

家族性平衡7/9易位与9号染色体部分三体患儿

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摘要 染色体部分三体的儿童往往伴有智力迟纯和其它表型异常。本文报道的一例部分三体的女性患儿也是如此。患者的智力低下, 面型异常。根据家系研究, 发现她的祖母和小弟是平衡易位杂合体, 从而知道她是平衡易位的不平衡分离产物。据我们已查阅的资料同样的病例国外尚未见有报告。

关键词

分类号

A Family with Balanced 7/9 Translocation and Unbalanced Offspring

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Abstract

A female patient with partial trisomy of chromosome 9 is described. The patient showed congenital heart disease, ocular hypertelorism, depressed nasal bridge, low-set ears and severe mental retardation. Cytogenetic analysis revealed that her paternal grandmother had a balanced translocation between the short arms of chromosomes 7 and 9[t(7;9) (p22; p13)]. Presumably the patient was the unbalanced segregational product of the balanced translocation carrier and hence the translocation 7/9 might have been transmitted at least through three generations.

Key words

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