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Estimation Haplotype Frequency of *Bgl*II/*Eco*RI/VNTR Markers at the *PAH* Gene Region in Iranian Population

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KEYWORDS Phenylalanine Hydroxylase (PAH). Phenylketonuria (PKU). Haplotype. Polymorphism Markers. Iranian Population.

ABSTRACT Deficiency in phenylalanine hydroxylase (PAH) is the main molecular characteristic of phenylketonuria (PKU). So far over 500 different mutations in the *PAH* gene have been identified **as** associated with the disease. Mutation analysis of the *PAH* gene is a time-consuming and cost-effective procedure. Therefore, molecular markers which are highly linked to the *PAH* gene, have been used in carrier detection and prenatal diagnosis in PKU families. These markers show a population dependent based haplotype frequency. In the present study, the haplotype frequency of three markers including *Bgl*II, *Eco*RI and VNTR, at the *PAH* gene region were investigated in Iranian population. Nine different alleles for VNTR marker with 3, 5, 6, 7, 8, 9, 10, 11 and 13 core repeats were detected. Alleles 4 and 13 were found specific to the Iranian population. The haplotype frequency was calculated using FBAT, PHASE and Arlequin (haplotype frequency estimation computer programs). Among the 36 possible estimated haplotypes identified for 2 alleles (+ and -) for *Bgl*II and *Eco*RI; and nine alleles for VNTR, ten haplotypes showed relatively high frequency (e" 5%), based on the above programs. Therefore, a combination of *Bgl*II-*Eco*RI-VNTR could be suggested as an informative haplotype in performing carrier and prenatal diagnosis of the *PAH* gene mutations in Iranian population.