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한국인 2형 당뇨 콩팥병에서 최초의 전장유전체 연관분석

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The First Genome-wide association Study of Diabetic Nephropathy in Korean Type II Diabetes Patients

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Background: It has been suggested that genetic susceptibility plays an important role in the pathogenesis of diabetic nephropathy. Recently, several genome wide association studies (GWASs) suggested that specific polymorphisms of candidate genes were associated with susceptibility to diabetic nephropathy. However, there was weak point in contents of GWAS DNA chip for GWAS. In fact, previous contents of GWAS DNA chip were fixed and did not cover SNPs in exon region and promoter region. In present study, we used the Axiom™ Genome-Wide Human Assay. The contents of assay are selected by researcher and useful to investigate association between several candidate SNPs and specific diseases.

Methods: To investigate whether specific polymorphisms are involved in the development of the diabetic nephropathy, 87 diabetic nephropathy patients and 104 diabetic controls in Korean with type II diabetes were studied. We firstly selected 47,777 genes of homo sapiens in NCBI gene database and searched the SNPs in dbSNP database. And the criteria for selection exonic SNPs, promoter SNPs, and intron SNPs in each gene were following: (1) SNPs with >10 % minor allele frequency (MAF), (2) >0.1 heterozygosity, (3) known genotype frequencies of SNPs in Asian population, (4) SNPs studied in previous study, and (5) unknown SNPs. Finally we selected 378,707 SNPs. Logistic regression models were performed to determine odds ratio (OR), 95% confidence interval (CI), and P value. The analysis were analysis using Helixtree program

Results and Conclusion: Among 378,707 SNPs, three SNPs (rs3214159 in *ABCC8* gene, *rs3747636* in *PIK3C2B* gene, and rs3765156 in *PIK3C2B* gene) showed strongly significant association with diabetic nephropathy ($p<0.00001$). These results suggest that these significant SNPs may be useful to investigate the development of diabetic nephropathy.

Key Words: 당뇨병성 신증, 단일 염기 다형성, 전장유전체 연관분석
Diabetic nephropathy, SNP, GWAS