P-122 - BIOCHEMICAL DIAGNOSIS OF METHYLMALONIC ACIDURIA IN CUBA DURING 2014 TO 2018.

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INTRODUCTION: Methylmalonic aciduria (MA) is the most frequent organic aciduria (OA) in Cuba. It is a group of genetic diseases characterized by the excretion of high levels of methylmalonic acid (MMA) in the urine. The excretion of this metabolite may or may not be accompanied by elevated levels of homocysteine (Hcy) depending on the affected metabolic pathway.

OBJECTIVE: The aim of this work is to describe a laboratory methodology that combines the analysis of MMA and Hcy in the differential diagnosis and follow-up of the MA.

MATERIALS AND METHODS: Organic profile of 731 children with clinical suspicion of OA were analyzed by Gas Chromatography/Mass Spectrometry, in a period from 2014 to 2018. Hcy was quantified in plasma, in patients with high levels of MMA. The quantification of Hcy was performed by High Resolution Liquid Chromatography.

RESULTS: Seven patients with clinical features and high levels of MMA were diagnosed with MA. In them, we quantified plasmatic Hcy levels for realized the differential diagnosis. The concentration of Hcy in four patients was higher than the reference values, suggesting an MA combined with homocystinuria. From them, three patients once initiated the treatment with vitamin B12 and L-carnitine showed a decrease in the levels of both metabolites, corresponding to a satisfactory evolution. The remaining patients with high MMA levels had normal levels of Hcy, suggesting an isolated MA.

CONCLUSIONS: The determination of both markers permitted the differential diagnosis and biochemical monitoring of these diseases. The analysis of these two markers will allow us to infer the possible defect variant.