Genetic screening for patients with azoospermia and severe oligo-asthenospermia

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摘要

Abstract

In order to explore the genetic defects of patients with azoospermia or severe oligo-asthenospermia, screening examinations were carried out for the chromosome disorder and gene deletion of the Y chromosome for 220 male infertility patients. The present results show that the total prevalence of genetic defects is 23.6%, including 38 patients (28.4%) with chromosome disorder and 14 patients (16.8%) with gene deletion in the Yq arm. The most prevalent chromosome anomaly is 47XXY (Klinefelter's syndrome), which includes 18 cases of pure type and three cases of mosaic type. Variable autosomal translocations occurred in both the azoospermia group (5.2%) and the oligo-astheno-spermia group (5.8%) with similar prevalence. A total of 22 patients had deletions of the variable, interstitial portion of the Yq arm. These gene deletions are distributed not only inside the AZF region, but also outside of this region. The severity of deletions is not well correlated to the clinical testicular function of the patients. We conclude that chromosome disorder and gene deletions are the causative factors of patients with azoospermia and oligo-asthenospermia. Genetic screening should be a routine examination for them before the use of assisted-reproductive technologies.