F837 Mutation Analysis of the 3' Region of PKD1 Gene in Patients with Autosomal Dominant Polycystic Kidney Disease (ADPKD)

Un Kyung Kim*[1], Jea Hyun Shin[2], Sung Han Kim[2], Kyu-Baek Lee[4], Dong-Kyu Jin[5], Yong Namkoong[3], Chung Choo Lee[2]
Dept. of Biology[1] and Molecular Biology[2], Seoul National Univ. Seoul, 152-742, Dept. of Biology[3], Kangnung National Univ.210-702, Dept. of Nephrology, SMC[4], 100-634, Dept. of Pediatics, SMC[5], 135-710

Autosomal dominant polycystic kidney disease(ADPKD) is a common genetic disorder with the frequency of 1:1000 and results in renal failure due to progressive cyst development. ADPKD is caused by mutations in at least three different genes (PKD1,PKD2, PKD3). The major locus, PKD1, locates to 16p13.3 and encodes a 14kb mRNA that is an integral membrane protein of 4302 amino acids with cell–cell or cell matrix interactions. In previous studies, a few mutations have been founded in the 3'single copy region of the gene. In this study, we have screened PKD1 gene from affected individual to assess frequency and nature of disease-causing mutations in Korean population. We detected novel aberrant bands in exon 36, 40, 41, 44 of the single copy region of the PKD1 gene using single strand conformation polymorphism (SSCP) analysis. The newly described mutations are one small deletion (8bp del at 11548-11555), two missense mutations (T→C at 10956, C→G at 12213), an insertion of G nucleotide causing a frameshift. In addition, polymorphisms were detected. Our findings indicate that many different mutations are likely to be responsible for ADPKD in the Korean population. In order to understand the molecular mechanisms underlaying ADPKD, we must screen for duplicated region of the PKD1 gene and this will help in elucidating the physiopathological role of the gene product.

F838 Haplotype analysis of the *apoB* gene in Hypertensives

Byung Yong Kang*, Hyun Seon Eo, Kyung Ho Lee and Chung Choo Lee
Department of Biology, Seoul National University

ApoB is the sole protein component of LDL, which plasma levels are associated with an increased risk of cardiovascular disease. The association of the six polymorphisms (*I/D*, *Xba*I, *Eco*RI, *Pvu*II, *Msp*I and 3'-VNTR) of *apoB* gene with essential hypertension was studied in 136 normotensive and 100 hypertensive subjects. Genotype and allele frequencies of *apoB* gene were not significantly different between cases and controls. However, significant linkage disequilibrium was detected between *I/D* and *Pvu*II polymorphisms by haplotype analysis (D' = 0.5882, P<0.05). Also, *I-X1-E1* and *D-X1-E1* haplotypes were not detected in normotensives. Therefore, the finding of two haplotypes in essential hypertensives suggests that *apo*B locus may be a useful tool for population association study in essential hypertension, at least Koreans.