P-164 - BIOCHEMICAL AND CLINICAL DIAGNOSE OF AMINOACIDOPATHIES AND ORGANIC ACIDURIA IN CUBA

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INTRODUCTION: The Inborn Errors of Metabolism (IEM) are biochemical disorders caused by gene mutations and several consequences in metabolic pathways. The aminoacidopathies and organic acidurias are two genetic diseases with severe clinical features and several affected systems. In Cuba, only hyperphenylalaninemas are include in the neonatal screening program. So, the biochemical diagnosis of other IEM is performed after the appearance of suggestive clinical manifestations. OBJECTIVES: The aim of this work is to perform the biochemical diagnosis of amino acid and/or organic acidurias in patients with clinical suspicion and to determine the frequency of risk in the population studied. MATERIALS AND METHODS: A selective investigation was realized in Cuban patients under 12 years of age with clinical manifestations suggestive of an IEM during the period 2014-2018. We analyzed samples of serum and urine from 107 patients and 23 cerebrospinal fluid (CSF). The urine samples were subjected to qualitative chemical tests, thin-layer chromatography for amino acids and the profile of organic acids by GC/MS. The samples of serum and CSF were evaluated by thin-layer chromatography for amino acids and the quantification of the amino acid profile by HPLC. RESULTS: The suggestive biochemical diagnosis was made in 22 patients: 3 patients with maple syrup urine disease (MSUD), 7 patients with non-ketotic hyperglycinemia (NKH), 4 patients with hyperglycinemia secondary to an organic aciduria (1 isolated methylmalonic aciduria, 1 propionic aciduria, 1 pyruvate dehydrogenase deficiency and 1 dihydrolipoyl dehydrogenase deficiency), 3 patients with hyperglycinemia secondary to a fatty acid oxidation, 1 patient with ornithine aminotransferase deficiency (OAT), 1 patient with tyrosinemia type I, 2 patients with deficiency of methionine metabolism and 1 deficiency of methionine metabolism secondary to congenital disorders glycosylation (CDG). The risk frequency of these diseases was 20.6 % in Cuban studied population. CONCLUSIONS: During the period evaluated, the biochemical diagnosis of aminoacidopathies and organic aciduria was performed in Cuban patients; and the frequency of risk was estimated.