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Molecular Diagnosis of Lung Cancers

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Abstract

Recent advances in cancer diagnosis have seen a rapid and accurate diagnostic test. Initially used as research tools the molecular diagnostic tests have been applicable in the clinical scenario.

Purpose: Current study focuses on the significant approach of detection of epidermal growth factor receptor (EGFR) mutations in exon 18-21 and anaplastic lymphoma kinase gene (ALK) rearrangement analysis using molecular methods such as pyrosequencing and florescence *in-situ* hybridization (FISH) on formalin fixed paraffin embedded (FFPE) from tissue biopsy and cell blocks of 267 lung cancer patients.

Methods: In this study, FISH using a break-apart probe for the ALK gene was performed on formalin fixed paraffin-embedded tissue to determine the incidence of ALK rearrangements and hybridization patterns in 267 patients with a referred diagnosis of non-small cell lung cancer. EGFR mutations detection was performed on 267 samples by using Pyrosequencing.

Results: Among the 267 patients 11 were positive for ALK-1 gene rearrangement. 260 of the 267 cases were confirmed of adenocarcinoma and 7 of the 267 cases were squamous cell carcinoma. EGFR mutations were detected in 55 patients. The most common mutations found were exon 19 (56.36%), exon 21 (27.27%), exon 20 (3.63%) and exon 18 (9.09%). The influence of gender, non-smoking, and histological type on the EGFR mutations showed increase in female group.

Conclusions: Mutation detection testing is mandatory in lung carcinomas for the diagnosis and management of NSCLC.

Keywords: EGFR mutation analysis, Molecular diagnosis, ALK gene rearrangement

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Introduction

Lung cancer is the leading cause of cancer-related deaths in the in the world. Histologically, the World Health Organization (WHO) classifies lung cancer into non-small cell lung cancer (NSCLC, 85%) and small-cell lung cancer (15%). Depending upon the stages of the disease, there are four treatment strategies, including surgery, radiation therapy, chemotherapy, and/or targeted therapy. The use of targeted kinase inhibitors is dependent on the targets such as epidermal growth factor receptor (*EGFR*) or activating chromosomal fusions involving the *ALK* kinase, which are effective if there is an association of the molecular alterations in the tumors [1]. Therefore, molecular tests are routinely performed to identify mutations in oncogenes in lung cancer, including *EGFR* and *ALK*. Molecular tests help in identifying those patients who respond to targeted therapy; and reduce side effects of ineffective treatments. The use of tyrosine kinase inhibitors (TKI), gefitinib or erlotinib, requires confirmation of somatic activating *EGFR* mutation.

ALK gene rearrangements have been reported in 2% to 13% of patients with NSCLC [1]. *ALK*-rearranged tumors typically lack *EGFR* and *KRAS* mutations. Recent studies have demonstrated that lung cancers harboring ALK rearrangements do not respond to EGFR-specific tyrosine kinase inhibitors, but benefit from *ALK* kinase (i.e., crizotinib).

Now-a-days along with the routine histology testing molecular tests have been used frequently in diagnosis. However, some of