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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

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Using a human phenylalanine hydroxylase cDNA probe, the restriction fragment length polymorphisms at the human phenylalanine hydroxylase locus were determined with the 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 a Chinese population is approximately 54%, which is significantly lower than that in Caucasians.

No DNA rearrangement or deletion of phemylalanine hydroxylase locus was detected among mutant phenylalanine hydroxylase genes in seven Chinese classical phenylketonuria families. Haplotype analysis of these seven families revealed that the mutant alleles belong to 5 different haplotypes, i.e., haplotype 4, 11 and three unreported haplotypes. The majority of normal and mutant phenylalanine hydroxylase genes are confined to haplotype 4. These results indicate that approximately 42% of Chinese PKU families are informative for prenatal diagnosis of PKU when eight restriction sites linked to the phenylalanine hydroxylase locus are examined.

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