Validation of the Idylla GeneFusion Assay to detect fusions and MET exon-skipping in non-small cell lung cancers

Gilson, et al. Sci Rep 13, 12909 (2023). doi:10.1038/s41598-023-39749-4

STUDY AIM

Gene fusions and MET exon skipping drive oncogenesis in 8–9% and 3% of non-small cell lung cancers (NSCLC) respectively. Their detection are essential for the management of patients since they confer sensitivity to specific targeted therapies with significant clinical benefit over conventional chemotherapy. The aim of the study is to evaluate the Idylla™ GeneFusion Assay, a fully-automated test that can identify a panel of specific gene fusions (including 16 specific ALK fusions, 13 ROS1 fusions, 7 RET fusions and MET exon 14 skipping) as well as expression imbalance (in ALK, ROS1, RET, NTRK1, NTRK2 and NTRK3 genes) without pre-analytical RNA extraction.

STUDY DESIGN

In this retrospective study, an evaluation of the Idylla™ GeneFusion Assay was performed in **35 clinical FFPE** specimens from NSCLC patients using IHC/FISH and RNASeq as the gold standards. This included the analysis of 11 clinical samples that did not reach RNA quality requirements for RNASeq.

RESULTS

PERFORMANCE

Idylla™ *versus* NGS (RNAseq)

100% sensitivity (18/18)

100% specificity (5/5)

Idylla™ *versus* IHC/FISH

91.7% sensitivity (11/12)

89.5% specificity (17/19)

Idylla™ validity

94.3% (33/35)

SALVAGE OF INCONCLUSIVE NGS SAMPLES

Idylla™ GeneFusion Assay was able to retrieve **90.9%** (10/11) of the FFPE samples that were found inconclusive by RNASeq due to insufficient RNA quality. Those included two samples **with actionable alterations** detected by Idylla™ that could benefit from targeted therapies.

PROPOSED LABORATORY WORKFLOWS

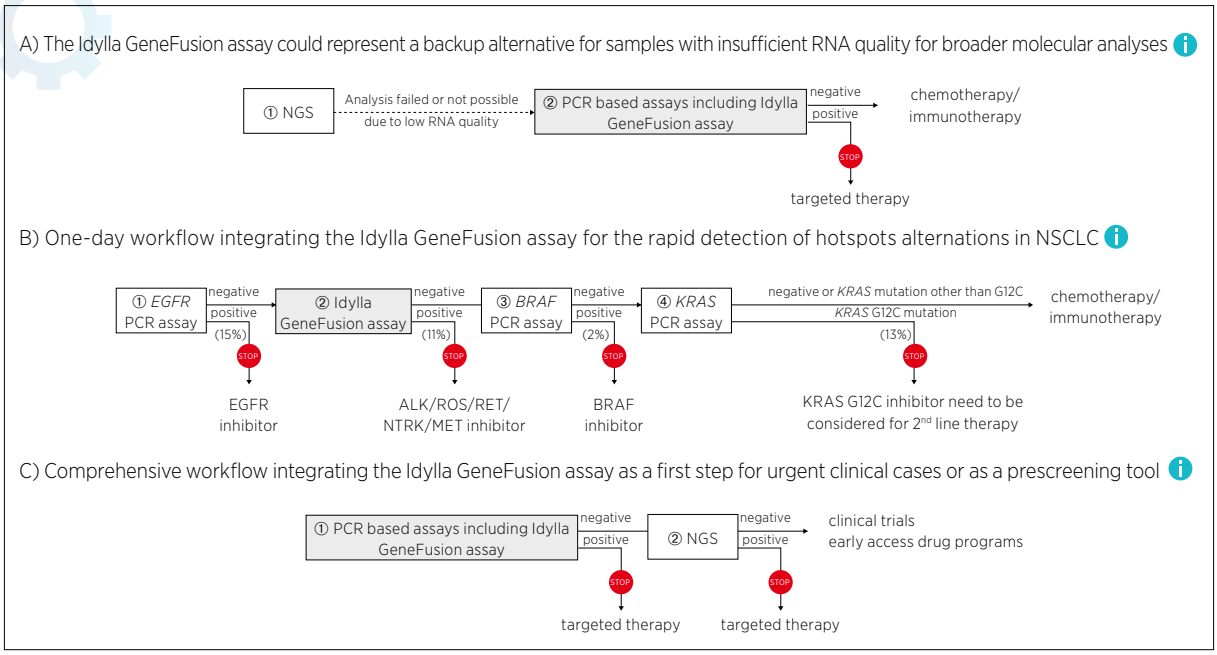

Figure 1. Proposition of laboratory workflows integrating the Idylla GeneFusion assay.

The Idylla™ GeneFusion Assay appears as a fast and reliable alternative to reference methods to detect gene fusions.

Due to its minimal hands-on-time and low turnaround time, it could be easily integrated into laboratory workflows in order to give more rapid tumor molecular profiles or retrieve samples rejected by other molecular techniques.

Biocartis NV
Generaal De Wittelaan 11B
2800 Mechelen, Belgium
+32 15 632 888
www.biocartis.com
customerservice@biocartis.com

Biocartis US, Inc
2 Pierce Place, Suite 1510
Itasca, IL 60143
1-844-443-9552
www.biocartis.com/US
customerserviceUS@biocartis.com

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