

DNA TEST REPORT

Full Name	Christopher Kalinski	Order ID/Sample ID	1228078/9018634
Age/Gender	74 Years/Male	Sample Type	FFPE
Referring Clinician	Dr. Senaka Kandegedara Aegle Omics Private Limited (Colombo)	Block No & Tumor content	RP3326 (A4) / 65%
		Date & time of Sample Receipt	13-03-2025, 11:15:00
		Date & time of Report	20-03-2025, 12:37:44
Collection Center/ Partner Lab			
Test Requested	BRAF V600 mutation analysis [MGM177]		

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Biopsy of lung - Squamous cell carcinoma of lung

RESULTS

No V600 mutation is detected in the *BRAF* gene

CLINICAL CORRELATION AND INTERPRETATION

No V600 mutation is detected in the *BRAF* gene

TEST DETAILS

The *BRAF* gene encodes a protein involved in regulating cell growth and proliferation. Somatic mutations in *BRAF* have been found in cancers, including non-Hodgkin lymphoma, colorectal cancer, malignant melanoma, papillary thyroid carcinoma, non-small cell lung carcinoma, and adenocarcinoma of lung. However, the highest frequency of *BRAF* mutations, 50%, is observed in malignant melanoma [1]. Majority of the mutations in cancer are within the kinase domain leading to a single amino-acid substitution (V600). "The mutational status of *BRAF* will help identify patients that would respond to B-raf inhibitors therapy in melanoma and prognostication in colorectal cancer" [2]. As recommended in the NCCN guidelines (2021), the combination of Dabrafenib and Trametinib is a targeted therapy for the *BRAF* V600 positive metastatic non-small cell lung cancer [3], Hairy cell leukemia [4], colorectal cancer [5], malignant melanoma [6], anaplastic thyroid cancer [7] and glioma [8] patients. This Assay screens for V600 variation in exon 15 of *BRAF* gene by Real-time PCR technology.

METHODOLOGY

DNA extracted from the sample is tested for the presence of indicated hotspot mutations (V600) in *BRAF* gene using Real-time PCR. The target exons are amplified with mutation specific primers. Mutations are reported according to HGVS guidelines for mutation nomenclature (www.hgvs.org) and according to the reference sequence NM_004333.4.

The scope of this assay does not distinguish between what mutation is present in given patient sample. However, the assay screens for four different mutations in any given patient sample. The mutations could be V600E/K/R/D.

LIMITATION

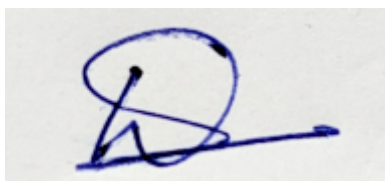
This test is performed using a CE, IVD marked commercial kit. However, there may be limitations imposed by the sensitivity and the specificity of the assay and should be interpreted in conjunction with clinical presentation and other related investigations. Results of the test could be affected by contamination during specimen collection, inappropriate specimen storage and transport.

DISCLAIMER

The analytical sensitivity of the test allows detection of the mutation when the mutant clone comprises at least 1% of the total genomic DNA. Real-time PCR is a highly sensitive technique; reasons for apparently contradictory results may be due to improper quality control during sample collection and fixation, intrinsic heterogeneity of the sample, tumor depletion, high degree of necrosis, mucin content and/or presence of PCR inhibitors.

REFERENCES

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Debabrata Mishra

Senior Research Associate
Operations - Oncology



Dr. Syed Muqlisur Rehman, MD

Molecular Pathologist
KMC Reg No. - 71468

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APPENDIX- HGVS NOMENCLATURE

BRAF V600 Variants				HGVS format reported by VariMAT	
ENS_TRANS_ID: ENSP00000288602.6				CDNA_CHG	AA_CHG
ENS_PROT_ID: ENSP00000288602.6					
REFSEQ_ID: NM_004333.4					
Mutation	Base Change	Cosmic ID	Name		
V600E1	1799T>A	476	BRAF-M1	c.1799T>A	p.Val600Glu
V600K	1798_1799GT>AA	473	BRAF-M2	c.1798_1799delGTinsAA	p.Val600Lys
V600E2	1799_1800TG>AA	475	BRAF-M3	c.1799_1800delTGinsAA	p.Val600Glu
V600R	1798_1799GT>AG	474	BRAF-M4	c.1798_1799delGTinsAG	p.Val600Arg
V600D1	1799_1800TG>AC	308550	BRAF-M5	c.1799_1800delTGinsAC	p.Val600Asp
V600D2	1799_1800TG>AT	477	BRAF-M6	c.1799_1800delTGinsAT	p.Val600Asp

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