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Sex Chromosome Alignment at Meiosis of Azoospermic Men With Azoospermia Factor Microdeletion

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Deletions in the q arm of the Y chromosome result in spermatogenesis impairment. The aim of the present study was to observe the X and Y chromosome alignment in the spermatocytes of men with Y chromosome microdeletion of the azoospermia factor (AZF) region. This was performed by multicolor fluorescence in situ hybridization probes for the centromere and telomere regions. Testicular biopsies were performed in a testicular sperm extraction-intracytoplasmic sperm injection set-up in 11 azoospermic men: 8 (nonobstructive) with AZF deletions and 3 (obstructive) controls. Histological sections, cytology preparations of the testicular biopsies, and evaluation of the meiosis according to the percentage of XY and 18 bivalents formation were assessed. Spermatozoa were identified in at least one location in controls and specimens with AZFc-deleted Y chromosomes. Complete spermatocyte arrest was found in those with a deletion that included the entire AZFb region. Bivalent formation rate of chromosome 18 was high in all samples (81%-99%). In contrast, the rate of bivalent X-Y as determined by centromeric probes was lower but in the range favorable with spermatozoa findings in controls and patients with the AZFc deletion (56%-90%), but not in those with AZFb-c deletions (28%-29%). A dramatic impairment in the normal alignment of X and Y telomeres in the specimen with AZFb-c deletion was shown (29%), compared to the specimens with AZFc deletion (70%-94%). It is suggested that the absence of sperm cells in specimens with the entire AZFb and with AZFb-c deletions is accompanied by meiosis impairment, perhaps as a result of the extent of the deletion or because of the absence of genes that are involved in the X and Y chromosome alignment.

Key words: Azoospermia, chromosome pairing, FISH, Y chromosome microdeletion

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[Abstract] [Full Text] [PDF]