Genetic analysis of exon 5 of AKAP3 gene in infertile men with short tail sperm defect

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One kind of sperm abnormalities that leads to men infertility is short flagella of sperms. In this defect, fibrous sheath and axoneme are disorganized, the sperms tail is short, the numbers of sperms in the semen fluid reduce and the sperms are immotile. Recently, demonstrated that two A- kinas anchoring protein (AKAP3 and AKAP4) are the most abundant structural proteins of the fibrous sheath. AKAP3 is involved in organizing the basic structure of the fibrous sheath. A Kinas anchoring protein 3 (AKAP3) gene encodes a protein that is involved in the fibrous sheath structure, regulation of sperm motility, head-associated functions such as capacitation and the acrosome reaction. AKAP3 interacts with the regulatory subunit of Protein Kinas A via its dimerization / docking domain. To our knowledge, no study has been done on this gene in males with short tail sperm disorder. In the present study, 30 patients with short tail sperms and 40 males with normal spermogram referred to Røyan institute were enrolled as case and control groups respectively. The genetic variation in exon 5 of AKAP3 gene which encodes the functional domain of this protein was studied. PCR - sequencing was undertaken on extracted DNA from blood samples of control and patient groups'. According to the results, a polymorphism 1499 T>C was seen in 5 of the patients whereas none of the individuals in the control group had this alternation. A significant difference was seen between the studied groups after analyzing the data by SPSS. In this alternation isoleucine is changed to threonine. This alternation causes the hydrophobic (I) amino acid with medium size changes to a medium size and polar (T) amino acid. Accordingly it can be concluded that this genetic variation can be proposed as none of the etiological reseans of short tail sperm defect in men.

Keywords: short tail sperm defect, AKAP3 gene.

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