

## Genome wide assay를 이용한 IgA 신증 환자의 유전자 감수성 연구

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### Discovery of New Risk Gene Loci in IgA Nephropathy: Genome Wide Human Assay

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IgA nephropathy (IgAN) is the most common form of glomerulonephritis in Korea. The etiology of IgAN is complex with high genetic heterogeneity. Recently, several genome wide association studies (GWAS) suggested that specific polymorphisms of candidate genes were associated with susceptibility to IgAN. 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. To investigate whether specific polymorphisms are involved in the development of the IgAN, 182 IgAN patients and 455 healthy controls 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 Helix tree program

Among 378,707 SNPs, 13 SNPs showed strongly significant association with IgAN ( $p < 0.00001$ ). We identified at rs55730189, rs2296136 and rs2597775 that implicated the genes encoding unc-51 like autophagy activating kinase 2 (ULK2), ankyrin repeat domain 16 (ANKRD16) and quinoid dihydropteridine reductase (QDPR) as susceptibility genes. These results suggest that these significant SNPs may be useful to investigate the development of IgAN.

**Key Words:** IgA 신증, 유전자, 전장 유전체

IgA nephropathy, SNP, Genome wide assay