Correlation between chromosomal polymorphisms and male infertility in a Northeast Chinese population

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ABSTRACT. The aim of this study was to evaluate the correlation between chromosomal polymorphisms and male infertility. The patients were diagnosed with azoospermia or oligospermiaby a semen analysis. Chromosomal analysis was performed on peripheral blood lymphocytes obtained from the patients, with standard G-banding and C-banding. Y chromosome microdeletions were detected by multiplex polymerase chain reaction (PCR) amplification. The parents of 35 polymorphic probands were also subjected to chromosomal analysis, and their detailed reproductive histories were surveyed. The frequency of autosomal polymorphisms did not differ significantly among the infertile patients and fertile control individuals. The frequency of the Yqh-variant increased with the decrease in sperm count; this appeared at a significantly higher frequency in the azoospermia group (57.2 *vs* 24.3 *vs* 0%). The results of PCR amplification indicated that 32.14% of the patients with Yqh ± had microdeletions in the Y chromosome. The parents of the probands with the same chromosomal polymorphisms as the probands (among the 35 recalled families) did not show any adverse

reproductive history. We observed no significant correlations between autosomal polymorphisms and male infertility. However, we observed a significant increase in the frequency of Yqh- in the azoospermic patients. This may be attributed to Y chromosome microdeletions, although the association between Y chromosome microdeletions and Y chromosome variants remains to be elucidated.

Key words: Chromosomal polymorphisms; Male infertility; Azoospermia; Pedigree analysis; Y chromosome microdeletion

