P-075 - NEURONAL CEROID-LIPOFUSCINOSES TYPE 8 (CLN8), A CASE REPORT

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INTRODUCTION: Neuronal ceroid-lipofuscinoses (NCLs) type 8 is a rare neurodegenerative disease characterized by motors and cognitive deterioration, seizures, visual loss, retinophaty, cerebellar and cerebral atrophy. The symptoms start between the ages of 4 and 8 years. It is an autosomal recessive lysosomal disorder caused by the mutations in the CLN8 gene.

OBJECTIVE: We report a female patient with a rare metabolic disease: CLN8. We discuss clinical and molecular findings.

METHODS: retrospective medical record review of a patient diagnosed with CLN8.

RESULTS: We report a 19 year old patient that initiated at the age of 6 with epilepsy and learning disability. At the age of 10 the epilepsy turned refractory to multiple antiepileptic drugs and developed progressive ataxia. At 17 years old, due to visual loss, she was diagnosed with bilateral macular degeneration. Brain MRI showed pronounced cerebellar atrophy. With this clinical phenotype, NCL was suspected. Using targeted next-generation sequencing, a variant in CLN8 was detected in this patient.

DISCUSSION: NCLs are the leading cause of dementia in children and adolescents. It presents with a varying age of onset, symptoms and disease course. Historically recognized by a debilitating set of symptoms: seizure/epilepsy, dementia and vision loss. CLN8 is one of them. We think it is very important to be aware of this disease to ensure early diagnosis, optimal patient care and family support.