P-177 - SINGLE CASE REPORT: ABNORMAL MOVEMENT DISORDER AND CEREBRAL MRI CORRELATION IN NEONATAL CITRULLINEMIA TYPE I

Garcia J, Bernal AC, Eiroa HD

JUAN P. GARRAHAN. CIUDAD AUTÓNOMA DE BS. AS. ARGENTINA

INTRODUCTION: Citrullinemia type I is an autosomal recessive disorder caused by argininosuccinate synthetase deficiency (ASS1D). ASS1D is the second most common urea cycle disorder (UCD) and it has a variable clinical phenotype. Although movement disorders are described in UCD, with some exceptions (arginase deficiency), these are uncommon. OBJECTIVES: We describe the clinical case of a patient with citrullinemia diagnosed at birth [compound heterozygote: c.970G>A(Missense)/c.601C>T(Nonsense)], with a history of a deceased brother in the neonatal period. There was a correlation between the clinical evolution and the cerebral MRI findings. MATERIALS AND METHODS: Review of the clinical records, including laboratory results, amino acid profile and cerebral MRI findings, at diagnosis and during follow up, from birth to 6 years of age. RESULTS: Since he was approximately 5-6 months of age, the patient evolved with a severe generalized dyskinetic-dystonic movement disorder of insidious onset. Nevertheless, he achieved an independent gait. Borderline cognitive profile with language skills mostly affected (IQ: 74). Brain MRI performed at 6 years of age showed bilateral and symmetric hyperintense signals in T2 and FLAIR in the globus pallidus. No significant metabolite disturbance observed in spectroscopy. While diffuse posterior white matter involvement was observed in the first MRI (1 month), there was no evidence of basal ganglia injury. First ammonia control was 272 uMol/L (16h of life). Adequate response to initial management. During the first 50 days, ammonia did not exceed 150 uMol/L. Prior to the second brain MRI, he had been hospitalized 25 times (4 per year). In 14/25 (56%), ammonia was higher than 100 uMol/L(Max. 333 uMol/L). Only twice ammonia exceeded 250 uMol/L. Response to emergency treatment was optimal in all cases. Ambulatory controls: Mean ammonia 68.5 uMol/L; Mean glutamine: 731.2 uMol/L. Mean citrulline: 1767.8 uMol/L. Leucine slightly decreased during the first year. CONCLUSION: Despite the early beginning of treatment and apparently unrelated to acute episodes of decompensation or chronic hyperammonemia, the patient developed a severe movement disorder of insidious onset with an evident image correlation. A second disease as well as epigenetic factors could determine the patient evolution. More case reports are needed in order to arrive general conclusions.