P-141 - DIAGNOSTIC DILEMMA OF INBORN METABOLIC DISEASE WITH HYPERAMMONEMIA IN A COLOMBIAN BOY WITH SUSPECTED DISEASE ON INTERMEDIARY METABOLISM

Beltrán OI, Márquez-Peñaranda W, Cuellar MC, Ladino LY

(1) Grupo de Investigación GenHOMI-Hospital Pediátrico de la Misericordia Fundación HOMI. (2) Grupo de investigación BioGenEtica & BioDerecho-Universidad Militar Nueva Granada. Bogotá DC-Colombia. beltran.orietta.gen@gmail.com

INTRODUCTION: Hyperammonemia is a metabolic emergency, which can lead to acute and chronic complications like neurological damage and/or death. It can be caused by different pathologies but in the pediatric population inborn metabolic disease should always be suspected. OBJECTIVE: Describe clinical follow-up and biochemical data collected in a boy with suspected disease on intermediary metabolism. METHODS: Retrospective study of clinical history and biochemical analysis of a case from July 2009-October 2018 at the Hospital Pediátrico de la Misericordia Fundación HOMI (Bogotá, Colombia). RESULTS: A 12-year-old boy, the third of three male children of unrelated parents with Colombian origin. Gestation and caesarean delivery without any complications (birth weight of 3225g). He had normal psychomotor development during the first six months of life until the administration of vaccines, when he presented fever, lethargy, irritability, hypotonia and vomiting. Subsequently, he presented developmental regression and progressive spastic tetraparesis. At 16-mo EEG showed generalized paroxysmal changes and biochemical analysis showed mild hyperammonemia, respiratory alkalosis and high orotic acid in urine, compatible with partial OTC deficiency. Whereby, he was treated with hypoproteic diet and sodium benzoate. At 5-yo, health care provider ceases sodium benzoate due to shortage, but surprisingly ammonium levels did not increase. Treating physicians re-evaluated the clinical evolution and the possible triggers and decided to perform biochemical analysis (plasma amino acids, urine organic acid, blood gases and others) several times, but the results were consistently normal. He continued treatment with hypoproteic diet and rehabilitation. Nowadays, the patient has severe motor and language delay, as well as, spastic tetraparesis. Recent analysis of serum metabolomic profile showed a type II glutaric aciduria. CONCLUSIONS: We report the case of an adolescent boy with neurodevelopmental delay since the age of 6 months, as well as, vomiting, lethargy, seizures, spastic tetraparesis and episodic hyperammonemia. He received the treatment of an urea cycle disorder. However, a detailed clinical follow-up suggested an organic aciduria as a more compatible diagnosis, but urine organic acids were always negative. Complex and overlapping diseases require high-throughput technologies with better sensitivity to distinguish the type of metabolic disorders and provide a clear diagnosis.