

BRAF V600 Somatic Mutation Analysis, Bone
Marrow

#### Overview

#### **Useful For**

Classification and/or possible targeted therapies of hematological neoplasms such as hairy cell leukemia, Langerhans cell histiocytosis, Erdheim-Chester disease

#### **Method Name**

Real-Time PCR Amplification and Detection

#### **NY State Available**

No

# **Specimen**

## **Specimen Type**

**Bone Marrow** 

#### **Shipping Instructions**

Specimen must arrive within 7 days of collection.

# **Necessary Information**

The following information is required:

- 1. Pertinent clinical history
- 2. Clinical or morphologic suspicion
- 3. Date of collection
- 4. Specimen source

# Specimen Required

Container/Tube: Lavender top (EDTA)

**Specimen Volume:** 3 mL **Collection Instructions:** 

- 1. Invert several times to mix bone marrow.
- 2. Send bone marrow specimen in original tube. Do not aliquot.
- 3. Label specimen as bone marrow.

# **Specimen Minimum Volume**

1 mL

#### **Reject Due To**

Gross	Reject
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hemolysis	
Moderately to	Reject
severely	
clotted	

## **Specimen Stability Information**

Specimen Type	Temperature	Time	Special Container
Bone Marrow	Ambient (preferred)	7 days	
	Refrigerated	7 days	

# Clinical & Interpretive

#### **Clinical Information**

This test uses DNA extracted from the bone marrow to test for the presence of BRAF V600E/D and V600K/R/M alterations. BRAF mutations occur in many different types of human cancers. Testing for BRAF mutations in bone marrow specimens facilitates classification and possible targeted therapies of hematological neoplasms, such as hairy cell leukemia, Langerhans cell histiocytosis, and Erdheim-Chester disease. This is test is not designed for detection of BRAF mutations in liquid biopsy of tumors.

#### **Reference Values**

An interpretive report will be provided.

#### Interpretation

The interpretive report includes an overview of the findings.

Results will be characterized as positive, negative, or indeterminate for a V600 somatic mutation.

### **Cautions**

Test results should be interpreted in context of clinical findings 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.

#### Clinical Reference

- 1. Maitre E, Cornet E, Troussard X. Hairy cell leukemia: 2020 update on diagnosis, risk stratification, and treatment. Am J Hematol. 2019;94(12):1413-1422
- 2. Rodriguez-Galindo C, Allen CE. Langerhans cell histiocytosis. Blood. 2020;135(16):1319-1331
- 3. Haroche J, Cohen-Aubart F, Amoura Z. Erdheim-Chester disease. Blood. 2020;135(16):1311-1318

#### **Performance**



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### **Method Description**

Polymerase chain reaction-based assay using bone marrow sample. This test detects the most common mutation V600E and other variants at the same codon. The limit of detection for this assay is approximately 1%. The mutation is reported using the current standard nomenclature. (Unpublished Mayo method)

#### **PDF Report**

No

# Day(s) Performed

Monday through Friday

#### Report Available

1 to 6 days

## **Specimen Retention Time**

Bone marrow: 2 weeks; Extracted DNA: 1 year

#### **Performing Laboratory Location**

Mayo Clinic Jacksonville Clinical Lab

#### **Fees & Codes**

#### **Fees**

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

## **Test Classification**

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

#### **CPT Code Information**

81210

# **LOINC®** Information

Test ID	Test Order Name	Order LOINC® Value
MBRAF	BRAF V600 Somatic Mutation, BM	85101-4

Result ID	Test Result Name	Result LOINC® Value
616930	Result	85101-4
616928	Interpretation	59465-5
616939	Signing Pathologist	18771-6



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616936	Method Summary	85069-3
616934	Sample ID	80398-1
616938	Disclaimer	62364-5
616931	Indication for Testing	42349-1
616952	Specimen	31208-2
616933	Source	31208-2