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DETERMINATION OF URINARY PTERINS BY HIGH PERFORMANCE LIQUID CHROMATOGRAPHY FOR DIFFERENTIAL DIAGNOSIS OF VARIANT FORMS OF PHENYLKETONURIA.

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Differential diagnosis of the classical phenylketonuria (PKU) and the atypical PKU caused by tetrahydrobiopterin deficiency is very important, becuase their medical managements are different. For differential diagnosis, the determination of urinary pterins by high performance liquid chromatography (HPLC) was studied. Urinary pterins were oxidized by maganes dioxide in acidic condition, followed by purifying with Florisil and Lewatit microcolumns. Analysis of total biopterin (B) and neopterin (N) was finally achieved by reverse phase (C-18) HPLC with fluorescent detection (ex.350nm, em.450nm). 6-methylpterin was used as internal standard. Pterins were eluted by a gradient of 3% methanol and methanol/isopropanol/acetic acid. Revoceries of B and N were both above 80%. Chinese reference ranges (nmol/mmol creatinine) of urinary B and N were 382-1140 & 147-521 for adults, and 350-2968 & 288-2608 for children, respectively. The percentage of biopterin, B%=B/(B+N), in adults and children were 53-76 and 43-76, respectively.. The B (39-252) and B% (0.4-2.8) of 7 atypical PKU patient were lower than normal control, but N (4161-22959) were higher. Urinary pterins of 7 classical PKU patients were also studied. The B% were all in the normal range, but some of them without dietary therapy had B and N higher than noromal control. The results indicated that the urinary pterins analysis is a good diagnostic aid for differential variant forms of PKU and should be used to select appropiate therapy for patients discovered by neonatal screening program.