

A comparison of bronchofiberscopic washing cytology and FFPE tissue in the analysis of EGFR mutations in advanced NSCLC

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Background: In the treatment of advanced NSCLC, EGFR mutation status is one of the most predictive factors for the efficacy of EGFR tyrosine kinase inhibitors, and the evaluation of EGFR mutation status using the formalin-fixed paraffin-embedded (FFPE) tissue has been widely used for this analysis throughout the world. However, whether bronchofiberscopic brushing (BB) cytology samples can be used as an alternative for FFPE samples in the analysis of EGFR mutations is unknown. Therefore, in the current study, we compared the freeze stock solution of BB cytology with FFPE for the determination of EGFR mutation status in a large sample set. **Methods:** In diagnostic BFS examinations, after curetting or brushing and biopsy to target lesions, subsequent bronchial washing by saline was performed. Thereafter, the saline fluid in which the forceps were washed and the bronchial washing fluid were mixed in a sterilized tube and were immediately frozen in a -20°C freezer. EGFR mutation testing for both BB cytology and FFPE was performed using high-sensitivity PCR (BML, PCR-Invader). **Results:** A total of 659 BFS examinations were performed from Aug 2010 to Aug 2012 in our hospital. The BB cytology samples of 437 suspected cases of lung cancer were successfully obtained. Of these, 68 cases that were pathologically confirmed as adenocarcinoma based on both BB cytology and FFPE samples were analyzed in this study. EGFR mutations were identified in 32 cases, while the remaining 36 cases had wild-type EGFR. In 66 of 68 cases, the results of EGFR mutation status were the same for BB cytology and FFPE samples, and the kappa coefficient was 0.94. In one case, an exon-18 mutation was detected only by BB cytology sample. In another case an exon-21 mutation was detected only by FFPE sample. In 29 of 30 cases of EGFR mutation, the mutation site was the same in both samples. The kappa coefficient was 0.92. **Conclusions:** This is the largest genetic study to date demonstrating a head-to-head comparison of BB cytology and FFPE samples for the evaluation of EGFR mutations. Both methods showed high reliability and concordance using high-sensitivity PCR. BB cytology is considered a simple, rapid method and represents an effective alternative for FFPE in EGFR mutation testing.