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Determination of pterines as markers of hyperphenylalaninemias

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In humans, genetic defects of the synthesis or regeneration of tetrahydrobiopteridin (BH4), an essential cofactor in hydroxylation reactions, are associated with severe neurological disorders. The diagnosis of these conditions relies on the determination of BH4, and its metabolites and precursors in biological fluids. This paper describes a reversed-phase high-performance liquid chromatographic (HPLC), new method with fluorometric detection (ERRECI, Milano) for precise and sensitive quantification in urine of the pterins after oxidation: neopterin, monapterin, isoxanthopterin, pterin, biopterin and primapterin. The HPLC method employs a C18, 5- μ m particle size analytical column (250 mm \times 4.6 mm), 10- μ L injection volume, column at room temperature, excitation at 350 nm and emission at 450 nm, and an acetate buffer mobile phase at a flow rate of 1.1 mL/min. These conditions resolve the six molecules as well as diluent peaks within 20 min. The method is linear for all the six molecules (0.2-34.0 μ mol/L). The detection limits were <0.08 μ mol/L at a signal-to-noise ratio of two. The relative standard deviation was <5% (<10% only for monapterin)

for the within-assay imprecision (n=10) and <9% for the between-assay imprecision (n=6). The recovery of different amount (1.3-5.5 μ mol/L) of the six molecules added to an urine sample was 92-108%. The specific and sensitive method described may offer a means for determining BH4 and five metabolites. The method is characterized by high recovery and good reproducibility; it is well suited for routine operation in every newborn with even slight but persistent hyperphenylalaninemia unresponsive to a low-phenylalanine diet.

Speaker Biography

A Barassi has completed her PhD from Insubria University School of Medicine (Varese, Italy) and Post-doctoral studies from Insubria University School of Medicine (Varese, Italy). She is working as an Assistant Professor of Clinical Biochemistry and Clinical Molecular Biology, University of Milan from 2006 and Assistant of Laboratory of Clinical Chemistry, San Paolo University Hospital, Milan, Italy from 2007. She has published 67 papers in reputed journals and has been serving as an Editorial Board Member of *repute*.

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