



© *Kamla-Raj 2004*
PRINT: ISSN 0972-3757 ONLINE: 2456-6360

Int J Hum Genet, 4(3): 179-185 (2004)

A Study of Y Chromosome Microdeletions in Infertile Indian Males

**Arundhati S. Athalye, Prochi F. Madon, Nandkishor J. Naik, Dattatray J. Naik,
Smita S. Gavas, Suresh B. Dhumal, Vijay M. Bandkar,
Mahadev T. Kawle and Firuza R. Parikh**

*Department of Assisted Reproduction and Genetics, Jaslok Hospital and Research
Center, 15, Dr. G. Deshmukh Marg, Mumbai 400 026, Maharashtra, India
E-mail: prochimadon@hotmail.com*

KEYWORDS Male infertility; ICSI; semen analysis; PGD; AZF deletions; azoospermia; OAT

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.

[Home](#)

[Back](#)
