

THE CLINICAL VALUE OF URINE ACYLCARNITINES.

¹Scheepers, J.C., ²Pretorius, P.J., ³Erasmus, E., ³Knoll, D.P. and ⁴Mienie, L.J.

¹Metabolic Unit, Biochemistry Division, School for Chemistry and biochemistry, North West University.

Acylcarnitine (AC) profiling in dried blood spots by means of MRM scans with electrospray ionisation tandem mass spectrometry (ESI-MS/MS) has proven to be a useful method in diagnosis of inborn errors of mitochondrial fatty acid oxidation. The short analysis time, use of filter paper for sending the material, possibilities for automatization and small volumes required, makes this method particularly suitable for neonatal screening programs. During the last decade ESI-MS/MS was developed as the heart of neonatal screening programs. Although less often reported ESI-MS/MS is becoming increasingly applied in metabolic diagnostic programs where urine is commonly used as specimen of interest. Urinary acylcarnitine excretion is affected by several external factors like diet, medication and other metabolic and non-metabolic diseases and several unknown metabolites are often present on the profile. In the last three years we have done thousands of urinary acylcarnitine analyses and a method for the identification of unknown metabolites, using different derivatization procedures, have been developed.

We have identified several of the unknown metabolites present in urine. Most of the metabolites identified, have diagnostic values. These metabolites included FIGLU, methyllysine, pyroglutamic acid and intermediates of the carnitine biosynthesis pathway. The excretion of intermediates of the carnitine biosynthesis pathway indicated that 2-hydroxyglutaric aciduria, a metabolic disease due to a defect of a yet unknown metabolic pathway, could be due to defective carnitine biosynthesis. All the results of these investigations will be presented.

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