Introduction: Advances in molecular biology are improving the understanding of non-small cell lung cancer (NSCLC) and changing the approach to treatment. Targeted therapy is available for the subgroup of NSCLC patients with specific mutations of the Epidermal Growth Factor Receptor (EGFR) tyrosine kinase domain. EBUS-TBNA is increasingly used for diagnosis and staging of NSCLC. The reported feasibility of molecular testing using EBUS-TBNA specimens has varied from 70 to 100%, when specimens are acquired with rapid on-site evaluation (ROSE) by a cytopathologist. The minimum number of tumor cells required for successful mutation testing, and procedural factors that may affect specimen quality, remain unclear. The aim of this study was to assess the feasibility of EGFR testing using EBUS-TBNA samples acquired in the absence of ROSE.

Methods: This was a prospective observational study. Demographic and procedural data was collected from consecutive patients who underwent EBUS-TBNA for diagnosis and/or staging of lung cancer from September 2012 to September 2013. Specimens were processed using liquid cytology, with cell block preparation in the presence of a visible cell pellet after centrifugation. EGFR mutation analysis was performed at the request of the treating physician, using Taqman quantitative PCR following cell block macrodissection. Tumor cell count, percentage of tumor cells and tumor cell volume were examined in each specimen. The final result of EGFR mutation analysis was recorded.

Results: 193 patients underwent EBUS-TBNA for diagnosis and/or staging of lung cancer (22G needle); 29 patients were diagnosed with adenocarcinoma and 2 patients were diagnosed NSCLC not otherwise specified. EGFR testing was requested in 16 of 31 patients. In 15 of 16 patients (93.7%) EBUS-TBNA specimens from a single lymph node station were found to be adequate for EGFR testing. A median of 3.5 needle passes per lymph node station tested (range 2-5) had been performed. In 13 of the 16 EBUS-TBNA specimens used for molecular analysis, tumor cell counts were above 400 cells. The minimum tumor cell count that allowed successful EGFR testing was 100 cells. Average tumor cell population among nucleated cell was 63%. The median tumor volume over the entire cut surface area of the cell block was 45%. In 2 of 16 patients (12.5%), EGFR mutations were detected (exon 19 in one and exon 21 in the other).

Conclusion: When performed in the absence of ROSE, EBUS-TBNA provides an adequate specimen for EGFR mutation testing in a majority of patients in whom it is requested.