遗传学报 1993 20 (1): 7-11 ISSN: 0379-4172 CN: 11-5450/R

一个22p+家系的临床及分子细胞遗传学研究

刘淑敏，高春生，胡原，刘妙良，程在玉

中国医学科学院基础医学研究所，北京 100005

收稿日期 修回日期 网络版发布日期 接受日期

摘要 本文对一例男性性腺发育不全伴有22p+标记染色体的患者及其家庭成员进行了分子细胞及临床细胞遗传学研究。结果表明，该家系中共有6名成员有22p+染色体，它们来源于先证者的外祖母。标记染色体的p+部分几乎与22q等大，C-带呈深染，G-、R-带在p+中间分别可见一条较窄的浅、深带型，Ag-显带在p+末端见到较大的银染区，部分p+出现双NOR，核型分析未见其它D、G组染色体短臂上平均数的3.9倍。家系研究表明，在6例22p+携带者中，2例女性具有多次自然流产史，4例男性中除一例年龄12岁未见明显性腺异常外，其余3人均有不同程度性腺或外生殖器异常，结合文献，我们认为此家系中22p+可能与上述异常表型存在着一定的关系

关键词 性腺发痛不全,标记染色体,近端着丝粒染色体,核仁组织者区,染色体原位杂交

分类号

Molecular and Clinical Cytogenetic Studies of a Family with a 22p+ Marker Chromosome

Liu Shumin1), Gao Chunsheng, Hu Yuan 1),Lin Miaoliang, Cheng Zaiyu

Institute of Basic Medicine, Chinese Academy of Medical Sciences, Beijing 10005

Abstract

A male with gonadal dysgenesis and a 22p+ was observed. Molecular and Clinical Cytogenetic studies have been carried out on the members of the faily. The results showed that there was a 22p+ marker chromosome transmitted from the maternal grandmother of the proband to 6 members of this family. Its short arm showed a homogeneously dull stained region in C-banded preparations and a narrow dark or light stained band in R-, G-banded preparations respectively. A large Ag-band or double NORs was also observed on p+. The chromosomal in situ hybrididation with tritium labelled rRNA gene probe demonstrated that the distribution of the silver grains was along the entire p+ of the marker chromosome. The number of silver grains on the short arm of the p+ was 3.9 times as that of any other normal acrocentric chromosomes. Two cases of female with repeated spontaneous abortions and 2 cases of male with gonadal dysgenesis were found by family study. Our studies, combining with previous literatures. suggested that these abnormalities were probably in association with p+ marker chromosome.

Key words Gonadal Dysgenesis Acrocentric Chromosome Marker Chromosome Nucleolar Organizing Region Chromosomal in Situ Hybridization rRNA Gehes Probe

DOI:

通讯作者