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Leukodystrophy with premature ovarian failure: think on vanishing white matter disease (VWMD)

Leucodistrofia com falência ovariana prematura: pense na doença da substância branca evanescente

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A 46-year-old female presented progressive hand tremor at age of 8, associated to cognitive and motor deterioration. She developed incapacitating head tremor and was wheelchair-bound 12 years after onset. Premature ovarian failure (POF) occurred at 27-year-old. In the last 3 years, head tremor became less intense, but she remained with horizontal bidireccional nistagmus.

Vanishing white matter disease (VWMD) is an autosomal recessive disorder characterized by cerebellar ataxia, spasticity, and cognitive impairment[1,2]. Brain MRI discloses symmetric and diffuse white matter lesions (figure). VWMD manifests from infancy to adulthood; in female, POF may occur[3]. It is caused by mutations in both alleles of one of five genes coding for subunits of eukaryotic translation initiation factor 2B.

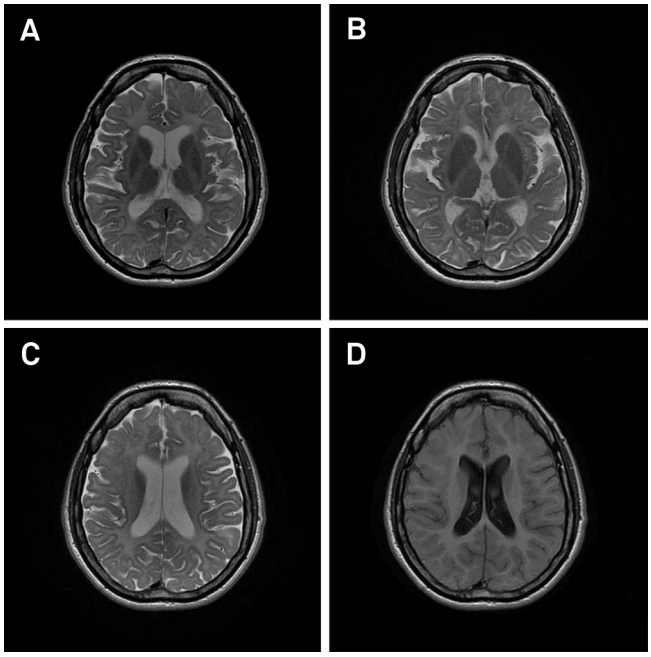

Figure. (A, B, and C) T2-weighted acquisition showing diffuse abnormal white matter signal, with intensity close to of the cerebrospinal fluid; and (D) Fluid-attenuated inversion recovery (FLAIR), showing diffuse hyperintensity in white matter.

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