

# Association Study of Aromatase Gene (CYP19A1) in Essential Hypertension

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## Abstract :

**Background:** As aromatase-deficient mice, which are deficient in estrogens, reportedly have reduced blood pressure, the aromatase gene (CYP19A1) is thought to be a susceptibility gene for essential hypertension (EH). The aim of the present study was to investigate the relationship between CYP19A1 and EH by examining single nucleotide polymorphisms (SNPs).

**Methods:** Five SNPs in the human CYP19A1 gene (rs1870049, rs936306, rs700518, rs10046 and rs4646) were selected, and an association study was performed in 218 Japanese EH patients and 225 age-matched normotensive (NT) individuals.

**Results:** There were significant differences between these groups in the distribution of genotypes rs700518 and rs10046 in male subjects, and genotypes rs700518, rs10046 and rs4646 in female subjects. On multiple logistic regression analysis, a significant association between rs700518 ( $p=0.023$ ) and rs10046 ( $p=0.036$ ) in male subjects and rs700518 in female subjects ( $p=0.018$ ) was noted. Interestingly, the risk genotypes of rs700518 and rs10046 showed a sex-dependent inverse relationship. Both SBP and DBP levels were higher in total (cases and controls) male subjects with the G/G genotype with rs700518 or the T/T genotype with rs10046 than in male subjects without the G/G genotype or T/T genotype. SBP levels were lower in female subjects with the G/G genotype with rs700518 than in female subjects without G/G. The A-T haplotype constructed with rs1870049 and rs10046 was a susceptibility marker for EH.

**Conclusions:** We confirmed that rs700518 and rs10046, as well as a haplotype constructed with rs1870049 and rs10046, in the human CYP19A1 gene can be used as genetic markers for gender-specific EH.

## Key Word :

Essential hypertension, aromatase, CYP19A1, single nucleotide polymorphism, genetic