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Poster Presentations

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Y chromosome-AZFc atypical partial microdeletions in Iranian severe oligozoospermia men

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Background: Azoospermia Factor c (AZFc) region contains important gene families (including DAZ, CDY1) for male spermatogenesis. Meiotic homologous recombination within amplicons is the etiologic cause for various partial microdeletions as well as the changes in copy number of mentioned genes. The association of AZFc partial microdeletions with impaired spermatogenesis and infertility is still debated.

Objective: The aim of this study was to investigate the nature and frequency of partial microdeletions (gr/gr,

b2/b3 and b1/b3) in AZFc region.

Materials and Methods: Total DNA extracted from the peripheral blood of 200 oligozoospermic male (sperm count < 5 mil./mL) as patients and 200 fertile males as controls was used to detect partial microdeletions by multiplex PCR and six sequence-tagged sites (STS) markers. Also PCR-RFLP technique was used to detect deletion of gene copies.

Results: Other than 18/400 (4.5%) cases of typical partial AZFc microdeletions (gr/gr, b2/b3 and b1/b3), we detected two cases of deletion pattern of sY1191 and sY1161 (1%), 1 case of sY1201 (0.5%) and one case of sY1258 (0.5%) in the oligozoospermia group. The mechanism of the rearrangement occurring within the Y chromosome of these individuals is complex and unknown.

Conclusion: Similar to some other studies, we have shown that in addition to known AZFc partial microdeletions, other atypical types may also been detected in patients with spermatogenic defects. Although, these findings are extremely rare, very small in size, and may not have much effect on spermatogenesis, there is a risk of vertical transmission and even expansion in the size of deletions in male offspring leading to a boost of infertility rate in the mentioned generation. Therefore, genetic counselling and informed decision making (PGT as an option) will be necessary for such couples before ART. Hence, testing for AZFc partial deletion may be suggested in oligozoospermia cases with idiopathic reason.

Key words: Male infertility, Y Chromosome, Oligospermia.