Male infertility in Northeast China: Molecular detection of Y chromosome microdeletions in azoospermic patients with Klinefelter’s syndrome


Center for Reproductive Medicine, Center for Prenatal Diagnosis, First Hospital, Jilin University, Changchun, Jilin Province, China

Corresponding author: R.-Z. Liu
E-mail: lrz410@126.com

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ABSTRACT. The prevalence of microdeletions of azoospermia factor (AZF) among azoospermic Klinefelter’s syndrome (KFS) patients shows conflicting data. We aimed to detect this frequency in a Northeast Chinese population, and to investigate the possible association between AZF microdeletions and KFS by comparison with previous conflicting reports. Eighty men affected with KFS and a random healthy control group comprising 60 fertile men and women were recruited. AZF microdeletions were detected by multiplex polymerase chain reaction using 9 specific sequence-tagged sites. Karyotype analyses were performed on peripheral blood lymphocytes using standard G-banding. Finally, azoospermia was confirmed in 77 men affected with KFS and no AZF microdeletions were found. Karyotype analysis revealed 1 patient with karyotype 47,XXY,inv (9) (p11, q13), and 2 with mosaic karyotypes (46,XX/47,XXY and 46,XY/47,XXY). All other patients had karyotype 47,XXY. Review of the literature showed that these results
were similar to those of other regions of Northeast Asia, but differed from those obtained from Caucasian populations. Our results supported the proposal that AZF microdeletions and KFS result from separate genetic defects. The prevalence of AZF in azoospermic KFS patients varies among populations, and it might result from genetic drift or selective pressure. These results suggest that routine screening for classical AZF microdeletions among infertile azoospermic men with a 47,XXY karyotype might not be necessary in Northeast Chinese individuals. However, it remains imperative for patients considering assisted reproductive treatments, particularly for those with mosaic karyotypes.

**Key words:** Y chromosome; AZF microdeletion; Azoospermia; Klinefelter’s syndrome