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Y染色体单倍群与西南地区男性生精障碍的相关性

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摘要 男性不育中, 原发无精、少精是最为重要的因素之一, 核型异常和无精子症因子(Azoospermia factor, *AZF*)微缺失能解释部分原发无精、少精的原因, 然而还有许多致病因素尚不清楚。Y染色体作为男性特有的染色体, 与男性生殖系统的正常功能密切相关。文章主要对Y染色体单倍群这一分子遗传背景与男性原发无精、严重少精症之间是否存在相关性进行探讨, 为进一步探索原发无精、严重少精症的遗传学致病原因提供依据和可行的方向。采集265名生精障碍患者(原发无精症患者193名, 原发严重少精症患者72名)以及193名正常男性样本的外周血, 进行核型分析和AZF缺失分析, 以排除有此两类异常的样本。将经过筛选的样本进行Y染色体单倍群分析, 并对其单倍群分布情况进行统计分析。分析显示, 生精障碍组和对照组分别在D1*、F*、K*、N1*和O3* 上有显著性差异(P=0.032, 0.022, 0.009, 0.009, 0.017, <0.05)。Y染色体单倍群, 这一Y染色体遗传背景与男性原发生精障碍的发生有相关性。

关键词： Y染色体单倍群 原发生精障碍 无精子症因子(*AZF*)

Abstract： Idiopathic azoospermia and oligospermia are one of the most important reasons for male infertility. Abnormal karyotype and azoospermia factor (*AZF*) microdeletion are two widely acknowledged reasons, but the most causes remain unclear. Y chromosome, as the male-specific chromosome, is closely related to the development of male reproductive system. To understand better the etiology of idiopathic azoospermia and oligospermia, we investigated the possible association between Y-haplogroup distributions and susceptibility to idiopathic azoospermia and severe oligospermia. Pe-ripheral blood was collected from 193 men with normal reproductive history, 193 men with idiopathic azoospermia, and 72 men with idiopathic severe oligospermia. All the subjects underwent karyotyping and *AZF* deletion analysis to screen out those with *AZF* deletion and abnormal karyotype. The comparison of Y-haplogroup distribution between experimental group and control group was performed with SPSS V.18.0 software. Significant difference of Y-haplogroup distribution was observed in D1*, F*, K*, N1* and O3*(P=0.032, 0.022, 0.009, 0.009, 0.017, <0.05). The results suggest that Y chromosome haplogroup plays a important role in spermatogenic impairment.

Keywords： Y chromosome haplogroups, spermatogenic impairment, azoospermia factor(*AZF*)

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