P-132 - IDENTIFICATION OF ERYTHRO 2,3-DIHYDROXY-2-METHYLBUTIRATE IN URINE

Uicich RE, Moix CF, Cases GG, Gimenez MI

Laboratorio de Espectrometría de Masa. Hospital Italiano de Buenos Aires, Argentina. raul.uicich@hospitalitaliano.org.ar

INTRODUCTION: deficiencies of 3-hydroxyisobutyryl-CoA-hydrolase (HIBCH) and short-chain enoyl-CoA-hydrazetase (ECHS1) are characterized by the urinary excretion of 2,3-dihydroxy-2-methylbutyrate (23DH2MB). This metabolite can be identified by the routine method of GC-MS. It can be present in small amounts in normal samples and has been suggested as an identifier for these pathologies. However, the pure metabolite standard is not easy to obtain, therefore it is difficult to differentiate normal samples from pathological ones. 23DH2MB have two isomers, erythro and threo, both of which are present in the urine of normal patients. In some samples analyzed in our laboratory, the chromatograms show peaks of 23DH2MB with heights similar to those published and/or presented in congresses, but it is the ratio of areas between the erythro and threo metabolites that must be taken into account in order to associate it with any pathology. We do not have the metabolite standard. OBJECTIVE: to present a practical and simple example for the identification of the peak of erythro-23DH2MB in samples of urinary organic acids, by comparing the retention time of the same with the one that appears in other pathologies such as propionic and methylmalonic acidurias where it is elevated with respect to the threo metabolite. METHOD: gas chromatography with mass spectrometry detection, and derivatization of the urine with BSTFA + TMCS, previous extraction with two organic solvents. RESULTS: chromatograms of overlapping suspected samples are presented with chromatograms of samples with methylmalonic aciduria. In this pathology, as in propionic acidemia, the erythro isomer is elevated in a ratio greater than 10:1 with respect to the threo isomer (slightly displaced). Therefore both isomers can be differentiated and correlated with pathologies by comparison of the chromatograms. CONCLUSIONS: in the absence of a 23DH2MB standard, the superposition of chromatograms of suspected samples with samples of methylmalonic or propionic aciduria as well as the isomer ratio allows us to rule out HIBCH and ECHS1 diseases.