CDKN2A基因突变与晚期非小细胞肺癌患者临床病理特征及预后的相关性分析

《现代肿瘤医学》[ISSN:1672-4992/CN:61-1415/R] 期数: 2019年07期 **页码:** 1154-1157 **栏目:** 论著（胸部肿瘤） **出版日期:** 2019-02-28

Title: **The analysis of CDKN2A gene mutation with clinicopathological features and prognosis in advanced non-small cell lung cancer patients**

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关键词: CDKN2A; 突变; 预后; 肺癌

Keywords: CDKN2A; mutation; prognosis; lung cancer

分类号: R734.2

DOI: 10.3969/j.issn.1672-4992.2019.07.013

文献标识码: A

摘要: 目的：评估细胞周期依赖性激酶抑制基因（CDKN2A）突变与晚期非小细胞肺癌（NSCLC）患者的临床病理特征和预后的关系。方法：采用第二代测序技术筛选肿瘤标本中的CDKN2A基因突变。卡方检验分析CDKN2A基因突变与晚期NSCLC临床病理特征之间的相关性。Logistic回归分析CDKN2A基因突变与临床一线治疗评效之间的相关性。Kaplan-Meier曲线和COX模型评估患者生存。结果：NSCLC患者CDKN2A基因突变率为3.81%（8/210）。CDKN2A基因突变在鳞状细胞癌患者中更常见（P=0.005）。多数CDKN2A基因突变型患者一线治疗评效是疾病进展（PD），而野生型患者多为疾病控制（DCR）（P=0.012）。CDKN2A基因突变型患者的中位生存时间（OS）明显短于野生型患者（19.1 vs 42.8个月，P=0.010），并且突变型患者的无进展生存时间（PFS）也明显缩短（3.5 vs 9.7个月，P=0.001）。多因素分析显示CDKN2A基因突变、T4期、淋巴结转移和ECOG高评分是影响晚期NSCLC患者生存的独立危险因素。结论：CDKN2A基因突变对晚期NSCLC患者的临床病理特征和预后有重要影响。CDKN2A基因突变患者的生存期明显缩短。

Abstract: Objective：To evaluate the association of CDKN2A gene mutation with clinicopathological features and prognosis in advanced non-small cell lung cancer（NSCLC) patients.Methods：CDKN2A gene mutation was screened in tumor specimens using second generation sequencing technology.The correlation between CDKN2A mutation and clinicopathological characteristics was analyzed by chi-square test.The correlation between CDKN2A mutation and clinical first-line evaluation was analyzed by Logistic regression analysis.The survival of patients was assessed by using Kaplan-Meier curve and COX model.Results：CDKN2A gene mutation was 3.81%（8/210) in NSCLC patients.Patients harboring CDKN2A mutation were more commonly observed in squamous carcinoma patients（P=0.005).Most of patients with CDKN2A mutation were progression disease（PD) in the assessment of first-line treatment efficacy，while the patients with CDKN2A wild type were disease control rate（DCR)(P=0.012).The median survival time（OS) was significantly shorter in patients with CDKN2A mutation than in wild-type patients（19.1 vs 42.8 months，P=0.010).The progression-free survival time（PFS) was also significantly shorter in the mutant patients（3.5 vs 9.7 months，P=0.001).Multivariate analysis showed that CDKN2A mutation，T4 phase，lymph node metastasis，and ECOG high score were independent risk predictors for the survival of advanced NSCLC patients.Conclusion：CDKN2A gene mutation had an important influence on the clinicopathological features and prognosis of advanced NSCLC patients.The patients with CDKN2A mutation had more poor survival.

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备注/Memo: National Natural Science Foundation of China(No.81673025）；国家自然科学基金（编号：81673025）

更新日期/Last Update: 2019-02-28