Correlation between Y chromosome microdeletion and male infertility

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ABSTRACT. Dyszoospermia due to genetic factors is the leading cause of male infertility. To explore the correlation between azoospermia factor (AZF) microdeletion of the Y chromosome and male infertility, we evaluated AZF microdeletion on the long arm of the Y chromosome in 166 infertile males and 50 fertile males using multiplex polymerase chain reactions amplification and gel electrophoresis. The results demonstrated that 28 individuals had varying degrees of microdeletion in the AZF region (16.90%); 12 out of the 76 males with azoospermia and 16 out of the 90 males with oligospermia had AZF microdeletion. AZF microdeletion was not observed in any of the healthy controls. In addition, 53.60% of the AZF microdeletions occurred in the AZFc region. It can be concluded that AZF microdeletion on the long arm of the Y chromosome can result in male spermatogenesis dysfunction. Detection of AZF microdeletion can provide a theoretical basis for genetic counseling, as well as improve the diagnosis and treatment of this disease.

Key words: Y chromosome; Microdeletion; AZF; Azoospermia; Male infertility