Technical sheet Idylla™ EGFR Mutation Test

EGFR CE IVD

The **Idylla™ EGFR Mutation Test**, performed on the Biocartis Idylla™ System, is an *in vitro* diagnostic test for the qualitative detection of **exon 18 (G719A/C/S), exon 21 (L858R, L861Q), exon 20 (T790M, S768I) mutations, exon 19 deletions and exon 20 insertions** in the *EGFR* oncogene. The Idylla™ EGFR Mutation Test, from **sample-to-result**, starts with formalin-fixed, paraffin-embedded (FFPE) human tissue from non-small cell lung cancer (NSCLC) to liberate DNA for subsequent real-time PCR amplification and detection.

Features

EGFR mutation detection		
Exon 18	G719A	c.2156G>C
	G719C	c.2155G>T; c.2154_2155delinsTT
	G719S	c.2155G>A
Exon 19	Del9	c.2238_2248delinsGC
		c.2239_2248delinsC
		c.2240_2248del
		c.2239_2247del
	Del12	c.2239_2251delinsC
		c.2240_2251del
	Del15	c.2235_2249del
		c.2236_2250del
		c.2239_2253del
		c.2240_2254del
		c.2238_2252del
		c.2237_2251del
		c.2235_2252delinsAAT
		c.2237_2252delinsT
		c.2234_2248del
		c.2236_2253delinsCTA
		c.2237_2253delinsTA
		c.2235_2251delinsAG
		c.2236_2253delinsCAA
		c.2230_2249delinsGTCAA

Exon 19	Del18	c.2240_2257del c.2237_2255delinsT c.2239_2256del c.2236_2253del c.2239_2258delinsCA c.2237_2254del c.2238_2255del c.2237_2257delinsTCT c.2236_2255delinsAT c.2236_2256delinsATC c.2237_2256delinsTT c.2237_2256delinsTC c.2235_2255delinsGGT
	Del21	c.2238_2258del c.2236_2256del
	Del24	c.2253_2276del
Exon 20	T790M	c.2369C>T
	S768I	c.2303G>T
	insG	c.2310_2311insGGT
	insASV9	c.2308_2309insGCCAGCGTG
	insASV11	c.2308_2311delinsCCAGCGTGGAT
	insSVD	c.2311_2312insGCGTGGACA
	insH	c.2319_2320insCAC
Exon 21	L858R	c.2573T>G c.2573_2574delinsGT c.2573_2574delinsGA
	L861Q	c.2582T>A
EGFR Total (acting as Sample Processing Control)		
Specimen requirements		
Sample Type	1 x 5µm FFPE tissue section	
Neoplastic cells	≥10%, if less macrodissection is required	
Performance		
Analytical Sensitivity	LOD ≤5% for most prevalent *EGFR* mutations	
Between Laboratory Reproducibility (600 results at 3 sites)	100% agreement for 10% EGFR G719S 100% agreement for 10% EGFR Del15 100% agreement for 10% EGFR T790M 100% agreement for 10% EGFR L858R 100% agreement for 10% EGFR L861Q	

Between Lot Reproducibility (300 results on 3 lots)	100% agreement for 10% EGFR G719S 100% agreement for 10% EGFR Del15 100% agreement for 10% EGFR T790M 100% agreement for 10% EGFR L858R 100% agreement for 10% EGFR L861Q
Total turnaround time	
Time	150 minutes

Accuracy - Clinical Performance Evaluation

95.9% overall agreement for the *EGFR* gene was obtained during the clinical performance evaluation comparing Idylla™ with Therascreen (Qiagen), a PCR-based reference method.

95.9% overall concordance

Therascreen / Idylla™	DelEx19	DelEx19, T790M	L858R	L867Q	G719X	G719X, S768I	L858R, T790M	InsEx20	No mutation detected	Totals without invalids	Invalids	Totals
DelEx19	18		–	–	–	–	–	–	–	18	8	26
InsEx20	–	–	–	–	–	–	–	–	–	–	1	1
DelEx19,T790M	–	4		–	–	–	–	–	–	4	–	4
L858R	–	–	20		–	–	1	–	–	21	2	23
L861Q	–	–	–	2	–	–	–	–	–	2	–	2
G719X	–	–	–	–	1	–	–	–	–	1	–	1
G719X,S768I	–	–	–	–	–	1	–	–	–	1	–	1
L858R,T790M	–	–	–	–	–	–	1	–	–	1	–	1
T790M	–	1	–	–	–	–	–	–	1	2	2	4
DelEx19, S768I	1	–	–	–	–	–	–	–	–	1	–	1
S768I	–	–	–	–	–	–	–	–	–	0	1	1
No mutation detected	–	1	2	–	1	–	–	1	66	71	30	101
Totals without invalids	19	6	22	2	2	1	2	1	67	**122**	–	/
Invalids	–	1	–	–	–	–	–	–	2	–	10	13
Totals	19	7	22	2	2	1	2	1	69	/	54	**179**

↓ Discordant analysis by NGS and/or ddPCR (excluding invalids)

97.5% overall concordance

Therascreen and further analysis by NGS and/or ddPCR / Idylla™	DelEx19	DelEx19, T790M	L858R	L867Q	G719X	G719X, S768I	L858R, T790M	T790M	No mutation detected	Totals
DelEx19	18		–	–	–	–	–	–	–	18
DelEx19,T790M	–	4		–	–	–	–	–	–	4
L858R	–	–	22		–	–	–	–	–	22
L861Q	–	–	–	2	–	–	–	–	–	2
G719X	–	–	–	–	1	–	–	–	–	1
G719X,S768I	–	–	–	–	–	1	–	–	–	1
L858R,T790M	–	–	–	–	–	–	1	–	–	1
T790M	–	–	–	–	–	–	–	2	–	2
DelEx19, S768I	1	–	–	–	–	–	–	–	–	1
S768I	–	–	–	–	–	–	–	–	–	0
No mutation detected	1	–	1	–	1	–	–	1	66	70
Totals	20	4	23	2	2	1	1	3	66	**122**

Idylla™ EGFR Posters & Publications

- Troncone G. et al. EGFR mutation detection on lung cancer cytological specimens by the rapid and fully integrated Idylla molecular diagnostics system. Poster ASCO 2016.
- De Luca C. et al. EGFR mutation detection on lung cancer cytological specimens by the novel fully automated PCR-based Idylla EGFR Mutation Assay. J Clin Pathol 2016.
- Reijans M. et al. Fully automated and sensitive detection of EGFR exon 18, 19, 20 and 21 mutational status in less than 2.5 hours from a single FFPE slice. Poster ESMO 2016.
- Laetitia Lambros et al. Evaluation of a fast and fully automated platform to diagnose EGFR and KRAS mutations in formalin-fixed and paraffin-embedded non-small cell lung cancer samples in less than one day. J Clin Pathol 2017.
- Mirko Marabese et al. Comparison of technologies for EGFR analysis within a subset of a randomized clinical trial. Poster AACR 2017.
- Vincent Thomas De Montpréville et al. EGFR and KRAS molecular genotyping for pulmonary carcinomas: Feasibility of a simple and rapid technique implementable in any department of pathology. Pathology Research and Practice 2017. http://www.sciencedirect com/science/article/pii/S0344033817302947
- Marius Ilie et al. Optimization of EGFR mutation testing by the fully-automated qPCR-based Idylla on whole slide and biopsy tumor tissue of non-small cell lung cancer. Abstract e20632, 2017 ASCO Annual Meeting. http://abstracts.asco.org/199 AbstView_199_191634.html

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