P-168 - DELAY IN THE DIAGNOSIS OF INTERMEDIATE FORM OF MAPLE SYRUP URINE DISEASE AND ITS GLOBAL REPERCUSSIONS: A CASE REPORT

Santos ES, Andrade KESN, Barbosa AF, Duarte SRS, Garcia SZ, Mendes LAM, Santana GCS, Santos JS, Santos PAR, Silva SW

Federal University of Sergipe. Lagarto - SE - Brazil. vicani@uol.com.br

INTRODUCTION: Maple syrup urine disease (MSUD) is an inborn error of metabolism caused by defects in the branched-chain α-ketoacid dehydrogenase complex (BCKAD), which results in elevations of the branched-chain amino acids (BCAAs) in plasma, α-ketoacids in urine, and production of the pathognomonic disease marker, alloisoleucine. The disorder varies in severity and the clinical spectrum is quite broad with five recognized clinical variants that have no known association with genotype. The intermediate form of MSUD is characterized by up to 30% of BCKAD residual activity. These individuals may appear healthy during the neonatal period, although maple syrup odor in cerumen may be present. During the first years of life, they may experience feeding problems, poor growth, and intellectual disability, and are susceptible to similar neurologic features as individuals with the classic form. OBJECTIVE: To report a case of intermediate form of MSUD diagnosed only after two years of progressing symptoms.

METHODOLOGY: A retrospective study was conducted. Clinical data were collected through the patient’s medical record. RESULTS: A two-and-a-half-year-old male, born from a young, healthy and non-consanguineous couple started with progressive hypotonia, feeding problems and poor interaction at the age of 4 months. Developmental delay and stereotypical movements became even more evident during the second year of life. He still cannot walk and has severe language delay. After normal initial exams (Karyotype, Creatine Kinase-CK, skull CT scan, echocardiogram, etc) he was referred to Medical Genetics evaluation. Quantitative plasma amino acid analysis showed an increase of leucine, isoleucine and valine, confirming MSUD diagnosis. Genetic Counseling was then offered and dietary treatment was introduced with a mild positive response. CONCLUSIONS: Unfortunately newborn screening for MSUD is not available in Brazil’s public health system. Therefore, Brazilian pediatricians and neuropediatricians should be alert to MSUD possible diagnosis in young children with development delay.