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研究报告

无精和严重少精症患者的遗传缺陷分析---附2例世界首报染色体异常核型

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摘要 对217例无精和严重少精症患者外周血淋巴细胞染色体核型进行分析，并采用聚合酶链反应对7例Y染色体结构异常患者的AZFc区进行检测。发现187例无精症患者中检出异常核型77例（41.18%）（其中46,XY,t(6;14)(p21;p13)，46,XY,t(8;12)(p21;q24)为世界首报核型），主要涉及染色体异常（数目异常和结构异常）；染色体异态（Y染色体异态和9号染色体臂间倒位）及46,XX性反转；30例严重少精症患者中检出异常核型4例（13.33%）（结构异常和46,XX性反转）。由此可见，性染色体数目和结构异常是精子发生障碍的主要原因，其次常染色体的某些断裂点也可能影响精子发生。AZFc区的缺失与否与精子发生也有直接关系。

关键词 无精症 严重少精症 染色体畸变 AZFc区

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Analysis on genetic defects of the patients with azoospermia and severe oligospermia---A report of 2 abnormal karyotypes of the first reported in the world

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Abstract

G banding karyotype analysis of peripheral lymphocytes in 217 patients with azoospermia or severe oligospermia were performed and the polymerase chain reactions(PCR) for AZFc of Y-chromosome in the blood from 7 cases with Y chromosome abnormality. The result is: out of 187 cases of azoospermia, 77 (41.18%)patients with abnormal chromosome(number and structural aberration); heteromorphic chromosome(Y chromosome polymorphisms and pericentric inversion of 9) and 46,XX sex reversal. Out of 30 cases of severe oligospermia, 4 patients with abnormal chromosome(structural aberration and 46,XX sex reversal). It is revealed that the aberration of sex chromosome causes the most serious spermatogenesis failure and some breakpoints of autosome may also affect spermatogenesis. The deletion of AZFc is also affect spermatogenesis.

Key words azoospermia severe oligospermia chromosome aberration AZFc region

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