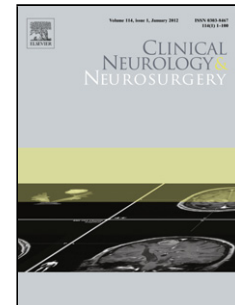


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Epilepsy and ovarian failure: Two cases of adolescent-onset ovarioleukodystrophy.

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HIGHLIGHTS

- 1- Vanishing white matter disease (VWM) is a leukodystrophy caused by mutations in eIF2B.
- 2- Association of neurological decline, ovarian failure and cystic changes in white matter suggests an ovarioleukodystrophy.
- 3- Adolescent-onset ovarioleukodystrophy is extremely rare.
- 4- Vanishing white matter disease may start as epilepsy without other neurological symptoms for years.

Abstract: Vanishing white matter disease (VWM) was described by Van der Knaap in 1996. This association with premature ovarian failure is known as ovarioleukodystrophy. This is a rare entity caused by a mutation in one of the subunits of eukaryotic initiation factor 2B (EIF2B). The onset in adulthood or late in adolescence is very infrequent. A 41-years-old woman and her 37-years-old sister developed epilepsy in association with premature ovarian failure at the age of 13 and 18 respectively. The oldest-one started 17 years later progressive subcortical cognitive decline with predominant behavioural disorders and a progressive spastic paraparesis in association with symmetric cystic changes in the white matter of both hemispheres. In both patients we found the c.1117C>T (p.Arg373Cys) mutation in homozygosis in the EIF2B4 gen.

KEYWORDS: Vanishing white matter disease, Ovarioleukodystrophy, Epilepsy, Ovarian failure.

INTRODUCTION

Vanishing white matter disease (VWM) was first described by Van der Knaap in 1996 after observing cystic degeneration of cerebral white matter in the necropsy of nine patients [1]. VWM is an autosomal recessive disease caused by a mutation in one of the five genes (EIF2B1-EIF2B5) that encode the eukaryotic translation initiation factor 2B (eIF2B) [2]. It is one of the most common leukodystrophies and classically presents in childhood. It was reported that clinical criteria for VWM were met by 40 % of patients with leukodystrophy of unknown

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