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148. STUDY OF RESTRICTION FRAGMENT LENGTH POLYMORPHISMS AT THE HUMAN PHENYLALANINE HYDROXYLASE LOCUS AND EVALUATION OF ITS POTENTIAL APPLICATION IN PRENATAL DIAGNOSIS OF PHENYLKETONURIA IN CHINESE. S.H.Chen*[1], K.J. Hsiao[2], L.H. Lin[2], T.T. Liu[2], R.B. Tang[3] and T.S. Su[1,2] Graduate Institute of Microbiology and Immunology[1], National Yang-Ming Medical College and Depts. of Medical Research[2] and Pediatrics[3], Veterans General Hospital, Taipei, Taiwan 11217; R.O.C.

Using a human phenylalanine hydroxylase cDNA as a probe, the restriction fragment length polymorphisms at the human phenylalanine hydroxylase locus were detected with restriction enzymes: BglII, PvuII, EcoRI + BamHI, MspI, XmnI, HindIII and EcoRV. The frequency of the observed heterozygosity of the restriction site polymorphisms at this locus in the Chinese population is 53.6%, which is significantly lower than that in Caucasians.

By Southern blot analysis using a human phenylalanine hydroxylase cDNA as a probe, no DNA rearrangement or deletion of phenylalanine hydroxylase locus was detected among mutant phenylalanine hydroxylase genes in seven Chinese classical phenylketonuria families analyzed. Restriction site polymorphic haplotype analysis of these seven phenylketonuria families reveals the mutant alleles belong to 5 different haplotypes, i.e., hapoltype 4, 11 and three unreported haplotypes. 71.4% of the mutant phenylalanine hydroxylase genes are confined to hapoltype 4. The status of Chinese classical phenylketonuria can be determined about 42.8% of the time by the analysis of eight restriction site polymorphisms at the phenylalanine hydroxylase locus.