

论著

无精子症、严重少精子症遗传缺陷的研究

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摘要 背景与目的: 探讨无精子症和严重少精子症患者的遗传缺陷与精子生成障碍的关系。材料与方法: 采用G显带技术对205例无精子症和39例严重少精子症患者进行外周血染色体核型分析, 采用多重聚合酶链式反应对其中染色体核型正常的36例无精子症和严重少精子症患者进行Y染色体上基因微缺失检测, 30例正常已生育的男子设为对照组。结果: 无精子症和严重少精子症患者中发现异常染色体核型74例, 异常核型发生率为30.33%(24.56%~36.10%), 正常对照组只发现1例异常核型, 占3.33%(0%~17%); 其中染色体核型正常的无精子症和严重少精子症患者发现Y染色体上基因微缺失3例, 缺失率为8.33%(2%~22%), 正常对照组无1例Y染色体基因微缺失。结论: 染色体核型异常和Y染色体微缺失均与无精子症和严重少精子症的发生有关。同时采用这两种遗传学筛查方法可以更为准确、有效地评价无精子症和严重少精子症患者的遗传缺陷, 更好地为患者提供病因诊断, 遗传咨询和治疗方案的选择。

关键词 [无精子症](#); [严重少精子症](#); [染色体核型](#); [聚合酶链反应\(PCR\)](#); [基因微缺失](#)

Research on Genetic Defects of the Patients with Azoospermia and Severe Oligospermia

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Abstract **BACKGRONUD & AIM:** To investigate the relationship between spermatogenesis disorder and genetic defects of patients with azoospermia or severe oligospermia. **MATERIAL AND METHODS:** G banding karyotype analysis of peripheral blood lymphocytes from 205 cases with azoospermia and 39 cases with severe oligospermia were performed and the multipolymerase chain reactions(PCR) for Y-chromosome microdeletion screening in the blood from 36 cases of azoospermia with normal karyotype were done. **RESULTS:** The incidence of abnormal chromosome karyotype was 30.33%(74 cases) in cases with azoospermia and severe oligospermia group and 3.33%(1 case)in the control group respectively. 3 cases with microdeletion in different segments of Azoospermia Factors(AZF)region on Y-chromosome were found in 36 cases of azoospermia with normal karyotype, and the microdeletion rate was 8.33%. No microdeletion in corresponding sites was discovered in the control group. **CONCLUSION:** Both of abnormal chromosome karyotype and Y-chromosome microdeletion are important to cause azoospermia and severe oligospermia. It is more accurate and effective in evaluating the genetic defect of the patients with azoospermia or severe oligospermia to combine two methods of karyotype analysis and microdeletion screening, which can offer the patients with etiologic diagnosis, genetic counseling and choices for therapeutic strategies.

Keywords [azoospermia](#) [oligospermia](#) [chromosome karyotype](#) [multipolymerase\(PCR\)](#) [gene microdeletion](#)

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