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研究报告

严重寡精症ICSI精子供体的*DAZ*基因拷贝缺失研究

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摘要

*DAZ*基因拷贝缺失与人类的生精障碍有关。为了解中国正常生精男性和ICSI中严重寡精症精子供体*DAZ*基因拷贝缺失的分布, 探讨*DAZ*基因拷贝数检测在严重生精障碍精子供体遗传缺陷筛查中的意义, 本研究运用多重PCR和PCR-RFLP技术, 对128例严重寡精症ICSI精子供体和287个正常生精男性的*DAZ*基因缺失进行了研究。发现*DAZ1/DAZ2*、*DAZ3/DAZ4*和全部4个拷贝缺失等3种拷贝缺失类型, 其中全部4个拷贝缺失仅见于严重寡精症患者, 频率为11.7%; *DAZ1/DAZ2*缺失的频率在严重寡精症患者中显著高于正常男性(9.4% vs 2.8%, $P = 0.004$); 在严重寡精症患者中*DAZ*基因拷贝完全缺失与*DAZ1/DAZ2*缺失的总发生率为21.1%。*DAZ3/DAZ4*缺失的频率在两组人群中无显著差异(7.0% vs 3.8%, $P > 0.05$)。这些结果提示, *DAZ*基因全部拷贝缺失是严重寡精症患者生精障碍的常见遗传病因, 而*DAZ1/DAZ2*缺失则可能是一种高风险因素。鉴于上述*DAZ*基因缺失在严重生精障碍精子供体中较高的发生率, 在应用ICSI进行辅助生育前, 建议对严重寡精症的精子供体进行*DAZ*基因全缺失与*DAZ1/DAZ2*共缺失筛查, 以评估其男性后代患病的风险。

关键词 无精症缺失基因 基因缺失 严重寡精症 ICSI

分类号 R349.5 R544.1

Study on *DAZ* Gene Copy Deletion in Severe Oligozoospermia Sperm Donor for ICSI

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Abstract

<P><SPAN lang=EN-US style="FONT-SIZE: 10pt; FONT-FAMILY: 'Times New Roman'; LETTER-SPACING: 0.1pt; mso-fareast-font-family: 宋体; mso-ansi-language: EN-US; mso-fareast-language: ZH-CN; mso-bidi-language: AR-SA; mso-font-kerning: 1.0pt"><FONT face=Verdana><FONT face=Verdana><FONT face=Verdana>Deletion of <EM>DAZ</EM> gene copies is related to spermatogenesis impairment. To investigate the distribution of <EM>DAZ</EM> gene copy deletions among Chinese men, we analyzed <EM>DAZ</EM> gene deletions by multiplex polymerase chain reaction (multi-PCR) and polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) in 128 infertile patients with severe oligozoospermia selected as semen donors for intracytoplasmic sperm injection (ICSI) and 287 normospermic men. Three patterns of <EM>DAZ</EM> gene deletions, namely <EM>DAZ1/DAZ2</EM> deletion, <EM>DAZ3/DAZ4</EM> deletion and complete deletion of all 4 <EM>DAZ</EM> copies, were found in the present study. Complete deletion of the entire <EM>DAZ</EM> family of genes was only present in 11.7% of severe oligozoospermic patients. The frequency of <EM>DAZ1/DAZ2</EM> deletion was significantly higher in severe oligozoospermic patients than that in the controls (9.4% vs 2.8%, p = 0.004. The total frequency of complete DAZ deletion and <EM>DAZ1/DAZ2</EM> deletion was 21.1%. No significant difference in the frequency of <EM>DAZ3/DAZ4</EM> deletion was observed between the patient and control group (7.0% vs 3.8%, <EM>p </EM>> 0.05). These results suggest that complete <EM>DAZ</EM> deletion is a frequent genetic cause of severe oligozoospermia, and <EM>DAZ1/DAZ2</EM> deletion is a high risk factor for the

disease. Thus, it is necessary to screen the two deletion patterns of <EM>DAZ </EM>genes in severely oligozoospermic sperm donors before ICSI during assisted reproduction.</FONT></FONT></FONT></SPAN></P>

Key words DAZ gene gene deletion severe oligozoospermia ICSI

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