



Test Definition: BARQ

BCR::ABL1, Rare Fusion Monitoring,
Quantitative, Varies

Overview

Useful For

Quantitative monitoring of rare (non-p210 [non-E13/E14A2], non-p190 [non-E1A2]) *BCR::ABL1* fusion transcript types occurring in myeloid neoplasms (eg, CML, myeloproliferative neoplasms) or B-cell acute lymphoblastic leukemias.

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
BADX	BCR/ABL1, RNA-Qual, Diagnostic	Yes	No
BA190	BCR/ABL1, p190, Quant, Monitor	Yes	No
BCRAB	BCR/ABL1, p210, Quant, Monitor	Yes	No

Testing Algorithm

If a previous *BCR::ABL1* rare fusion has not been identified by Mayo Clinic Laboratories (MCL), the qualitative, diagnostic assay for *BCR::ABL1* will be performed at an additional charge to identify the fusion form.

If MCL has previously identified a p190 or p210 *BCR::ABL1* fusion form, the appropriate quantitative testing will be performed and this test will be canceled.

Method Name

Droplet Digital Polymerase Chain Reaction (ddPCR)

NY State Available

Yes

Specimen

Specimen Type

Varies

Ordering Guidance

This test **should not** be used to screen for *BCR::ABL1* fusions at the time of diagnosis. For diagnostic evaluation, please order BADX / *BCR/ABL1*, Qualitative, Diagnostic Assay, Varies; or BCRFX / *BCR/ABL1* Qualitative Diagnostic Assay with Reflex to *BCR/ABL1* p190 Quantitative Assay or p210 Quantitative Assay, Varies.

For measurable residual disease (MRD) monitoring of patients with chronic myeloid leukemia (CML) or, less commonly,

B-cell acute lymphoblastic leukemia (B-ALL) or acute myeloid leukemia (AML) with a previously identified p210 (e13/e14-a2) fusion transcript, order BCRAB / *BCR/ABL1*, p210, mRNA Detection, Reverse Transcription-PCR (RT-PCR), Quantitative, Monitoring Chronic Myelogenous Leukemia (CML), Varies.

For MRD monitoring of B-ALL or rare CML or AML patients with a previously identified p190 (e1-a2) fusion transcript, order BA190 test / *BCR/ABL1*, p190, mRNA Detection, Reverse Transcription-PCR (RT-PCR), Quantitative, Monitoring Assay, Varies.

For more information, see [BCR/ABL1 Ordering Guide for Blood and Bone Marrow](#)

Shipping Instructions

Ambient specimens must arrive within 3 days (72 hours) of collection. Refrigerated specimens must arrive within 5 days of collection. Collect and package specimens as close to shipping time as possible.

Necessary Information

The following information is required:

1. Pertinent clinical history including if the patient has a diagnosis of chronic myeloid leukemia, B-cell acute lymphoblastic leukemia, or other *BCR::ABL1*-positive neoplasm
2. Specific fusion transcript if previously determined
3. Date of collection
4. Specimen source (blood or bone marrow)

Specimen Required

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD)

Specimen Volume: 10 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot.**
3. Label specimen as blood.

Specimen Type: Bone marrow

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD)

Specimen Volume: 4 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.

Forms

1. [Hematopathology Patient Information](#) (T676)
2. If not ordering electronically, complete, print, and send a [Hematopathology/Cytogenetics Test Request](#) (T726) with the specimen.

Specimen Minimum Volume

Whole blood: 8 mL; Bone marrow: 2 mL

Reject Due To

Gross hemolysis	Reject
Moderately or severely clotted	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Refrigerated (preferred)	5 days	PURPLE OR PINK TOP/EDTA
	Ambient	72 hours	PURPLE OR PINK TOP/EDTA

Clinical & Interpretive

Clinical Information

The t(9;22)/*BCR::ABL1* abnormality is associated with chronic myeloid leukemia (CML) and "Philadelphia-positive" acute lymphoblastic leukemia of B-cell lineage (Ph+ B-ALL). Very rarely has this abnormality also been identified in cases of acute myeloid leukemia and T-lymphoblastic leukemia/lymphoma. The fusion genes on the derivative chromosome 22q11 produce a chimeric *BCR::ABL1* messenger RNA (mRNA) transcript and corresponding translated oncoprotein. Because of substantial breakpoint heterogeneity at the genomic level, *BCR::ABL1* mRNA transcripts are utilized for sensitive detection and quantification by reverse transcription-polymerase chain reaction (RT-PCR) techniques. Common *BCR::ABL1* fusion transcript types include the p210 (E13/E14A2) product, which is associated with nearly all cases of CML and some cases of B-ALL, and the p190 (E1A2) product, which is associated primarily with B-ALL. However, rare alternate breaks in both *BCR* and *ABL1* are described in CML, B-ALL and other rare myeloid neoplasms, resulting in a variety of alternate *BCR::ABL1* fusion transcript types. These include, but are not limited to, the p230 (E19A2) fusion, *BCR::ABL1* fusions involving *ABL1* A3, and other rare types (eg, E6A2, E8A2). These alternate *BCR::ABL1* fusion transcript types can be identified with a comprehensive diagnostic RT-PCR screening assay (see test ID: BCRFX / *BCR/ABL1* Qualitative Diagnostic Assay with Reflex to *BCR/ABL1* p190 Quantitative Assay or *BCR/ABL1* p210 Quantitative Assay, Varies or BADX / *BCR/ABL1*, Qualitative, Diagnostic Assay, Varies); however, rare transcript targets cannot be monitored using specific quantitative assays that typically target the more common p210 or p190 events. To meet this need, this test was developed using a droplet digital PCR methodology (ddPCR). If a rare *BCR::ABL1* fusion type is identified by diagnostic RT-PCR screening, this test can subsequently be used to monitor patients with the specific rare fusion form. This assay provides quantitative evaluation of 10 alternate rare *BCR::ABL1* fusion transcripts and better individualizes treatment

