Study on association of four polymorphisms in TNP2 gene with spermatogenesis and causes of male infertility

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Abstract
Protamines are DNA-binding proteins in the sperm nucleus, which cause differentiation of human spermatogenesis. Protamines required for packaging nucleus sperm by replacement of histones with protamines. Sperm protamine deficiency has been associated with spermatogenesis failed and caused male infertility. In this study association of four SNPs in TNP2 gene were studied in Iranian idiopathic infertile men with azoospermia or oligospermia. Analysis of four SNPs include T1019G, G1272C, G deletion at 1036 and 1046 in TNP2 gene was performed by DNA extraction from blood samples of 96 idiopathic infertile men with azoospermia or oligospermia and 100 normal control men. Then using restriction fragment length polymorphism (PCR-RFLP) for detection of T1019G and G1272C, SNPs and for identification of G deletion at 1036 and 1046 used Single Strand Conformation Polymorphism (PCR-SSCP). Results were confirmed by DNA sequencing analysis. For (G1272C) in TNP2 gene, frequency of CC genotype was differented in fertile and infertile groups but statistical analysis showed no significant association related to this SNP in case and control groups. As also no polymorphisms were found for T1019G and G deletion at 1036 and 1046 nt. These results are consistent with previous studies and indicating that four tested SNPs was not associated with oligospermia and azospermia and idiopathic male infertility in Iranian population.

Keywords: Male infertility, PCR-RFLP, PCR-SSCP, SNP, TNP2 gene