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Determination of Urinary Pterins by High Performance Liquid Chromatography for Detection of Heterozygotes of Atypical Phenylketonuria Caused by Biopterin Synthetase Deficiency

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Atypical phenylketonuria (PKU) caused by biopterin synthetase deficiency is an inherited metabolic disease. In order to understand the genetic trait and to detect heterozygotes for genetic counseling, the determination of urinary pterins by high performance liquid chromatography (HPLC) was investigated. Urinary pterins were oxidized by maganese dioxide in acidic condition, followed by purifying with Florisil and Lewatit microcolumns. After the pretreatment, total biopterin (B), neopterin (N) and other pterins, including monapterin isoxanthopterin, and 6-hydroxymethylpterin, were separated by reverse phase (C-18) HPLC with an elution gradient of 3% methanol and methanol/isopropanol/acetic acid (49/49/2). The pterins were detected by fluorescence (ex.350nm, em.450nm) and 6-methylpterin was used as an internal standard. Recoveries of B and N were both above 80%. The percentages of biopterin, B%=B/(B+N), were

53-76 and 43-76 in apparently normal Chinese adults and children, respectively. The B% of 10 adult obligate heterozygotes (37-44) were lower than the normal control, but higher than that of 7 atypical PKU patients (0.4-2.8). In classical PKU patients, however, the B% were all in the normal range. A sibling of an atypical PKU patient was detected as heterozygotes (B%=27.2) by this method. The results indicated that urinary pterin analysis by HPLC is a good diagnostic aid for differential variant forms of PKU and for detection of heterozygotes of atypical PKU caused by biopterin synthetase deficiency. Besides, this urinary pterin analytical method may also be applied to the study of psychiatric and immunological disorders.