P-097 - OUTCOMES OF A CLN2 PATIENT TREATED EARLY WITH CEREBROVENTRICULAR ENZYME REPLACEMENT THERAPY

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INTRODUCTION: Neuronal CeroideLipofuscinosis(CLN)is a rare progressive disorder caused by a deficiency of tripeptidyl peptidasa 1(TPP1). CLN2 most commonly presents with seizures and/or ataxia in the late-infantile period (age 2-4), often in combination whit a history of language delay, followed by progressive childhood dementia, motor and visual deterioration, and early death. Recombinant human tripeptidyl peptidase1has been developed to treat CLN2.***AIM:*** to describe the outcomes of a CLN2 patient who received early treatmentwith cerebroventricular ERT. ***MATERIALS AND METHODS:*** we analyzed the CLN2 Clinical Rating Scale electroencephalographic (EEG) characteristics and MRIof a patient with CLN2 after 20 months of treatment with cerebroventricular ERT. ***RESULTS:*** a 4-year-old male wasdiagnosed after his affected16yo brother with classic phenotype. He started treatment at 2 years of agepresenting at the time of diagnosis, language delay, behavioral disorders and normal examination.MRI and EEG with photic stimulation were normal. After 20 months of treatment his neurological exam, the MRI and EEG with photic stimulation were still normal.. Language delay remained stable. He didn´t present complication regarding therapy. ***CONCLUSIONS:*** CLN2 natural history studies estimates a loss of 2 points per year in the Clinical Rating Scale. Our patient remained stable after 20 months of ERT so we conclude that ERT prevented the onset of CLN2 symptoms in him so far.