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研究报告

*EGFR*基因在非小细胞肺癌、乳腺癌中突变的研究

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摘要

表皮生长因子受体(*EGFR*) 基因酪氨酸激酶域体细胞突变与非小细胞肺癌(NSCLC)患者对酪氨酸激酶抑制剂吉非替尼敏感性密切相关。文章分析和检测本院75例非小细胞肺癌、10例乳腺癌患者石蜡包埋标本*EGFR*基因突变状况。采用PCR技术进行*EGFR*基因19和21外显子突变分析。结果显示：75例NSCLC患者中有13例(13/75，17.33%)酪氨酸激酶域存在体细胞突变。其中7例(7/75，9.33%)为19外显子缺失突变，6例 (6/75，8%)为 21外显子替代突变(2573T>G，L858R)。病理分型显示，腺癌突变率高于其他几种类型NSCLC。乳腺癌患者均为免疫组化HER-2阳性女性，*EGFR*基因的19、21外显子中未见突变发生。中国非小细胞肺癌患者总突变率高于高加索人种，女性患者较男性患者突变率高，提示肺腺癌的患者突变率高可能在吉非替尼的治疗中获益。

关键词 非小细胞肺癌 表皮生长因子受体 吉非替尼 突变

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Epidermal growth factor receptor mutation in non-small cell lung cancer and breast cancer

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Abstract

<P> Somatic mutation in the tyrosine kinase (TK) domain of the epidermal growth factor receptor (<EM>EGFR</EM>) gene is associated with the sensitivity of non-smal cell lung cancer (NSCLC) to TK inhibitor Gefitinib. Mutational analysis for <EM>EGFR </EM>exons 19 and 21 was performed in 75 NSCLC and 10 breast cancer patients. All patients had not received treatment of Gefitinib. Somatic mutations in TK domain of <EM>EGFR </EM>were identified in 13 of the 75(13/75, 17.33%) patients, including 7 cases of in-frame deletion in exon 19 (7/75, 9.33%) and 6 cases of amino acid substitution (2573T>G, L858R) in exon 21 (6/75, 8%) . No other mutations were found in 10 breast cancer patients who stained positive for HER2 immunhistochemistry. Adenocarcinoma has a higher rate of mutations than several other types of NSCLC, the mutations occurring more fre-quently in female patients. <EM>EGFR</EM> mutation rate in Chinese NSCLC patients was higher than that in Caucasians. Our data indicated that Chinese adenocarcinoma patients could benefit from TK inhibitor Gefitinib.</P>

Key words non-small cell lung cancer(NSCLC) epidermal growth factor receptor (EGFR) gefitinib mutation

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