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JOURNAL ARTICLE

Three-generation evaluation of Y-chromosome microdeletion

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Sperm cells can be retrieved directly from the testis (testicular sperm extraction [TESE] procedure) and used for intracytoplasmic sperm injection (ICSI), circumventing underlying spermatogenetic defects. Thus, it is important that added information be available on the genetic defects in men undergoing TESE for the ICSI procedure and on the transmission of genetic factors associated with infertility to the offspring. We report a three-generation genetic analysis of a family with a case of male factor infertility. The proband, previously diagnosed as infertile, was physically examined and laboratory tested for gonadotrophic hormones, semen analysis, karyotype and Y-chromosome microdeletion screening in the blood and testis. The Y-chromosome microdeletion screening was performed by multiplex polymerase chain reaction with 20 Y-chromosome sequenced, tagged sites located at the Y chromosome. A microdeletion including the AZF-c region was detected in the azoospermic patient. His father, four brothers, and three offspring born after ICSI also underwent Y-chromosome microdeletion screening. The genetic analysis of the male members of the patient's family did not reveal similar microdeletions. The newborn male was found to bear a Y-chromosome microdeletion similar to that of his father. The fertilization capacity of the proband testicular microdeleted spermatozoa by the ICSI procedure is described. The transfer of the genetic defect raises the possibility that the son will have the same fertility problem as his father.

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