

中国肿瘤临床 2012, Vol. 39 Issue (11): 785-787 DOI: doi:10.3969/j.issn.1000-8179.2012.11.008

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甲状腺乳头状癌BRAFV600E突变分析

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BRAFV600E Mutation in Papillary Thyroid Carcinoma

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摘要

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摘要 检测BRAFV600E突变在甲状腺乳头状癌(PTC)中的发生情况, 分析BRAFV600E突变与临床各病理参数以及与合并桥本氏甲状腺炎(HT)和结节性甲状腺肿的关系。方法: 天津医科大学附属肿瘤医院2011年3月至2011年8月所收治的临床考虑甲状腺癌患者112例, 术中取部分新鲜肿瘤组织, 送基因诊断室检测BRAFV600E突变情况, 其中30例患者同时取部分正常甲状腺组织进行检测对照。结果: 112例患者病理结果显示110例为PTC, 2例为结节性甲状腺肿。其中BRAFV600E突变在110例PTC的突变率为62.7%, 不存在于结节性甲状腺肿及正常甲状腺组织。年龄≤30岁PTC患者8例, 突变率为25.0%; 30~60岁患者86例, 突变率为62.8%; ≥60岁患者16例, 突变率为81.2%, 差异有统计学意义($P=0.027$)。BRAFV600E突变与其他临床病理参数间的差异无统计学意义。合并HT的PTC患者40例, 突变率42.5%, 未合并HT的PTC患者70例, 突变率74.3%, 差异有统计学意义($P=0.001$)。合并结节性甲状腺肿的PTC患者61例, 突变率72.1%; 未合并结节性甲状腺肿的PTC患者49例, 突变率51.0%, 差异有统计学意义($P=0.023$)。结论: PTC的BRAFV600E突变率可能与种族差异有关。BRAFV600E突变率可能与患者年龄构成比有一定的相关性。合并HT的PTC BRAFV600E突变率低, 而合并结节性甲状腺肿的PTC BRAFV600E突变率高。

关键词: 甲状腺乳头状癌 突变 BRAF 结节性甲状腺肿 桥本氏甲状腺炎

Abstract. To detect BRAFV600E mutation in papillary thyroid carcinoma (PTC); to analyze the relationship between BRAFV600E mutation and clinicopathologic parameters in PTC; and to investigate BRAFV600E mutation in PTC coexisting with Hashimoto's thyroiditis or nodular goiter. Methods: DNA was extracted from the fresh thyroid tumor tissues of 112 patients and from normal tissues of 30 patients who were treated in our institution from March 2011 to August 2011 and diagnosed with thyroid carcinoma before surgery. BRAFV600E mutation was detected via polymerase chain reaction and DNA sequencing assays. Clinical data were reviewed and evaluated using the SPSS17.0 statistical software package. Results: The pathology showed that the 112 Chinese patients consisted of 110 cases of PTC and 2 cases of nodular goiter. The presence of BRAFV600E mutation was found in 69 patients with PTC (62.7%); the mutation was detected only in PTC and not in the 2 patients with nodular goiter as well as in the 30 normal tissues near the tumor. A significant difference in the BRAFV600E mutation rates in the ≤ 30, 30-60, and ≥ 60 age groups (25.0%, 62.8%, and 81.2%, respectively) was found. However, statistical data did not show any correlation between BRAFV600E mutation and other clinicopathologic parameters in PTC. The BRAFV600E mutation rate in PTC coexisting with Hashimoto's thyroiditis was 42.5%, whereas that in PTC coexisting with nodular goiter was 72.1%. These results both showed a significant difference with PTC not coexisting with benign thyroid lesion. Conclusion: Differences in genetic backgrounds might explain the differences in the BRAFV600E mutation rates. The rate obtained in the study is higher than the results in western countries but lower than that in Korea. The BRAFV600E mutation rates are different in different age groups. The rate obtained in PTC coexisting with Hashimoto's thyroiditis is low, whereas a high rate was obtained in PTC coexisting with nodular goiter.

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