

论著

人脑星型细胞瘤神经细胞黏附分子基因突变分析

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摘要

目的: 了解神经细胞黏附分子 (neural cell adhesion molecule, NCAM) 基因突变在人脑星型细胞瘤发生中的作用。方法: 对NCAM基因的外显子进行聚合酶链反应-单链构象多态性分析 (polymerase chain reaction-single strand conformation polymorphism, PCR-SSCP), 找出可疑突变, 回收PCR产物纯化测序及序列分析, ORF finder 软件分析蛋白质序列。结果: 43例星型细胞瘤中有1例胶质母细胞瘤NCAM 7号外显子1 126号核苷酸由A颠换为C, 导致369号氨基酸由赖氨酸变为谷氨酰胺, 该病人1年后死于复发。结论: NCAM基因点突变导致的蛋白质结构改变在星型细胞瘤的发生中可能有重要意义。

关键词 [星型细胞瘤](#); [神经细胞黏附分子](#); [点突变](#)

分类号

Mutation analysis of neural cell adhesion molecules in human astrocytoma

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Abstract

Objective To identify the function of neural cell adhesion molecule (NCAM) mutation in the genesis of human glioma. Methods Mutations were found through polymerase chain reaction-single strand conformation polymorphism, and then the changed DNA fragments were purified and multiplied and sent to Shanghai for sequencing. Blast and ORF finder were used to find out the amino changes in NCAM. Results An A-C transversion was found at position 1 126 in NCAM's 7 exon in a patient with glioblastoma from 43 astrocytoma. Conclusion Structural change in the protein caused by point mutation may be the reason for tumorigenesis of astrocytoma.

Key words [astrocytoma](#) [neural cell adhesion molecule](#) [point mutation](#)

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