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Chromosomal Abnormalities and Y Chromosome Microdeletions in Infertile Men With Varicocele and Idiopathic Infertility of South Indian Origin

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Various factors cause spermatogenesis arrest in men and, in a large number of cases, the underlying reason still remains unknown. Little attention is paid to determining the genetic defects of varicocele-related infertility. The objective of our present study was to investigate the chromosomal abnormalities and Y chromosome microdeletions in infertile men of South Indian origin with varicocele and idiopathic infertility. Metaphase chromosomes of 251 infertile men with varicocele and unexplained infertility were analyzed using Giemsa-Trypsin-Giemsa (GTG) banding and fluorescence in situ hybridization (FISH). The microdeletions in 6 genes and 18 sequence-tagged-sites (STS) in the Yq region were screened using polymerase chain reaction (PCR) techniques. Out of 251 infertile men, 57 (22.7%) men were with varicocele, of which 8.77% were azoospermic, 26.31% were severely oligozoospermic, 21.05% were mildly oligozoospermic, and 43.85% were oligoasthenoteratozoospermic (OAT), and 194 (77.29%), with idiopathic infertility, of which 51% were azoospermic, 13.40% were severely oligozoospermic, 19.07% were mildly oligozoospermic, and 16.4% were with OAT. Genetic defects were observed in 38 (15.13%) infertile individuals, including 14 (24.56%) men with varicocele and 24 (12.37%) men with idiopathic infertility. The frequencies of chromosomal defects in varicocele and idiopathic infertility were 19.3% and 8.76%, respectively, whereas Y chromosome microdeletions were 5.26% and 3.60%, respectively. Overall rate of incidence of chromosomal anomalies and microdeletions in 251 infertile men were 11.5% and 3.98%, respectively, indicating a very significant higher association of genetic defects with varicocele than idiopathic male infertility. Our data also demonstrate that, among infertile men with varicocele, severely oligozoospermic and OAT men with varicocele have higher incidences of genetic defects than mildly oligozoospermic and azoospermic men.

Key words: Genetic defects, male infertility, cytogenetic study, STS markers

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