P-138 - URINARY ORGANIC ACID ANALYSIS EXPERIENCE IN DETECTING ORGANIC ACIDURIAS AND OTHER INBORN ERRORS OF METABOLISM IN BRAZIL OVER A TEN-YEAR PERIOD

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INTRODUCTION: Gas chromatography/ mass spectrometry (GC/MS) of urine samples is the method of choice in the diagnosis and follow-up of Organic acidurias/ acidemias. However, due to the high costs of equipment purchase, operation and maintenance, there are few reports on the detection of these IEM in Brazil. OBJECTIVE: To describe the organic acidurias/ acidemias, aminoacidopathies, Urea Cycle disorders, and fatty acid beta-oxidation defects detected by the analysis of urinary organic acids in a Brazilian private IEM reference laboratory. METHODOLOGY: A cross-sectional study was performed. Urine samples were obtained over a ten-year period from patients with a clinical suspicion of IEM. Neurological dysfunctions, neuroimaging abnormalities, liver disorders, metabolic acidosis, hypoglycemia, hyperlacticemia, ketonuria, and hyperammonemia were the main causes for the request of this analysis. These samples were maintained at -20°C until analysis. The analytes of interest were extracted with ethyl acetate/ethyl ether following acidification of the urine and addition of internal standard. Sample extracts were thoroughly dried before derivatization with a 100: 1 MSTFA/TMS-Cl reagent mixture and identified as trimethylsilyl compounds on a gas chromatograph/ mass spectrometer. RESULTS: Of the 8,873 urine specimens analyzed, 610 (7%) presented an IEM specific profile; other 929 samples (10%) presented nonspecific profiles. The majority of the analyzed samples, 6,838 (77%), were negative, while 496 (6%) presented undetermined results that would have to be characterized by other methodologies. Characteristic profiles of 33 different IEM were revealed from 262 patients. Organic acidemias/acidurias, aminoacidopathies and fatty acid beta-oxidation disorders were predominant. Methylmalonic acidemia, glutaric aciduria type 1, undefined Urea Cycle disorders, maple syrup urine disease and propionic acidemia were the most frequent IEM. CONCLUSIONS: The analysis of urinary organic acids, especially when combined with the analysis of acylcarnitines in dried filter paper blood spots, is very useful in the diagnostic evaluation of patients with a clinical suspicion of an IEM. This analysis may be included in the diagnostic workup of neonates with severe symptoms due to acute and early forms of IEM, often confounded with sepsis, as well as of patients with insidious or late-onset forms, frequently with a predominance of neurological symptoms - “neurological organic acidosis“.