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. Brain MRI discloses symmetric and diffuse white matter lesions (figure). VWMD manifests from infancy to adulthood; in female, POF may occur. It is caused by mutations in both alleles of one of five genes coding for subunits of eukaryotic translation initiation factor 2B.

References