MAYO CLINIC LABORATORIES

1-800-533-1710

BAKDM

BCR/ABL1 Mutation, Sequencing

Patient ID	Patient Name		Birth Date	Gender	Age
SA00141367	TESTINGRNV, VALIDATION		1999-09-09	M	21
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00141367	SA00141367	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		11 Jan 2021 07:00			

BCR/ABL1 Mutation, Sequencing

Specimen Type MCR

Peripheral blood

BCRABL Fusion (210, 190, 205, 230) MCR

210

Final Diagnosis: 1 MCR

Peripheral blood, BCR-ABL1 Kinase Domain Mutation Analysis for Therapeutic Resistance:

Positive. Mutation identified in the BCR-ABL1 kinase domain region. This mutation and its corresponding amino acid change is: g. 133748283C>T; c.944C>T; p.Thr315Ile (T315I)

The presence of BCR-ABL1 kinase domain mutation may be associated with acquired resistance to imatinib.

The T315I mutation is associated with pan-resistance to nearly all tyrosine kinase inhibitors, but is sensitive to ponatinib.

References: 1) Baccarani M, et al. Blood 2013; 122: 872–884. 2) Soverini S, et al. Blood 2011;118(5):1208–1215. 3) Press RD, et al. J Mol Diagn 2013;15(5):565–576. 4) NCCN guidelines, Chronic Myeloid Leukemia (version 1. 2019-August 1, 2018)

Per provided client information, this patient is reported to have a p210 BCR-ABL mRNA transcript type.

Signing Pathologist: BENJAMIN BAILEY

ADDITIONAL INFORMATION

This assay detects acquired mutations in the kinase domain region of the BCR-ABL1 oncogene (ABL1 amino acids 218–401; cDNA reference GRCh37(hg19)NM_005157.5), which have been associated with significant clinical or in vitro resistance to tyrosine kinase inhibitor therapy.

Method Summary: Total RNA was extracted and nested reverse transcription PCR was performed to amplify the BCR-ABL1 transcript including the ABL1 kinase domain (KD) region. The presence of kinase domain mutations (KDM) was evaluated by Sanger sequencing of the ABL1 region. If the BCR-ABL1 quantitative PCR level is very low, RT-PCR amplification of BCR-ABL1 mRNA may be unsuccessful in this assay. In general, a normalized BCR-ABL1 quantitative level above 0.1% (International scale) is required in order to detect a KDM by this assay.

Received: 12 Jan 2021 09:28 **Reported:** 13 Jan 2021 11:22

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