

## Overview

### Useful For

Therapy selection for patients with cancer (eg, melanomas that may respond to BRAF inhibitors, colon cancers than may not respond to EGFR inhibitors)

Aiding in the diagnosis/prognosis of certain cancers (eg, hairy cell leukemia, papillary thyroid cancers, and association with aggressiveness)

Aid in determining risk for Lynch syndrome (eg, an adjunct to negative *MLH1* germline testing in cases where colon tumor demonstrates MSI-H and loss of MLH1 protein expression)

### Additional Tests

Test Id	Reporting Name	Available Separately	Always Performed
SLIRV	Slide Review in MG	No, (Bill Only)	Yes

### Testing Algorithm

When this test is ordered, slide review will always be performed at an additional charge.

See [Lynch Syndrome Testing Algorithm](#) in Special Instructions.

### Special Instructions

- [Lynch Syndrome Testing Algorithm](#)

### Method Name

Digital Droplet Polymerase Chain Reaction (ddPCR)

### NY State Available

Yes

## Specimen

### Specimen Type

Varies

### Necessary Information

**Pathology report** (final or preliminary) **must accompany specimen in order for testing to be performed.** At minimum, it should contain the following information:

1. Patient name
2. Block number-must be on all blocks, slides and paperwork (can be handwritten on the paperwork)
3. Tissue collection date
4. Source of the tissue

**Specimen Required**

**Preferred:**

**Specimen Type:** Tissue

**Container/Tube:** Tissue block

**Collection Instructions:** Submit a formalin-fixed, paraffin-embedded tissue block.

**Acceptable:**

**Specimen Type:** Tissue

**Container/Tube:** Slides

**Specimen Volume:** 1 stained and 5 unstained

**Collection Instructions:** Submit 1 slide stained with hematoxylin and eosin and 5 unstained, nonbaked slides with 5-micron thick sections of the tumor tissue.

**Forms**

If not ordering electronically, complete, print, and send an [Oncology Test Request](#) (T729) with the specimen.

**Reject Due To**

Specimens that have been decalcified (all methods) Specimens that have not been formalin-fixed, paraffin-embedded Bone marrow in EDTA	Reject
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**Specimen Stability Information**

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Refrigerated		

**Clinical & Interpretive**

## Clinical Information

This test assesses for somatic (tumor-specific) *BRAF* V600E and V600K alterations. The *BRAF* gene is a member of the mitogen-activated protein/extracellular signal-regulated (MAP/ERK) kinase pathway, which plays a role in cell proliferation and differentiation. Dysregulation of this pathway is a key factor in tumor progression and *BRAF* alterations occur frequently in many different tumor types. *BRAF* variant analysis aids in the diagnosis of cancer types including anaplastic and papillary thyroid carcinoma, hairy cell leukemia, and papillary craniopharyngioma.

*BRAF* V600E and V600K alterations are associated with response or resistance to specific targeted therapies in cancers such as melanoma, colorectal cancer, and lung cancer. Targeted cancer therapies are defined as antibody or small molecule drugs that block the growth and spread of cancer by interfering with specific cell molecules involved in tumor growth and progression. Multiple targeted therapies have been approved by the US Food and Drug Administration (FDA) for treatment of specific cancers. Molecular genetic profiling is often needed to identify targets amenable to targeted therapies and to minimize treatment costs and therapy-associated risks.

*BRAF* variant analysis can provide helpful diagnostic information in the context of evaluation for Lynch syndrome. [See Lynch Syndrome Testing Algorithm](#) in Special Instructions.

## Reference Values

An interpretive report will be provided.

## Interpretation

An interpretive report will be provided.

## Cautions

Not all tumors that have *BRAF* alterations respond to *BRAF*-targeted therapies.

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

Test results should be interpreted in context of clinical findings, tumor sampling, 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.

Colon cancer is relatively common and it is possible for a sporadic colon cancer to occur in a Lynch syndrome family. Therefore, evaluation of other family members should still be considered in cases with *MLH1* promoter hypermethylation and absence of the *BRAF* V600E alteration if there is high clinical suspicion of Lynch syndrome.

## Clinical Reference

1. Chapman PB, Hauschild A, Robert C, et al: BRIM-3 Study Group. Improved survival with vemurafenib in melanoma with BRAF V600E mutation. *N Engl J Med*. 2011 Jun;364(26):2507-2516
2. Di Nicolantonio F, Martini M, Molinari F, et al: Wild-type BRAF is required for response to Panitumumab or Cetuximab in metastatic colorectal cancer. *J Clin Oncol*. 2008; 26(35):5705-5712
3. Hyman DM, Puzanov I, Subbiah V, et al: Vemurafenib in Multiple Nonmelanoma Cancers with BRAF V600 Mutations. *N Engl J Med*. 2015 Aug 20;373(8):726-736
4. Domingo E, Laiho P, Ollikainen M, et al: BRAF screening as a low-cost effective strategy for simplifying HNPCC genetic testing. *J Med Genet*. 2004;41(9):664-668

## Performance

### Method Description

Digital droplet polymerase chain reaction (PCR)-based assay to detect the presence of the *BRAF* V600E and *BRAF* V600K alterations.(Unpublished Mayo method)

### PDF Report

No

### Day(s) Performed

Monday through Friday

### Report Available

5 to 7 days

### Specimen Retention Time

Unused slides: 5 years;. Extracted DNA: 3 months. Unused portions of blocks will be returned.

### Performing Laboratory Location

Rochester

## Fees & Codes

### Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

### Test Classification

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 US Food and Drug Administration.

### CPT Code Information

81210

88381-Microdissection, manual

### LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
BRAFD	BRAF V600 Somatic Mutation Analysis, Tumor	97025-1

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Result ID	Test Result Name	Result LOINC® Value
608306	Result Summary	50397-9
608307	Result	97025-1
608308	Interpretation	69047-9
608309	Additional Information	48767-8
608310	Specimen	31208-2
608311	Source	31208-2
608312	Released By	18771-6
608235	Method	85069-3
608222	Tissue ID	80398-1
606746	Disclaimer	62364-5