MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

MELP

Melanoma Targeted Gene Panel by Next Generation Sequencing, Tumor

Patient ID	Patient Name		Birth Date	Gender	Age
SA00000835	SAMPLEREPDMGLM, VLD20150713A0095		1981-01-01	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00000835	SA00000835	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jul 2015 15:25			

Result Summary

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Alteration Identified

Result

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BRAF | GNA11 | GNAQ | KIT | NRAS

Provided diagnosis: melanoma

The following alteration was identified:
Gene: BRAF
DNA change: c.1799T>A
Amino Acid change: p.V600E (Val600Glu)
Classification: MUTATION

No additional reportable somatic alterations were identified within the tested genes.

Interpretation

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ASSOCIATIONS BETWEEN BRAF MUTATIONS AND MELANOMA

Approximately 40% of patients with melanoma have a somatic mutation in the BRAF gene (1). BRAF mutations, primarily the p.V600E mutation, result in constitutive activation of the RAS/MAPK signaling pathway (2, 3).

Current data suggests that the efficacy of BRAF-targeted therapy and anti-MEK therapy in melanoma is limited to patients whose tumors harbor a p.V600E/K mutation. Thus, the presence of the p.V600E mutation in this tumor specimen suggests that this patient may respond to such therapy (3–8).

REFERENCES

1. http://cancer.sanger.ac.uk/cancergenome/projects/cosmic/
2. Nature. 2002 Jun 27;417(6892):949–54 (PMID 12068308)
3. J Transl Med. 2010 Jul 14;8:67 (PMID 20630094)
4. Lancet. 2012 May 19;379(9829):1893–901 (PMID 22608338)
5. N Engl J Med. 2012 Feb 23;366(8):707–14 (PMID 22356324)
6. N Engl J Med. 2015 Jan 1;372(1):30–9 (PMID 25399551)
7. N Engl J Med. 2014 Nov 13;371(20):1877–88 (PMID 25265492)
8. Lancet Oncol. 2014 Mar;15(3):323–32 (PMID 24508103)

ADDITIONAL INFORMATION

Microscopic examination was performed by a pathologist to identify areas of tumor for enrichment by macrodissection. Next generation sequencing is performed to test for the presence of a mutation within targeted regions of the following genes: BRAF, GNA11, GNAQ, KIT, and NRAS. Mutation nomenclature is based on the following GenBank accession numbers (build GRCh37 (h19)): BRAF NM_004333, GNA11 NM_002067, GNAQ NM_002072, KIT NM_000222, and NRAS NM_002524. See www.mayomedicallaboratories.com (Test ID MELP) for additional information about this test.

CLINICAL CORRELATIONS

Test results should be interpreted in context of clinical findings, tumor sampling, histopathology, and other laboratory data. If results obtained do not match other clinical or laboratory findings, please contact the laboratory for possible interpretation. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

The presence or absence of a mutation may not be predictive of response to therapy in all patients.

TECHNICAL LIMITATIONS

This test does not detect large insertions, deletions, or duplications or genomic copy number variants.

This assay has been shown to detect >99% of single base

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

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substitutions and >93% of known COSMIC insertions and deletions up to 22bp in length within the reportable range of this assay.

A negative (wild type) result does not rule out the presence of a mutation that may be present but below the limits of detection of this assay. The analytical sensitivity of this assay is 5–10% with a minimum coverage of 100X.

Rare polymorphisms may be present that could lead to false negative or false positive results.

This test cannot differentiate between somatic and germline alterations. Additional testing may be necessary to clarify the significance of results if there is a potential hereditary risk.

Metastatic and corresponding primary lesions may have discordant results.

Additional Information

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CLINICAL TRIALS

Possible clinical trials of benefit for this patient can be found at the following sites:
1) ClinicalTrials.gov:
http://clinicaltrials.gov/ct2/search/advanced
2) Mayo Clinic:
http://www.mayo.edu/research/clinical-trials/
3) National Cancer Institute:
http://www.cancer.gov/clinicaltrials/search

Specimen

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Tissue, Tumor

Tissue ID

MCR

S15–5999

Released By

MCR

EMILY LAUER

Received: 13 Jul 2015 20:21 **Reported:** 24 Jul 2015 15:46

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905