Urinary acylcarnitine\enoylcarnitine ratio provides a sensitive method for the detection of acyl-CoA dehydrogenase deficiencies

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Acylcarnitine profiling in dried blood spots with ESI-MS-MS has proven to be a useful method in diagnosis of mitochondrial fatty acid oxidation defects. During the last 2 years ESI-MS-MS has become increasingly applied for the detection of fatty acid defects in urine. Traditionally GC-MS were used for diagnostic purposes but the sensitivity of these analytical methods, is considerably limited in detecting fatty acid disorders, especially during remission. Contrary to organic acids, the excretion of ACs is nearly unaffected by the patients clinical condition but urinary carnitine and AC concentration are more often affected by other factors like medication, tubular immaturity and other metabolic disorders and therefore difficult to interpret The ratios of different acylcarnitine species (short medium and long chain) are often used for a more sensitive detection of abnormalities. We have studied the possibility to use the urinary acylcarnitine\enoylcarnitine ratios according to the normal β-oxidation of fatty acids for the identification of acyl-CoA-dehydrogenase deficiencies. We discovered that this approach is not only much more sensitive, and reliable than calculating the real concentrations, but also showed the potential of detecting currently unknown metabolic diseases. The use of expensive isotopes, used as internal standards, are not necessary and we found that other metabolic diseases as well as medication seldom have an affect on the ratios.

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2. James J. Pitt, Mary Eggington, and Stephen G. Kahler Comprehensive Screening of Urine Samples for Inborn Errors of Metabolism by Electrospray Tandem Mass Spectrometry Clinical Chemistry 48:11 (2002) 19701980