Rapid EGFR Mutation Testing in Lung Cancer Samples: The Idylla™ System

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Introduction
Test for possible mutations in EGFR exons 18, 19, 20, and 21 is the current standard of care in advanced non-small cell lung cancer. Moreover, EGFR gene mutations should be obtained quickly given the rapid tumor progression and availability of effective targeted therapy. However, lack of time or tissue for molecular testing are among the main reasons why patients are often deprived of quick access to the proper targeted therapies.

Methodology
Idylla™ is a fully integrated and automated molecular diagnostics platform that combines speed and ease-of-use with high sensitivity and high multiplexing capabilities. Moreover, it overcomes the current problem of lack of tissue, and the time-consuming step of processing FFPE tissue samples. After insertion of a single FFPE slice into the cartridge, the complete process of sample liquefaction, nucleic acid preparation, real-time PCR, data analysis and reporting is fully automated. The Idylla™ EGFR Mutation Test allows the sensitive detection of 51 mutations including insertions and deletions in exons 18, 19, 20 and 21, in one single test. The complete process takes less than 2.5 hours and has a LOD of ≤5% for all most prevalent EGFR mutations.

PERFORMANCE
Comparison study versus Therascreen, Qiagen
• Overall concordance with Therascreen was 97.5%.
• Inter-laboratory reproducibility (3 sites, day, 2 operators, 2 instruments): 100% on 600 samples, 120 replicates per sample.
• Inter-lot reproducibility (3 lots, 2 instruments, 5 days, 1 operator): 100% on 300 samples, 60 replicates per sample.
• No tissue-size related correlation with valid and concordant results observed on 179 metastatic NSCLC samples, with a tissue area ranging from 1 to 567 mm².

LOW INVALID RATE
• Invalid rate for Idylla™ was 7% (13/179) whereas for Therascreen 30% (54/179)
• 80% (20/25) of the samples that resulted in an invalid test result with NGS (25/68: 37%) could be ‘rescued’ by the Idylla™ test.

TURN AROUND TIME
• EGFR and KRAS testing with rapid systems could spare time, statistically in about 45% of cases.
• Compared to pyrosequencing, it has been shown that idylla™ had a substantial shorter turn around time (3 hours versus 12 hours).
• It has been shown that testing on a local on-site system (cfr. Idylla™) versus external biopathological platform is much quicker, <1 week versus <3 weeks.

SAMPLE TYPE
• The Idylla™ EGFR Mutation Test has been validated for FFPE use. A single 5mm FFPE tissue section can be used.
• A liquid biopsy test, ctEGFR Mutation Assay, will be available soon. 2mL plasma can be used.
• Several researchers have conducted feasibility studies to show the suitability of other sample types:
  - Extracted DNA of cytological samples and cytological smears
  - Extracted DNA of FFPE samples
  - EBUS-FNA samples and Touch prep fresh lung tissue

REFERENCES