MAYO CLINIC LABORATORIES

1-800-533-1710

# BRAFD

BRAF V600E/V600K Somatic Mutation Analysis, Tumor

| Patient ID | Patient Name | | Birth Date | Gender | Age |
|---|---|---|---|---|---|
| SA00136052 | BRAFDABNORMAL, ATLASVAL | | 1985-08-02 | F | 35 |
| **Order Number** | **Client Order Number** | **Ordering Physician** | **Report Notes** | | |
| SA00136052 | SA00136052 | CLIENT,CLIENT | | | |
| **Account Information** | | **Collected** | | | |
| C7028846 DLMP Rochester | | 31 Aug 2020 00:00 | | | |

## BRAF V600 Somatic Mutation Analysis, Tumor

### Result Summary MCR

POSITIVE

### Result MCR

**Provided diagnosis:** metastatic melanoma involving the brain

**BRAF status:** Mutant (V600K)

### Interpretation MCR

#### ASSOCIATIONS BETWEEN BRAF MUTATIONS AND MELANOMA

Approximately 40% of patients with melanoma have a somatic mutation in the BRAF gene (1). BRAF mutations, primarily those occurring at codon 600, result in constitutive activation of the RAS/MAPK signaling pathway (2). The p.V600K mutation accounts for approximately 5–12% of BRAF mutations occurring at codon 600 (1–3).

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.V600K mutation in this tumor specimen suggests that this patient may respond to such therapy (3–8).

#### REFERENCES


1. 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)


### Specimen MCR

Tissue, Tumor

### Tissue ID MCR

4567

### Method MCR

Microscopic examination was performed by a pathologist to identify areas of tumor for enrichment by macrodissection. Tumor DNA was isolated and evaluated for the presence of BRAF V600E/K mutations using digital droplet PCR analysis.

### Disclaimer [1] MCR

Test results should be interpreted in the context of clinical findings, family history, 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.

Rare polymorphisms exist that could lead to false-negative or false-positive results.

This assay was designed to detect V600E and K mutations.

A negative result does not rule out the presence of the V600E or K mutation that may be present but below the limit of detection for this assay (approximately 0.1–0.5%).

### Released By MCR

Kevin C. Halling, M.D., Ph.D.

**Received:** 31 Aug 2020 12:30 **Reported:** 02 Sep 2020 17:39



### 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.

**Performing Site Legend**

| Code | Laboratory | Address | Lab Director | CLIA Certificate |
|---|---|---|---|---|
| MCR | Mayo Clinic Laboratories - Rochester Main Campus | 200 First Street SW, Rochester, MN 55905 | William G. Morice M.D. Ph.D | 24D0404292 |