P-025 - CLINICAL MANIFESTATIONS OF COBALAMIN DEFICIENCY (CD) IN CHILDREN

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Cobalamin is cofactor of methionine and nucleic acids synthesis. Human is dependent on dietary sources. Its deficiency causes central/peripheral nervous system compromise and megaloblastic anemia. The most frequent etiology of CD in children is nutritional. We report 5 children with CD, 4 infants (8-18 months), 1 newborn. Clinical picture in infants: 3 patients were suspected due to moderate to severe anemia. One patient presented with acute tremor of the extremities. Patients rejected solid food and were mainly breast feeding; they presented failure to thrive, mild to moderate psychomotor developmental delay (PMDD), axial hypotonia in two. Relevant tests: Hemoglobin 6.5-9.2 gr/dl, 2 patients with regenerative hemolytic anemia, 2 patients with macrocytic anemia. Very low cobalamin levels. The newborn was diagnosed at age of 12 days due to mild increment of propionylcarnitine in neonatal screening. CD causes: Four cases due to maternal CD. Two mothers presented subclinical pernicious anemia. Two mothers with CD of unknown cause. A patient was explained by a vegan diet due to parental option. Patients were treated with intramuscular hydroxycobalamin and have been followed for 6-20 months. Two present normal psychomotor development (including the newborn), 1 presents moderate PMDD, 2 present language delay. CONCLUSIONS: CD should be suspected in all infants with PMDD and/or hypotonia of non-evident cause. It can present with hemolytic anemia. The cause of maternal CD can be unknown. Despite the treatment, CD in infants can leave long-term neurological compromise. Neonatal diagnosis and early treatment of CD can avoid neurological and systemic symptoms.