P-161 - URINARY ORGANIC ACIDS CHROMATOGRAPHY ANALYSIS ON FILTER-PAPER IN PKU: AN ALTERNATIVE APPROACH IN THERAPEUTIC MONITORING OF SAPROPTERIN DIHYDROCHLORIDE RESPONSIVENESS?

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BACKGROUND: Sapropterin dihydrochloride is the synthetic form of tetrahydrobiopterin, the cofactor for the phenylalanine hydroxylase enzyme. Approximately 50% of patients with phenylalanine hydroxylase deficiency, also referred to as phenylketonuria (PKU), are responsive to sapropterin as evidenced by a clinically significant decrease in blood phenylalanine levels and/or an increase in dietary phenylalanine (PHE) tolerance. OBJECTIVE: To present a practical approach in therapeutic monitoring of a PKU patient being treated with sapropterin dihydrochloride.

MATERIALS AND METHODS: Retrospective biochemical data review of phenylalanine levels measured in blood and urinary organic acids chromatography performed on filter-paper and total urine (pre and post-trial of sapropterin dihydrochloride) in a Brazilian PKU patient (also affected by type 1 diabetes mellitus).

RESULTS: Markedly reduction of phenylpyruvic acid, phenylacetic acid and phenyllactate in urine were seen after 24-hours after sapropterin dihydrochloride introduction (20mg/kg/day) and remained reduced after one month of therapy. There was correlation between PHE levels decrease and better organic acids profile, however PHE blood levels showed greater variation in different measurements in comparison to phenylpyruvic and phenylacetic acids levels that remained stable after reduction. Ketonuria was also observed after one month therapy, possibly reflecting poor diabetes mellitus control.

CONCLUSIONS: Although PHE levels and the phenylalanine/tyrosine ratio in blood have been the gold standard for diagnosis and follow-up of PKU patients, urinary organic acids gas chromatography/mass spectrometry (GC/MS)-based analysis may be a potential adjunctive tool to assess sapropterin dihydrochloride-responsiveness on a less invasive way, showing other biomarkers to further delineate biochemical findings in both treated and non-treated PKU patients.