

## 4号染色体部分重复和缺失一例<sup>1)</sup>

萧广惠 陈争 赖伏英 吕冰清

湖南医学院附属第一医院中心实验室;长沙 湖南医学院附属第一医院神经内科;长沙

收稿日期 修回日期 网络版发布日期 接受日期

**摘要** 本文报道一例4号染色体部分重复和缺失的男婴。患儿发育迟缓,智力低下,体查多种畸形。其G显带核型为“6, XY, rec (4), dup c1, inv(4)(p16q31)。患儿父亲及祖母均为inv(4)(p16q31)的携带者,重复一缺失染色体源自患儿父亲的减数分裂重组。

**关键词**

**分类号**

## Partial Duplication and Deletion of Chromosome 4 in an infant of a Carrier of Pericentric Inversion of Chromosome 4

Xiao Guanghui, Chen Zheng, Lai Fuying, Lu Bingqing

(Central Laboratory of First Affiliated Hospital, Hunan Medical College, Changsha)(Department of Internal Neurology of First Affiliated Hospital, Hunan Medical College, Changsha)

### Abstract

A 9 months male infant with both physical and mental retardation and many deformities is found to be a karyotype of 46, XY, rec(4), dup q, inv(4)(p16q31). His father and grandmother are both the carriers of inv(4)(p16q31). The recombinant may result from the paternal meiosis.

### Key words

DOI:

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