

**DNA TEST REPORT – MEDGENOME LABORATORIES (\*RUO)**

Full Name	Nirosha M Karunarathna	Order ID/Sample ID:	1158793/8893200
Age/Gender	51 Years / Female	Sample Type:	Peripheral Blood in EDTA
Hospital Name	Aegle Omics Private Limited, Sri Lanka	Date and time of Sample Collection	NA
Physician Name	Dr. Sujeewa Siyambalapitiya	Date and time of Sample Receipt	04-01-2025 17:55:00
Test Requested	MGM177/ BRAF V600 mutation analysis	Date and time of Report	15-01-2025, 11:00:00

**CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY**

Biopsy of lung - Adenocarcinoma 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](http://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.

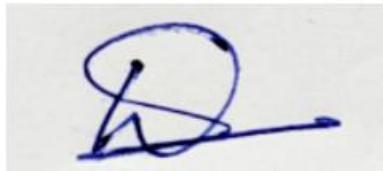
## DISCLAIMER

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.

**\*NOTE: This assay has not been validated on peripheral blood and bone marrow aspirate sample types. The test was performed based on clinician's request. Hence, the report is released on a RUO basis and interpret with caution.**

## REFERENCES

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-----End of Report-----

### APPENDIX- HGVS NOMENCLATURE

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

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