundus examination, pigmentary retinopathy was reported. Axial T2-weighted image showed hypersignal in the pale globes and internal capsules (Figure A), coronal T2 presented impairment of subcortical WM (Figure B), midbrain's hyperintensities (Figure C), and T2-weighted signal hyperintensities in cerebellar hemispheres (Figure D).

With clinical diagnosis already made, patient underwent a histopathological study whose results were fibres of varying size, subsarcolemmal and intermyofibrillar accumulations of anomalous mitochondria and glycogen, myofibrillar degeneration, and presence of ragged red fibers, which is characteristic of KSS[2,3]. Subsequently, the genetic study of SMB showed smaller fragment of mtDNA, suggestive of deletion of mitochondrial genetic material, which is suggestive of KSS[2,3].

Keywords: mitochondrial encephalomyopathies, mitochondrial myopathies, Kearns-Sayre syndrome

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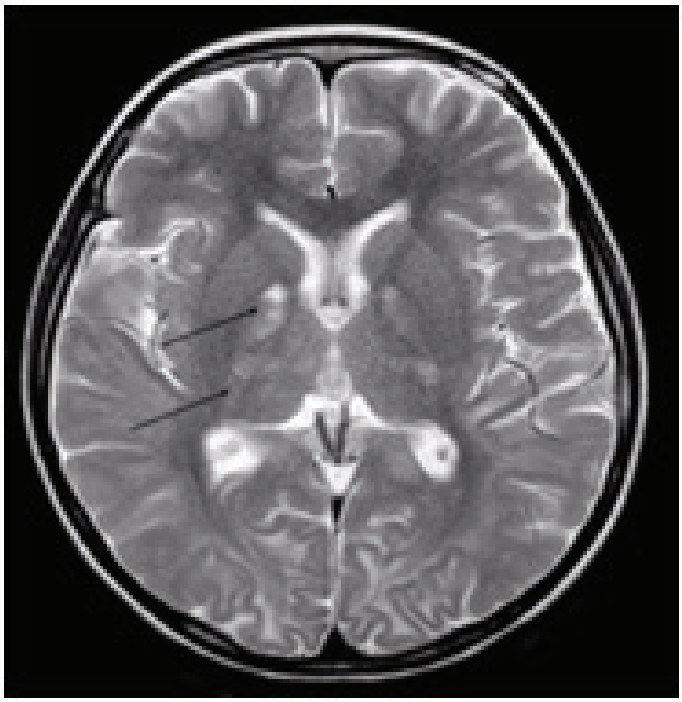

Figure A. Axial T2-weighted image demonstrating hyperintensities in the pale globes and internal capsules.

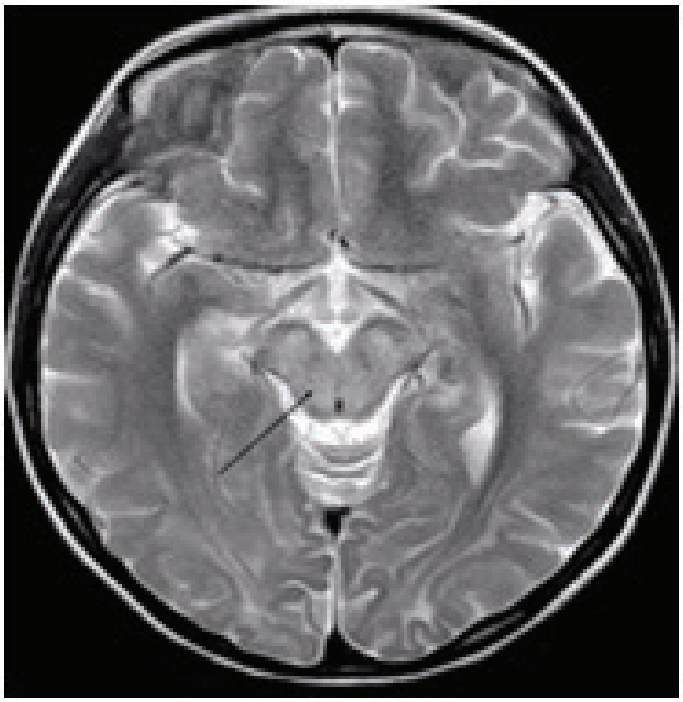

Figure C. Axial T2-weighted image showing impairment of the midbrain.

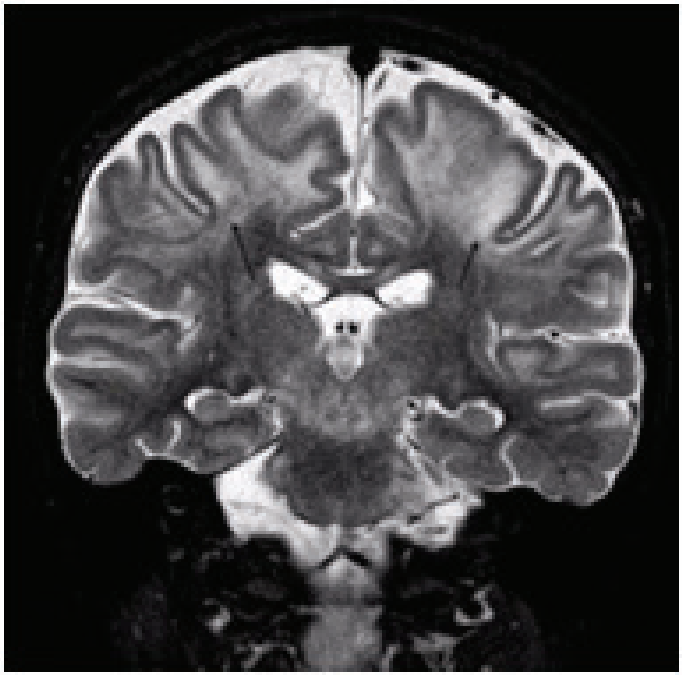

Figure B. Coronal T2-weighted image demonstrating impairment of the subcortical white matter.

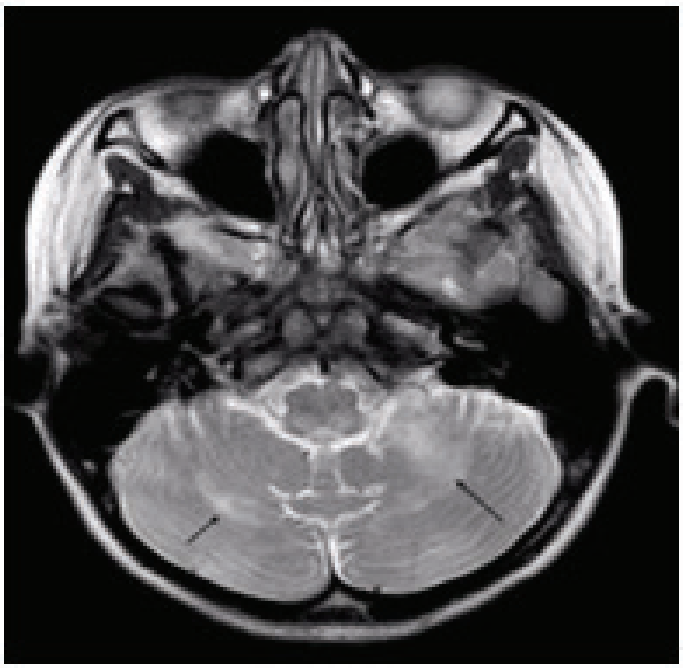

Figure D. Axial T2-weighted image with hyperintensities in the cerebellar hemispheres.

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