Evaluation of genetic variations in exon 4 and intron 4 of RABL2B gene in infertile men with Oligoasthenoteratospermic disorder.

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One of the main causes of male infertility is defect in structure and function of sperm cells. Normal spermogram must have appropriate morphology, count and motility. Oligoasthenoteratospermic infertile men have sperms with abnormalities in count, motility and morphology which are not able to fertilize oocyte. Numerous proteins are involved in sperm formation. One of these proteins is RAB Like 2B (RABL2B), which recently its essential role in sperm tail assembly and fertility in mouse has been demonstrated. So its gene, which called RAB Like 2B (RABL2B), is an appropriate candidate gene in human studies. RABL2B protein is a G protein with 4 GTP binding domains which have important roles in protein function. Exon 4 of RABL2B gene codes one of these main domains and intron 4 of RABL2B gene is the location for binding to some important transcription factors. The purpose of this study is to evaluate the genetic variations of exon 4 and intron 4 of RABL2B gene in Oligoasthenoteratospermic infertile men and controls. In this study, 30 infertile men with Oligoasthenoteratospermia and 30 normozoospermic men as controls were recruited. To study the genetic variations, DNA was extracted from peripheral blood, then PCR sequencing was done. Sequence analysis results did not identify any mutations or single-nucleotide polymorphisms (SNPs) in exon 4, but an intronic variant was found in heterozygote form in 5 patients. This intronic variant was a (C) nucleotide deletion (rs:144944885). No mutations or (SNPs) was identified in controls. As mutations and (SNPs) in introns can have a pronounced effect on altering pre-mRNA splicing, bioinformatics analysis and further research on transcription factors which binds to this region of RABL2B gene is recommended.

Keywords: Oligoasthenoteratospermia, RABL2B gene, Intronic variant.