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Identification of the origin of marker chromosomes by two-color fluorescence *in situ* hybridization and polymerase chain reaction in azoospermic patients

C.L. Wei[1,2]*, J.L. Cheng[1]*, W.C. Yang[1], L.Y. Li[3], H.C. Cheng[3] and J.J. Fu[1,2,3]

[1]Key Laboratory of Epigenetics and Oncology,
Research Center for Preclinical Medicine, Sichuan Medical University,
Luzhou, Sichuan Province, China
[2]State Key Laboratory of Quality Research in Chinese Medicine,
Macau University of Science and Technology, Macau, China
[3]Insistitute of Reproduction and Stem Cell Engineering,
Xiangya School of Medicine, Central South University, Changsha,
Hunan Province, China

*These authors contributed equally to this study.
Corresponding author: J.J. Fu
E-mail: fujunjiang@hotmail.com / fujunjiang@lzmc.edu.cn

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ABSTRACT. Y chromosomal microdeletions at the azoospermia factor locus and chromosome abnormalities have been implicated as the major causes of idiopathic male infertility. A marker chromosome is a structurally abnormal chromosome in which no part can be identified by cytogenetics. In this study, to identify the origin of the marker chromosomes and to perform a genetic diagnosis of patients with azoospermia, two-color fluorescence *in situ* hybridization (FISH) and polymerase chain reaction (PCR) techniques were carried out. The marker chromosomes for the two patients with azoospermia originated

in the Y chromosome; it was ascertained that the karyotype of both patients was 46,X, ish del(Y)(q11)(DYZ3+, DXZ1-). The combination of two-color FISH and PCR techniques is an important method for the identification of the origin of marker chromosomes. Thus, genetic counseling and a clear genetic diagnosis of patients with azoospermia before intracytoplasmic sperm injection or other clinical managements are important.

Key words: Azoospermia factor; Azoospermia; Marker chromosome; Fluorescence *in situ* hybridization; Polymerase chain reaction

