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### The Influences of RFLPs in the Cholesteryl Ester Transfer Protein (CETP) Gene on the Regulation of Plasma HDL Cholesterol Levels in the Koreans

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Cholesteryl ester transfer protein (CETP) is expected to play an important role in modulating lipid levels in the plasma. This protein may transfer lipid particles from cells to plasma lipoproteins or between circulating lipoproteins. In the present study, we studied several restriction fragment length polymorphisms (RFLPs) in cholesteryl ester transfer protein (CETP) gene and examined if CETP gene influences plasma lipid levels in general Koreans (n=270). The frequencies of the major alleles at *Bam*HI, *Eco*NI, *Msp*I, *Taq*IA, *Taq*IB, and *Hin*fI polymorphic sites were 0.77, 0.55, 0.94, 0.85, 0.62, and 0.81 respectively. The distribution of the genotypes of all RFLPs were in Hardy-Weinberg equilibrium and the differences in allele frequencies between the sexes were insignificant in this population. The *Bam*HI and *Msp*I RFLPs were informative for association studies with HDL cholesterol in this population. Subjects with genotype B2B2 of the *Bam*HI RFLP (polymorphism in intron 9) exhibited significantly lower HDL cholesterol levels and higher CETP activity than mean of total samples. Genotype M1M2 of the *Msp*I RFLP (M2 is the A to G substituted allele in exon 15) exhibited significantly higher HDL cholesterol levels in male. Multivariate analysis showed that allelic variation in *Bam*HI and *Msp*I sites accounted for 4.0 and 5.9% of the total inter-individual variation in plasma HDL cholesterol in male (F=2.29, p=0.10; F=3.4, P=0.03). In conclusion, the genetic variation of CETP gene is related to the modulation of plasma HDL cholesterol levels and the extent of the effect seems to be different between male and female in the Koreans.

## F108

### Two Partial Deletion Mutations of the LDL Receptor Gene

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Twenty-eight unrelated heterozygotes for familial hypercholesterolemia (FH) were screened to assess the nature of major structural rearrangements at the LDL receptor gene in Korean FH patients. Genomic DNA was analyzed by Southern blot hybridization with probes encompassing exons 1-18 of the LDL receptor gene. Two different deletion mutations (FH29 and FH110) were detected in three FH patients. Each of the mutations was characterized by the use of exon-specific probes and detailed restriction mapping, mediated long-PCR. The FH29 was 3.85-kb deletion extending from intron 6 to intron 8 and the FH110 was 5.71-kb deletion extending from intron 8 to intron 12. Sequence analysis revealed that both deletions have occurred between two Alu-repetitive sequences that are in the same orientation. This suggested that in those patients the deletions are caused by an unequal crossing-over event follows mispairing of two Alu sequences on different chromatids during meiosis. Moreover, in both deletions, the recombinations were related to an Alu sequence in intron 8 and the deletion breakpoints were shared within a specific sequence, 27-bp in length. This support a hypothesis that this region might have some intrinsic instability and act as one of the important factors in recombinational large rearrangements.