Proc 3rd Joint Annual Coference of Biomedical Sciences, Taipei, p.159

147. HETEROZYGOTE DETECTION OF CLASSICIAL PHENYLKETONURIA BY DETERMINATION OF PHENYLALANINE AND TYROSINE IN FASTING PLASMA.

K.-J. Hsiao, T.-T. Liu*, S.-J. Wu. Clinical Biochem. Res. Lab., Dept. of Medical Research, Veterans General Hospital; Taipei, Taiwan 11217; R.O.C.

Classical phenylketonuria(PKU) is an autosomal recessive inherited disease caused by defects in phenylalanine hydroxylase. Since direct measurement of this enzyme activity for heterozygote detection requires a liver biopsy. The non-invasive method for discriminating between normals and heterozygotes by determining plasma phenylalanine(Phe) and tyrosine(Tyr) was therefore studied. Fasting venous blood specimens were obtained from 42 apparently normal control subjects and 14 obligate heterozygotes of classical PKU. Plasma were separated immediately after collection and deproteinized by sulphosalicylic acid. Phe and Tyr were quantitated by ion exchange chromatographic amino acid analyzer. Using Phe and Tyr level as analysis parameters, the discriminant function for heterozygote detection derived from Discriminant Analysis (SPSS PC+ program) was DSC=0.1015[Phe]-0.0761[Tyr]-1.7922. None of the PKU gene carriers were missclassified, but one of the apparently normal subjects was missclassified. The discriminant function gave 98.2% of correct classification. With this discriminant function, we could determined carrier status and provide genetic counsling for Chinese PKU family simply by collecting fasting blood without liver biopsy or Phe loading test.

148 STUDY OF [illegible]