P3 Association study for single nucleotide polymorphisms in *INSR* gene and polycystic ovary syndrome

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Objectives: To evaluate associations between several single nucleotide polymorphisms of the *insulin receptor* (*INSR*) gene and polycystic ovary syndrome (PCOS) in a Korean population.

Materials and Methods: We have sequenced all exons of *INSR* to discover single nucleotide polymorphisms (SNPs) in 24 Korean DNA samples using the ABI PRISM 3730 DNA analyzer. After finding several candidate SNPs, we recruited 134 women who have been diagnosed for PCOS as a patient group and another 100 healthy women as a control group. For genotyping of polymorphic sites, we used TaqMan SNP genotyping assays.

Results: We analyzed frequencies of genotypes for SNPs in *INSR* gene, which include +109482 A)G, +109665 C>T, +125498 A>G, +127527 G>A, +143485 G)C, +161822 G)A, +168606 C>T, +168828 T>A and +176477 C>T. All subjects have a major genotype for two SNPs, +109482 A)G and +168828 T>A, in PCOS and control groups. The frequency of T allele for +109665 C>T in PCOS group was a little bit higher than in control group. In the case of +125498 A)G, major genotype AA was the most frequent genotype and frequencies of three genotypes were similar between two groups. The frequency of A allele shown in +127527 G)A was higher in PCOS group than in control group. After the frequency analysis for +143485 G)C SNP, we were able to find that three genotypes were shown to similar frequencies in two groups. For +161822 G>A, the frequency of A allele was higher in PCOS group than in control group. In the case of +168606 C>T, the minor allele T was shown more frequently than wild type CC in both groups and frequencies for three genotypes were similar between two groups. For the last study with +176477 C>T, we found that the frequency of a minor allele T was higher in control group than in PCOS group.

Conclusion: All analyzed SNPs of *INSR* gene in this report are not associated with PCOS due to the fact that they had similar frequencies of three genotypes between PCOS and control groups.

Key words: PCOS, SNP, INSR

P4 Association between single nucleotide polymorphism in exon 17 of insulin receptor gene and polycystic ovary syndrome in a Korean population

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Objectives: To assess the association between the His 1085 C/T polymorphism of the *insulin receptor* (*INSR*) gene and polycystic ovary syndrome (PCOS) in a Korean population.

Materials and Methods: We recruited 174 women who have been diagnosed for PCOS based on the revised diagnostic criteria announced in 2003 ASRM/ESHRE Rotterdam consensus and 93 healthy women as a control group. We extracted the genomic DNA from the blood of patients with PCOS and normal controls. RFLP (Restriction Fragment Length Polymorphism) analysis was performed to determine genotypes for the His 1085 C/T polymorphism at the tyrosine kinase domain in the *INSR* gene.

Results: The frequency of T allele was shown highly both in patient and control groups. The frequency of C allele which known as a normal allele was slightly higher in a patient group than in a control group. However, it was not a significant level.

Conclusions: The C/T polymorphism in exon 17 of the *INSR* gene is not associated with susceptibility of PCOS in a Korean population. And this result is opposed to previous reports published in America and China. We found that the cause of difference between our result and previous reports is come from experimental misinterpretation in genotyping procedure previously performed by other researchers.

Key words: PCOS, SNP, INSR