

论文

成人急性淋巴细胞白血病ETV6基因重排分析及其临床意义

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摘要:

目的 检测成人急性淋巴细胞白血病(ALL)ETV6基因重排情况并探讨其临床意义。方法 用Split-signal FISH技术检测32例初治及复发的成人急性淋巴细胞白血病患者骨髓样本ETV6基因重排情况, 阳性样本行巢式RT-PCR证实融合基因形成。结果 FISH检测出1例ETV6易位样本, 常规染色体检查为正常核型, RT-PCR证实形成ETV6-RUNX1融合基因。该患者为复发难治性白血病, 复发后产生耐药, 于确诊9个月后死亡。结论 FISH检测ETV6基因重排较常规染色体核型分析更敏感。成人ALL中ETV6-RUNX1表达率低, 可能与ALL复发患者的不良预后有关。

关键词: 基因, ETV6; 白血病, 淋巴样; 成人; 荧光原位杂交

ETV6 gene rearrangements in adult acute lymphoblastic leukemia and their clinical significance

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Abstract:

Objective To test the rearrangements of the ETV6 gene in adult patients with acute lymphoblastic leukemia (ALL) and analyze the clinical significance of the rearrangement. Methods Bone marrow was obtained from 25 ALL patients for the test. Split-signal fluorescence in situ hybridization(Split-signal FISH)was used for testing ETV6 rearrangements. Nested Reverse transcription polymerase chain reaction(Nested RT PCR)was used for confirmation if fusion was found through Split-signal FISH. Results The ETV6 gene was observed as broken and rearranged in one male patient who showed as a normal karyotype by conventional cytogenetic analysis. The arrangement was subsequently confirmed as expression of the ETV6-RUNX1 fusion gene identified by RT-PCR. The patient was a refractory and relapsed acute lymphocytic leukemia patient and resistant to re-induction therapy. Conclusion FISH is more sensitive than the conventional cytogenetic analysis to detect the ETV6 gene rearrangement. Our study indicates that the ETV6 RUNX1 fusion gene, which is caused by ETV6 rearrangement, may be an indication of a poor outcome in adult patients with ALL.

Keywords: Gene, ETV6; Leukemia,Lymphoid; Adult; Fluorescence in situ hybridization

收稿日期 2011-11-07 修回日期 网络版发布日期

DOI:

基金项目:

济南市科技发展基金资助项目(200905045)

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