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The azoospermia factor locus-c region was found to be related to Klinefelter syndrome in Turkish patients

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ABSTRACT. We looked for a possible association between Klinefelter syndrome (KFS) and microdeletions in the Y chromosome in Turkish KFS patients. We examined the frequency of KFS in male patients with proven non-obstructive azoospermia and the types of Y chromosome microdeletions in these KFS patients. Fifty azoospermic patients and 50 fertile men were included in this study. KFS was found in 14 azoospermic patients. Y chromosome microdeletions were found in eight KFS patients. Azoospermia factor locus c (AZFc) was the most commonly deleted interval in KFS patients. All KFS patients had elevated plasma follicle-stimulating hormone and luteinizing hormone concentrations, but they had normal plasma testosterone concentrations. Testis biopsy of five samples with Y microdeletions revealed Sertoli cell-only syndrome. No Y microdeletions were found in the fertile group. We concluded that there could be an association between the AZFc region and KFS. Screening for this should be part of diagnostic work-up, particularly in those considering assisted reproduction.

Key words: Azospermia; Klinefelter syndrome; AZF region

