

# 一例t(Y;15)并t(14;21)复合易位的细胞与分子遗传学研究\*

石玉平, 马绍武, 王玉梅, 高春生, 叶丽珍, 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** 本文对一例生育过先天愚型儿的个体进行了细胞与分子遗传学研究。发现先证者拥有t(14;21)及一个短臂增大变异的15号标记染色体。通过G-显带、C-显带、Q-显带、硝酸银染色及Y染色体长臂异染色质区特异探针pY3.4对先证者基因组DNA的斑点杂交和中期染色体的原位杂交,证实变异部分由Y染色体长臂异染色质区易位所形成,从而排除了巨大随体的存在或其他染色体参与重排形成变异的可能性。结果表明,常规显带与染色体原位杂交等分子生物学技术相结合对于鉴定人类染色体的微小结构畸变具有重要的应用价值。

**关键词** [复合易位,标记染色体,原位杂交,斑点杂交](#)

分类号

## Cytogenetic and Molecular Genetic Study on a Female with Complex Translocation of t(Y;15) and t(14;21)\*

Shi Yuping, Ma Shaowu, Wang Yumei, Gao Chunsheng, Ye Lizhen, Cheng Zaiyu

Institute of Basic Medical Sciences, Chinese Academy of Medical Sciences, Beijing 100005

### Abstract

A 27-year-old female having a baby with Down's syndrome was found to have a translocation t(14;21) and an acrocentric marker chromosome 15. The short arm of the marker was darkly stained in both G- and C-banded preparations, and had the normal Ag-NOR, but showed a homogeneously stained region in the distal part of the marker. By means of QM staining, the variant was found fluorescent, and Y-body like was found in about 9% interphase nuclei, which suggested that there might exist Y chromosome material in genomic DNA of proband. Dot blot hybridization using DNA probe pY3.4 derived from the heterochromatic region on the long arm of Y chromosome demonstrated that the proband carried Y chromosome material in her genome. Further, by in situ hybridization with the same probe, it showed that the variant of the marker had specific autoradiographic silver grains. So, the karyotype of the proband indicated that the combination of conventional banding analysis with chromosome-specific probe was of great significance in studying the microaberration of human chromosome.

**Key words** [Complex translocation](#) [Market chromosome](#) [Chromosomal in situ hybridization](#) [Dot blot hybridization](#)

DOI:

通讯作者

### 扩展功能

#### 本文信息

- ▶ [Supporting info](#)
- ▶ [PDF\(1176KB\)](#)
- ▶ [\[HTML全文\]\(0KB\)](#)
- ▶ [参考文献](#)

#### 服务与反馈

- ▶ [把本文推荐给朋友](#)
- ▶ [加入我的书架](#)
- ▶ [加入引用管理器](#)
- ▶ [复制索引](#)
- ▶ [Email Alert](#)
- ▶ [文章反馈](#)
- ▶ [浏览反馈信息](#)

#### 相关信息

- ▶ [本刊中包含“复合易位,标记染色体,原位杂交,斑点杂交”的相关文章](#)
- ▶ [本文作者相关文章](#)

- [石玉平](#)
- [马绍武](#)
- [王玉梅](#)
- [高春生](#)
- [叶丽珍](#)
- [程在玉](#)