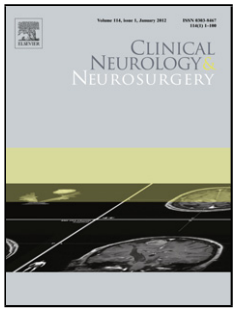

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Authors: Herrera-García José David, Guillen-Martínez Virginia, Creus-Fernández Carlota, Mínguez-Castellanos Adolfo, Carnero Pardo Cristóbal

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

**Authors:** Herrera-García, José David[1]. Guillen-Martínez, Virginia[1]. Creus-Fernández, Carlota[1]. Mínguez-Castellanos, Adolfo[1]. Carnero Pardo, Cristóbal[1].

[1]Neurology Unit. Hospital Universitario Virgen de las Nieves. Granada. Spain.

## 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 with 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