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# Cytogenetic and molecular analysis of infertile Chinese men: karyotypic abnormalities, Y-chromosome microdeletions, and CAG and GGN repeat polymorphisms in the androgen receptor gene

**T.T. Han, J. Ran, X.P. Ding, L.J. Li, L.Y. Zhang, Y.P. Zhang, S.S. Nie and L. Chen**

Key Laboratory of Bio-Resources and Eco-Environment,
Institute of Medical Genetics, Ministry of Education,
College of Life Science, Sichuan University, Chengdu, China

Corresponding author: T.T. Han / X.P. Ding
E-mail: han578388@gmail.com / brainding@scu.edu.cn

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**ABSTRACT.** Chromosome abnormalities, Y-chromosome microdeletions, and androgen receptor gene CAG and GGN repeat polymorphisms in infertile Chinese men featuring severe oligospermia and azoospermia were analyzed. Ninety-six fertile men and 189 non-obstructive infertile men, including 125 patients with azoospermia and 64 with severe oligozoospermia, were studied. Seventeen infertile men (9.0%) carried a chromosome abnormality. Twenty (10.6%) carried a Y-chromosome microdeletion. In the remainder of the patients and controls, GGN and CAG repeats were sequenced. Short GGN repeats ($n < 23$) appeared to be associated with defective spermatogenesis, with the number of GGN repeats strongly correlated with sperm counts. No significant difference in CAG repeats was found between patients and con-

 

trols, nor were CAG repeats correlated with sperm counts. However, for CAG repeats ranging between 24 and 25, there was a >2.5-fold risk (OR = 2.539, 95%CI = 1.206-5.344, $P < 0.05$) of severe oligospermia and azoospermia. Our results confirmed the significant role of chromosome abnormalities, Y-chromosome microdeletions, and GGN repeats in Chinese male infertility.

**Key words:** Glutamine and glycine repeat polymorphisms; Karyotype; AZF microdeletions; Azoospermia; Non-obstructive oligospermia

 