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Loss of the AZFc region due to a human Y-chromosome microdeletion in infertile male patients

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ABSTRACT. Infertility is a major reproductive health threat; the frequency of male infertility due to Y-chromosome microdeletions is 13-18% in the human population; these microdeletions involve recurrent loss of three non-overlapping regions designated as AZFa, AZFb and AZFc, associated with spermatogenic failure. Several contradictory reports have been published regarding deletion frequency based on sequence-tagged site markers and genotype-phenotype correlation. We examined the prevalence of Yq- deletion in 64 clinically diagnosed infertile male patients. We found a 3% frequency of microdeletion of the AZFc region; hormone profiles (FSH, LH and testosterone) showed significantly ($P < 0.001$) elevated levels compared to controls. No mutations were observed in the AZFa

and AZFb regions, perhaps due to the selective use of sequence-tagged site markers.

Key words: Male infertility; Y-chromosome microdeletion; AZF regions; STS markers

