O-7(기초) Novel Single Nucleotide Polymorphisms of VEGF Gene in Polycystic Ovary Syndrome

Eung-Ji Lee[1], Min-Young Lee[1], Sook-Hwan Lee[1], Bermseok Oh[2], Jong-Young Lee[2], Kuchan Kimm[2], Kwang-Hyun Baek[1]

[1]Genome Research Center for Reproductive Medicine and Infertility, CHA General Hospital,
[2]Center for Genome Science, Korean National Institute of Health

Background & Objectives: It has been suggested that many women with reproductive age have polycystic ovary syndrome (PCOS) and it is one of major factors for female infertility. Because of steroid hormone abnormalities, menstrual irregularities are shown in numerous patients with PCOS. Previous reports suggested that abnormal androgen steroidogenesis in PCOS can be occurred, resulting from increased vascularization by up-regulated VEGF. Therefore, it is suggested that VEGF may be involved in one of symptoms for PCOS. In this study, we considered VEGF as a candidate gene for pathogenesis of PCOS.

Method: To investigate single nucleotide polymorphisms (SNPs), we screened candidate SNPs in 6p12 of VEGF gene by direct sequencing and genotyped enlarged subjects for each selected SNP by TaqMan assay. Results were analyzed by HapAnalyzer for association test.

Results: After association tests for each SNP and haplotype, we found that among 10 SNPs which were analyzed with large subjects, one novel SNP at +9812 site and one known SNP at +13553 site are significantly associated with PCOS and one haplotype (ht4) has an association with PCOS in a Korean population. Interestingly, frequencies of minor alleles for these SNPs were much higher in a control group than in the PCOS patient group at significant levels ($p=0.0361$ and $p=0.0305$).

Conclusions: In this study, we identified a significantly associated novel SNP and a known SNP in INSR gene (+9812C>T and +13553C>T) with PCOS. From the results of association test, it is suggested that the minor allele T in +9812C>T and the minor allele T in +13553C>T of INSR gene may have protective effects in pathogenesis of PCOS in a Korean population.