P-124 - METHYLMALONIC ACIDEMIA’S CLINICAL EVOLUTION IN THE FIRST CHILDHOOD: DIFFERENCES BETWEEN EARLY AND LATE DIAGNOSIS.

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**INTRODUCTION:** Methylmalonic acidemia caused deficient cofactor of cyanocobalamin system or deficiency of methylmalonyl-CoA mutase enzyme, that can be unresponsive to vitamin B12 due to complete or partial deficiency. This disorder progress with crises of decompensation, precipitated by protein intake, with ketoacidosis, muscular hypotonia, irritability, and an important neuropsychomotor development delay, presented in neonatal period.

**OBJECTIVE:** Case report comparing clinical and neuropsychomotor development of two patients, one with late diagnosis and another with early diagnosis of Methylmalonic Acidemia.

**MATERIALS AND METHODS:** Review of medical records, interviews with patient’s parents and children’s physical examination, photographic record, and literary review with Medline / Pubmed and specialized literature.

**RESULTS:** CASE 1 - PFRS, female, DB: 06/11/2012, consanguineous parents, gestation and pregnancy without complications. Neonatal deaths in the family. 2 days: ketoacidosis, respiratory discomfort; At 3, 7, 9, 12, 15 months: uncontrollable vomiting, ketoacidosis, pancytopenia; 1 year 6 months: diagnosed. Methylmalonic acid and child = 3479.9 mmol/mol creatinine. Inception of adequate diet and supplementation. Neurological clinic in diagnosis: microcephaly, malnutrition, mixed tetraplegia, axial hypotonia (without cervical support), absence of social contact, swallowing disorder, irritability, sleep disorder. Evolution: periods of improvement and regression. Family did not follow proper dietary treatment. He presented several decompensations. Gastrostomy: 2 years 3 months. 3 years 3 months: weight / height adjusted for age. Social smile, adequate sleep, responding to stimuli, still with hypotonia and tetraplegia. 4 years 4 months: Death due to sepsis. CASE 2: ALSQ, female, DB: 05/08/13 3 days: Acidosis, sepsis. Presented two altered samples in the neonatal screening test. 20 days: Methylmalonic acid (VR <36 mmol/mol creatinine) and child = 140 mmol/mol creatinine. Done protein restriction. 35 days: Started diet and complementation. 2 years 11 months: started school, took personal snack and knew what she could eat. 4 years 2 months: Adequate neuropsychomotor development.

**CONCLUSIONS:** the reported cases reassert the importance of early diagnosis by demonstrating an enormous difference between the evolution of pathology in a child diagnosed early and
another with late diagnosis. It Reinforced the importance of investments in neonatal screening tests to avoid permanent damage in the neuropsychomotor development of children with metabolic errors.