P-137 - L-2 HYDROXYGLUTARIC ACIDURIA. A CASE CONFIRMED AT 43 YEARS OF AGE.

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**INTRODUCTION:** L-2-Hydroxyglutaric aciduria (L2HGA) is a rare neurometabolic disorder with an autosomal recessive mode of inheritance. It is characterized by elevated levels of L-2-hydroxyglutarate in biological fluids. It was first reported in 1980 in a Moroccan boy with psychomotor retardation. Patients have neurologic manifestations including mild-to-moderate psychomotor retardation, cerebellar ataxia, variable macrocephaly, and epilepsy. Has a slowly progressive disease course. Most patients reach adulthood. A consistent pattern of brain magnetic resonance (MR) with signal intensity abnormalities of the subcortical cerebral white matter (WM), putamen, and dentate nucleus had been described. We describe a case with symptoms and WM abnormalities since childhood that urine organic acid performed at 43 years of age, showed high 2-Hydroxyglutaric acid and molecular study confirmed pathogenic mutation in L2HGA gene. There is no specific treatment. **OBJECTIVE:** to describe clinical course and MR findings of a case confirmed at adulthood. **CASE:** female product of first pregnancy of consanguineous couple. Normal pregnancy and delivery. Development delay, mild mental retardation. She was evaluated for the first time when she was 27 years because she had cerebellar symptoms and gait ataxia since she was 4 years. Bilaterally symmetrical subcortical WM abnormalities were present since the first MR (29 years). 4 years latter involvement of both dentate nuclei was evident. There have been no clinical progression. Ammonia, lactic acid, amino acids were normal. At 43 years of age, qualitative urine organic acid profile showed high level of 2 hydroxyglutaric acid. At this age, epilepsy started. Because the clinical picture and MRI findings, sequencing of the L2HGA gene was performed. It was find a known pathogenic variant: homozygosis c.632G>T p.Gly211Val. **CONCLUSION:** L2HGA is a rare slow progressive neurometabolic disease inherited as an autosomal recessive trait. Characteristic brain MR findings helps for the diagnosis. It is important to be aware about this disease in order to perform urine organic acids earlier during childhood to give adequate genetic counselling to the family.