Patient Report

labcorp

Specimen ID: 046-225-9994-0
Control ID:

SAMPLE REPORT, 485140

Acct #: 90001325 **Phone:** (919) 361-7217 **Rte:** 99
Labcorp Of America
CMBP
1912 TW Alexander Dr
Durham NC 27709

Patient Details
DOB:
Age(y/m/d): 000/00/00
Gender: N
Patient ID:

Specimen Details
Date collected: 02/14/2022 0000 Local
Date received: 02/15/2022
Date entered: 02/15/2022
Date reported: 02/15/2022 1508 ET

Physician Details
Ordering:
Referring:
ID:
NPI:

Ordered Items
KIT (D816V) Digital PCR, SpTst

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
KIT (D816V) Digital PCR, SpTst					
CKIT Result [A]	Positive				01
The KIT D816V mutation was detected. Results should be interpreted in conjunction with all other clinical, molecular, and cytogenetic findings for the most accurate interpretation. For serial monitoring of transcript levels, consider resubmitting a specimen using KIT (D816V) Digital PCR test. (Labcorp test code 485126).					
CKIT Quant Value	2.32		%		01
Specimen Type	Blood				01
Background					01

Systemic mastocytosis (SM) is a hematopoietic neoplasm characterized by an abnormal growth of clonal mast cells in bone marrow and other extracutaneous organs.
KIT is a receptor tyrosine kinase involved in proliferation of mast cells, melanocytes, germ cells, and hematopoietic stem cells. Activation of KIT occurs upon binding of the stem cell factor ligand, which triggers autophosphorylation and dimerization of KIT. Activated KIT signals downstream protein kinase pathways which induces cell proliferation and survival.
The vast majority (>90%) of SM cases have a somatic A to T missense mutation at position 2447 in exon 17 of the KIT gene. This KIT D816V mutation (c.2447 A>T, p.D816V) results in the substitution of aspartate (D) to valine (V) at codon 816 in the kinase activation loop domain of the protein causing a conformational change in the receptor. This conformational change results in ligand-independent constitutive activation of KIT and leads to increased cell proliferation and accumulation in various organs, and a reduction in cell death. The detection of KIT D816V is one of the minor diagnostic criteria for SM per the WHO system. KIT mutation detection is correlated with the proportion of lesional cells in the sample as well as the sensitivity of the detection method employed.
Quantitative detection using digital PCR of the KIT D816V mutation may aid physicians in diagnosis and therapeutic monitoring of patients with SM. The reported values will allow clinicians to predict disease severity and/or monitor the effectiveness of treatment protocols and to detect increasing mutation levels that may be indicative of patient relapse. An effect of mutational dose on disease phenotype may also prove significant.

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
Method/Extraction					02

Nanoplate Digital PCR (dPCR)
Total genomic DNA is extracted and amplified using a multiplex digital PCR using wild-type and mutant-specific probes.
The assay is designed to detect the KIT D816V (KITc.2447A>T) mutation. Results are reported as percent mutated alleles. The mutation can be detected down to 0.03% mutated alleles.

References 01

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Intake Review Completed 01

Director Review

Dan Wang, PhD, FACMG 01
Director, Molecular Oncology
LabCorp Center for Molecular Biology and Pathology
Research Triangle Park, NC 27709
1-800-533-0567

Comments:

A This test was developed and its performance characteristics determined by Labcorp. It has not been cleared or approved by the Food and Drug Administration.

01	YU	Labcorp RTP 1904 TW Alexander Drive Ste C, RTP, NC 27709-0153	Dir: Anjen Chenn, MDPhD
02	TG	Labcorp RTP 1912 TW Alexander Drive, RTP, NC 27709-0150	Dir: Anjen Chenn, MDPhD

For inquiries, the physician may contact **Branch: 800-762-4344 Lab: 800-735-4087**