response monitoring and clinical management for these patients.

Reference Values

An interpretive report will be provided

Interpretation

An interpretive report will be provided.

Cautions

Although this quantitative droplet digital polymerase chain reaction (ddPCR) assay is comprehensive for detecting and quantifying 10 rare alternative (non-p210, non-p190) *BCR::ABL1* fusions, there are additional extremely rare fusions (eg, complex translocation/rearrangement events) that may produce highly unusual *BCR::ABL1* products that may not be detectable by this assay

The precision of this assay at very low *BCR::ABL1* levels is less reliable, such that inter-run results can be slightly variable. Significant changes during monitoring should be verified by testing a subsequent specimen.

Results of this assay cannot be directly compared with data generated from other polymerase chain reaction (PCR) assays, including similar assays performed in other laboratories.

The results of this assay cannot be directly compared with *BCR::ABL1* results obtained using fluorescence in situ hybridization (FISH) technology. FISH measures the presence of rearrangements in single cells, whereas this ddPCR-based assay measures relative expression of messenger RNA (mRNA) transcripts. FISH is generally not as sensitive as ddPCR.

Blood or bone marrow can be used for disease monitoring. While *BCR::ABL1* levels in blood and bone marrow drawn at the same time are generally similar, bone marrow may provide a slight increase in detection sensitivity (0.5-1 log).

Specimens with delayed transport or nearing the stability window as stated may result in sufficient RNA degradation to produce false-negative results. Thus, specimens should be shipped as quickly as possible. Ambient specimens over 3 days old and refrigerated specimens over 5 days old at the time of receipt are not acceptable.

Clinical Reference

1. Burmeister T, Reinhardt R. A multiplex PCR for improved detection of typical and atypical BCR-ABL fusion transcripts. *Leuk Res*. 2008;32(4):579-585
2. Melo JV. The diversity of BCR-ABL fusion proteins and their relationship to leukemia phenotype. *Blood*. 1996;88(7):2375-2384
3. Melo JV. BCR-ABL gene variants. *Baillieres Clin Haematol*. 1997;10(2):203-222
4. Baccarani M, Castagnetti F, Gugliotta G, et al. The proportion of different BCR-ABL1 transcript types in chronic myeloid leukemia. An international overview. *Leukemia*. 2019;33(5):1173-1183. doi:10.1038/s41375-018-0341-4
5. Petiti J, Lo Iacono M, Dragani M, et al. Novel multiplex droplet digital PCR assays to monitor minimal residual disease in chronic myeloid leukemia patients showing atypical *BCR-ABL1* transcripts. *J Clin Med*. 2020;9(5):1457. Published 2020 May 13. doi:10.3390/jcm9051457
6. Schafer V, White HE, Gerrard G, et al. Assessment of individual molecular response in chronic myeloid leukemia patients with atypical BCR-ABL1 fusion transcripts: recommendations by the EUTOS cooperative network. *J Cancer Res*

Clin Oncol. 2021;147(10):3081-3089. doi:10.1007/s00432-021-03569-8

Performance

PDF Report

No

Day(s) Performed

Weekly

Report Available

10 to 15 days

Specimen Retention Time

Whole blood, Bone marrow: 2 weeks; Extracted RNA: 3 months

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81208

BADX: 81206, 81207, 81208 (if appropriate)

BA190: 81207 (if appropriate)

BCRAB: 81206 (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
BARQ	BCR::ABL1 Rare fusion Quant	75892-0

Test Definition: BARQ

BCR::ABL1, Rare Fusion Monitoring,
Quantitative, Varies

	Monitor	
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Result ID	Test Result Name	Result LOINC® Value
MP097	Specimen Type	31208-2
MP098	BCRABL Fusion	75892-0
623494	Signing Pathologist	18771-6
623479	Interpretation	69047-9