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TITLE

ASSESSMENT OF EXON 8 OF DPY19L2 GENE IN PATIENTS WITH TOTAL & PARTIAL GLOBOZOOSPERMIA

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ABSTRACT

Globozoospermia is a rare but severe teratozoospermia disorder which causes male infertility. Total globozoospermia is diagnosed by the presence of 100% round-headed spermatozoa lacking an acrosome in semen analysis. Partial globozoospermic patients are also a larger group with 20-90% round-headed acrosome-less sperm cells in semen analysis. Recent studies have shown that in large majority of globozoospermic patients, deletion of a 200 kb segment including the DPY19L2 gene occurs. Among all the genes in this locus, DPY19L2 is the only gene which is dominantly expressed in the testis and has been shown to be involved in the cause of this phenotype. It must be mentioned that no studies have been done on its effect on partial globozoospermia patients until now. The aim of this study was to peruse exon8 variations in Iranian infertile men with total globozospermia and partial globozoospermia without DPY19L2 deletion In this study, 24 men with total globozospermia and 20 men with partial globozoospermia and 24 men with normal spermogram were selected. In a second step, in conjunction with screening for the DPY19L2 deletion, we sequenced exon8 and intron boundaries in the non-deleted patients using specific primers and PCR technique. The results showed a whole DPY19L2 gene deletion in 70.83 % of total globozospermia patients. None of partial globozoospermic patients and also control group showed this large deletion. No mutations were detected in exon 8 of the patients without the gene deletion. According to our data and based on the critical role of DPY19L2 protein in acroplaxome attachment to the nucleus, it can be concluded that absence of this protein is one of the major causes of total globozoospermia in Iranian infertile men. Therefore, it seems that an unknown factor or mutation instead of whole DPY19L2 gene deletion cause similar phenotype in partial globozoospermia.

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