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Single-Nucleotide Polymorphisms and Mutation Analyses of the *TNP1* and *TNP2* Genes of Fertile and Infertile Human Male Populations

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Previously, we examined the relationship between protamine gene variations and human male infertility. In this study, we show specific variability in the transition nuclear protein genes (TNPs) of sterile male patients. Transition nuclear proteins (TNPs) are major nuclear proteins that replace nuclear histones, leading to eventual substitution by protamines during human spermiogenesis. Analysis of the human *TNP1* and *TNP2* gene sequences in 282 sterile male patients and 270 (*TNP1*) and 266 (*TNP2*) proven-fertile male volunteers revealed 5 amino acid substitution-causing single nucleotide polymorphisms (SNPs) in the open-reading frame of the *TNP2* gene. On the other hand, a deletion of 15 nucleotides, which encompassed the recognition site for the cAMP response element (CRE) transcription factor, was found in the 5'-promoter region of the *TNP1* gene in infertile men. This deletion reduces *TNP1* expression and may cause human male infertility.

Key words: Protamine, transition nuclear protein, sperm, male infertility, genome, promoter, SNPs

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