P-092 - TREATMENT OF LATE- INFANTILE NEURONAL CEROID LIPOFUSCINOSIS TYPE 2 DISEASE (CLN2) WITH ENZYME - REPLACEMENT THERAPY WITH INTRAVENTRICULAR INFUSION OF CERLIPONASE ALFA: REPORT OF TWO CASES

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BACKGROUND: Late- infantile Neuronal Ceroid Lipofuscinosis type 2 disease (CLN2), is a progressive disease which typically has beginning during the stages of infant or preschool, with a progressive deterioration in mental, motor, vision, language skills and with epilepsy usually difficult to manage. The frequency depending on the ethnic origin, reaching frequencies of 1 / 385,000 inhabitants in Finland, up to 1 / 1,000,000 inhabitants in other Scandinavian countries. In Colombia there is no record that allows the estimation of the frequency of this disease. Typically the age of onset varies between 2 and 7 years. It is transmitted in an autosomal recessive way, by mutations of the TPP1.1-4 gene). Until April 2017, no treatment was available that potentially would change the natural course of disease. GOALS: Describe two cases of late infantile Neuronal Ceroid Lipofuscinosis type 2 (CLN2), currently in treatment with enzyme-replacement therapy with intraventricular infusion of Cerliponase alfa. RESULTS: We report two cases which are patients of 5 years of age, male and female, with myoclonic epilepsies, with a reflex component to sounds and low-frequency photostimulation, associated with progressive ataxia and regression of motor and language skills. Symptoms about 2 years of evolution, with neuroimaging that show cerebral and cerebellar atrophy; the diagnosis of Neuronal Ceroid Lipofuscinosis type 2 (CLN2) was confirmed by enzymatic and genetic method, with mutation confirmed in the TPP1 gene. The treatment of enzyme- replacement with Cerliponasa α was started, it was applied intraventricular every 2 weeks, through the Ommaya reservoir. When the treatment was started, the patients were in a phase of progressive skills regression, and after the beginning the stabilization of the disease was found. We have not found new clinical deterioration until now. No significant gains in neurodevelopment have been documented, although subjectively parents have reported improvements in some skills CONCLUSIONS: Treatment intraventricular with Cerliponase α every 2 weeks is effective in controlling and stabilizing the disease. In our patients we have not found deterioration from the basal point which they started the therapy.