

六例性反转综合征患者的分子遗传学分析*

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摘要 对六例性反转综合征患者(3例 XX 男性)(3例 XY 女性)用Y-特异性DNA探针进行了Southern印迹杂交分析,并用PCR技术扩增了SRY基因部分序列。结果表明,1例XX男性缺乏源于Y染色体的杂交信号,也无SRY基因;其余2例XX男性和3例XY女性都检测到Yp-DNA序列和SRY基因。这对进一步阐明性反转综合征的病因和SRY基因的作用机制具有重要意义。

关键词 [性反转综合征,性别决定,SRY 基因](#)

分类号

Molecular Genetic Analysis on Six Cases of Sex Reversal Syndrome*

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Abstract

Southern blot hybridization using Y-specific DNA probes and electrophoretic analysis of the SRY fragment polymerase chain reaction (PCR) have been performed in six cases of sex reversal syndrome (three 46,XX males and three 46,XY females). The result showed that the Y-specific DNA hybridization signal and SRY sequence were absent in one XX male and present in the remaining cases. This Southern blot hybridization using Y-specific DNA probes and electrophoretic analysis of the SRY gene fragment amplified through polymerase chain reaction indicates that testis development of this XX male was initiated in the absence of the SRY gene.

Key words [Sex reversal syndrome](#) [Sex determination](#) [SRY gene](#)

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