P-142 - AN INFANTILE CASE OF METHYLMALONIC ACIDURIA WITH HYPEROUMOCYSTEINEMIA (CBLC). A DIFFICULTO TO DIAGNOSIS DISEASE WITHOUT NEONATAL SCREENING AND WITH FEW EVIDENCES ABOUT THE BEST TREATMENT.

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The remethylation defects are rare diseases, with often significant delayed diagnosis and with few standardized treatment protocols. The objective of the case presentation is to see the difficulties in the diagnosis and the follow up with hydroxocobalamin dose escalation. The boy is the first son of non-consanguineous parents, with a normal pregnancy and delivery. He was hospitalized at 13 days of life, because feeding difficulties, 14% of weight loss, perianal lesions and bad general condition, with bone marrow aplasia. The diagnosis was sepsis until 35 days of life when a broad neonatal screening was done because the neurological symptoms persisted in spite of antibiotic treatment with negative bacterial and virus exams. With C3 elevation the assessment was expanded, finding low plasma methionine, methylmalonic acid in urine and high serum homocysteine, then a remethylation defect was suspected. It was confirmed with the finding of two pathogenic variant in MMACHC gene (c271duoAand c.331C>T). With 3.4kg he began treatment with IM Hydroxocobalamin 1mg/d, betaine 800mg/d and L-carnitine 300mg/d with breast feeding. The serum methylmalonic acid (MMA) decreased from 6994 to 629 ug/ml (normal <104ug/ml) and the homocysteine from > 50 umol/l to 35. After five months with the same B12 dose and with 8 kg of weight (0.12 mg/kg/d ) the MMA increased to 900 ug/ml and the homocysteine to 40 umol/l then the B12 dose was increased to 0,2 mg/kg and the betaine to 150mg/kg. Then the serum MMA decreased again to 345ug/ml, the homocysteine to 27 umol/l and the metionine was normal. He has had normal growth and development without retinal lesions but with persistent nystagmus that was present since1 month old. In conclusion it is difficult for pediatricians to think about the disease in spite of the severe clinical aspect. Patients like this could die in the first trimester of life no diagnostic. The increment of B12 doses seems again to be necessary. Longer follow up would tell us about neurological and ocular development.