

D-33

A *PvuII* Polymorphism of Human Apo B mRNA Editing Protein Gene

홍승호*, 임진희¹, 강병용¹, 김진규², 이정주¹

서울대 분자생물학과, ¹생물학과, ²입상병리과학교실

We have identified a genetic polymorphism of human apoB mRNA editing protein (HEPR) gene in Korean population. This polymorphism occurs according to absence (*P1* allele) or presence (*P2* allele) of the *PvuII* restriction site. The allele frequency in 84 healthy subjects was 0.38 for *P1* and 0.62 for *P2* allele, respectively. Subjects with the *P2P2* genotype was associated with the most elevated LDL cholesterol levels, and subjects with a heterozygous genotype had intermediate levels, indicating a gene dosage effect ($P < 0.05$). Thus, current study might be provided for basic data in elucidating the interactions between this genetic variants and disorders related to lipid metabolism.

D-34

Mae III Restriction Fragment Length Polymorphism of the Alcohol Dehydrogenase 2 (*ADH2*) Gene in Korean

목지원*, 김민영¹, 박경숙

성신여자대학교 자연대학 생물학과, ¹한효과학기술원

Alcohol dehydrogenase (ADH:alcohol:NAD⁺ oxidoreductase, EC 1.1.1.1) in human liver which is responsible for the oxidative metabolism of ethanol consists of eight genes (ADH 1-8) with corresponding subunits. The genes of *ADH* are polymorphic at *ADH2* and *ADH3* loci. *ADH2* gene located on 4q21-25 consists of nine exons. Three alleles of *ADH2* gene have been recognized in the exon 3 region such as : *ADH2*^{*1} for $\beta 1$ subunit (common in Caucasians), *ADH2*^{*2} for $\beta 2$ subunit (common in Orientals), and *ADH2*^{*3} for $\beta 3$ subunit (common in Blacks). The polymorphism of *ADH2* is due to an mutation of a single amino acid at position 47. *ADH2*^{*1} has Arginine (CGC) while *ADH2*^{*2} has Histidine (CAC). We determined *ADH2* genotype of exon 3 using the restriction fragment length polymorphism of leukocyte DNA in 287 unrelated Korean. *Mae* III RFLP analysis of *ADH2* gene showed fragments of 95bp and 60/35bp behaved as *ADH2*^{*1} and *ADH2*^{*2} allele. The allele frequencies for *ADH2*^{*1} and *ADH2*^{*2} were 0.294 and 0.706, respectively. The *ADH2*^{*2} frequencies vary in different ethnic groups with the range of the lowest 0.0 in the American Indians and the highest 0.65-0.80 in Mongoloids. The genetic variation of *ADH2* genotype may influence the risk of alcoholism through acetaldehyde formation. The frequency of *ADH2*^{*2} of the alcoholics (0.373) is significantly ($p < 0.01$) lower than that of the non-alcoholics (0.706).