

Evaluating Y Chromosome Microdeletions in Infertile Men with Severe Oligozoospermia or Azoospermia at Imam Reza Hospital In Meshad

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Abstract

Background: Male factors account for nearly 50% of infertilities, among which genetic defects constitute some of the major ones. Microdeletion of the long arm of Y chromosome has been seen in about 7% of infertile men. The importance of these microdeletions lies in the possibility of their occurrence in the off-springs in ART and their de novo appearance.

Methods: This cross-sectional descriptive-analytical study was performed on 47 individuals with azoospermia or severe oligozoospermia. The cases were recruited when they attended Imam Reza Hospital in Mashad during 2006–2008. Hormone profile, including FSH, was measured and karyotyping, testicular biopsy and Y chromosome microdeletion detection, using 11 pairs of sequence-tagged site (STS method) sets which were specific for AZF and SRY loci, were performed.

Results: Three out of four patients with azoospermia had Y chromosome microdeletion (8.5%). Klinefelter's syndrome and deletion of SRY region were each seen in two patients (4.3%). Multiple AZF region deletions were seen in 75% of Y chromosome microdeletions and deletions in AZFa, AZFb and AZFc regions were seen in 25%, 75% and 100% of the cases, respectively. The prevalence of AZF deletion in patients with and without FSH abnormality were 17.6% and 3.3%, respectively, however, the differences were not statistically significant ($p = 0.125$). In patients with azoospermia and severe oligozoospermia, AZF deletion were 11.1% and 5%, respectively ($p = 0.628$). In addition, there were no significant differences in AZF deletion between patients suffering from varicocele or other related disease ($p = 1.0$). Family history had no significant effect on AZF deletion ($p = 0.239$). Testicular biopsy showed Sertoli-cell-only syndrome in three out of four patients with AZF microdeletions.

Conclusion: Male factor infertility is associated with a high incidence of Y chromosome microdeletions and transmission of these defects to the off-springs in ART, aside from their de novo occurrence, seems probable. Therefore, it would be wise to look for microdeletions in cases with severe oligozoospermia or cases with non-obstructive azoospermia. There seems to be a correlation between the prevalence of AZF regional deletions and the degree of spermatogenesis disruption but this finding needs further scientific evidence.

Keywords: Azoospermia, Azoospermic factor, Male infertility, Oligozoospermia, Severe oligozoospermia, Y chromosome microdeletion.

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