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Genetic anomalies in patients with severe oligozoospermia and azoospermia in eastern Turkey: a prospective study

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ABSTRACT. Infertility is defined as the inability to conceive a child after one year of regular unprotected intercourse; it is a major health problem affecting about 10-15% of all couples. Infertility is due to a male factor in approximately 50% of cases. The human Y chromosome contains genes necessary for gonadal differentiation into a testis and genes for complete spermatogenesis. We examined the frequency and type of both chromosomal abnormalities and Y chromosome microdeletions in 90 patients with severe male factor infertility and 75 fertile control men. Thirty of the infertile patients had nonobstructive azoospermia, 30 had oligozoospermia and 30 had normozoospermia. Five of 30 were azoospermic, four of 30 were oligozoospermic and two of 30 were normozoospermic with Y chromosome microdeletions. The AZFc locus was the most frequently deleted region (64%). Ten cases with azoospermia, four cases with oligozoospermia and four cases with normozoospermia had chromosomal abnormalities. The 75 men with proven fertility were genetically normal. We conclude that various chromosomal

abnormalities and deletions of the Y chromosome can cause infertility; therefore, genetic screening is indicated for infertile patients.

Key words: Male infertility; Y chromosome microdeletion; Chromosomal abnormality; Azoospermia; Severe oligozoospermia

