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A Study of Y Chromosome Microdeletions in Infertile Indian Males

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ABSTRACT Male partners of infertile couples are known to frequently have abnormal semen parameters. Some of these cases are due to underlying genetic factors such as Y chromosome microdeletions, an abnormal karyotype or cystic fibrosis mutations. Y chromosome microdeletions generally cannot be detected by karyotyping. At our clinic we undertook a study of male partners of infertile couples to determine the frequency and common loci of Y chromosome microdeletions in India, using the PCR technique. We studied 100 patients mainly having azoospermia (AZ) or oligoasthenoteratozoospermia (OAT). Multiplex PCR analysis for 18 loci on the Y chromosome was carried out using commercially available kit (Promega Version 1.1). Y chromosome microdeletions were observed in 12/100 (12%) patients including 8/27 (29.63%) with azoospermia, 3/56 (5.35%) with oligoasthenoteratozoospermia and 1/7 (14.28%) with only asthenoteratozoospermia. All loci of the DAZ gene were deleted along with DYS237 and DYS236 from AZFd in 5/27 (18.52%) azoospermic males studied. The most commonly deleted loci were DYS240 in 11/12 (91.67%) and DYS219 in 7/12 (58.33%) patients with microdeletions. The use of ICSI in such patients can lead to transmission of Y chromosome microdeletions and subsequent infertility from father to son. Hence screening for Y chromosome microdeletions will help in the proper counseling and management of couples with male factor infertility.

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