# Test Definition: BADX

BCR::ABL1, Qualitative, Diagnostic Assay, Varies

## Overview

### Useful For

Initial diagnosis of *BCR::ABL1*-positive hematologic neoplasms (eg, chronic myeloid leukemia and acute lymphoblastic leukemia)

Detection and identification of *BCR::ABL1* fusion forms, including the common p210 (e13/e14a2) and p190 (e1a2) forms, as well as other rare fusion forms

This test is **not useful for** therapeutic monitoring of patients with a known *BCR*::*ABL1* fusion form.

### Testing Algorithm

For more information see:

-Myeloproliferative Neoplasm: A Diagnostic Approach to Bone Marrow Evaluation

-Myeloproliferative Neoplasm: A Diagnostic Approach to Peripheral Blood Evaluation

-BCR::ABL1 Ordering Guide for Blood and Bone Marrow

### Special Instructions

- Myeloproliferative Neoplasm: A Diagnostic Approach to Peripheral Blood Evaluation
- Myeloproliferative Neoplasm: A Diagnostic Approach to Bone Marrow Evaluation
- Hematopathology Patient Information
- BCR::ABL1 Ordering Guide for Blood and Bone Marrow

### Method Name

Reverse Transcription Polymerase Chain Reaction (RT-PCR) Multiplex PCR

### NY State Available

Yes

## Specimen

### Specimen Type

Varies

### Ordering Guidance

This test is qualitative only and **should not be used** for routine therapeutic measurable residual disease monitoring.

To monitor patients carrying *BCR::ABL1* fusion forms coding for p190 (e1a2), order BA190 / *BCR::ABL1*, p190 mRNA Detection, Reverse Transcription-PCR (RT-PCR), Quantitative, Monitoring Assay, Varies.

To monitor patients carrying *BCR::ABL1* fusion forms coding for p210 (e13/e14a2) protein, order BCRAB / *BCR::ABL1*, p210, mRNA Detection, Reverse Transcription-PCR (RT-PCR), Quantitative, Monitoring Chronic Myeloid Leukemia (CML), Varies.

To monitor patients carrying rare *BCR::ABL1* fusion forms coding for e19a2, e13/e14a3, e1a3, e6a2, e19a3, e8a2, e12a2, e6a3, e8a3, and e12a3, order BARQ / *BCR::ABL1*, Rare Fusion Monitoring, Quantitative, Varies.

## Shipping Instructions

**Refrigerate specimens must arrive within 5 days of collection, and ambient specimens must arrive within 72 hours of collection.** Collect and package specimens as close to shipping time as possible.

## Necessary Information

**Pertinent clinical history** including the patient diagnosis of chronic myelogenous leukemia or other *BCR*::*ABL1*-positive neoplasm **is required.**

## 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 whole 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 to 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+ ALL). Very rarely, this abnormality has also been identified in cases of acute myeloid leukemia and T-cell lymphoblastic leukemia/lymphoma. The fusion gene on the derivative chromosome 22q11 produces a chimeric *BCR*::*ABL1* messenger RNA (mRNA) transcript and corresponding translated oncoprotein. Despite substantial breakpoint heterogeneity at the DNA level, a consistent set of *BCR*::*ABL1* mRNA transcripts are produced that can be readily and sensitively detected by reverse transcription polymerase chain reaction (RT-PCR) technique. In CML, breakpoints in *BCR* result in either exons 13 or 14 (e13, e14) joined to exon 2 of *ABL1* (a2). The corresponding e13-a2 or e14-a2 *BCR*::*ABL1* mRNAs produce a 210 kDa protein (p210). Rare cases of CML are characterized by an e19-a2 type mRNA with a corresponding p230 protein. In Ph+ ALL, the majority of cases harbor an e1-a2 *BCR*::*ABL1* mRNA transcript, producing a p190 protein. However, chimeric mRNA type is not invariably associated with disease type, as noted by the presence of p210-positive Ph ALL and very rare cases of p190-positive CML. Therefore, positive results from a screening (diagnostic) assay for *BCR*::*ABL1* mRNA need to be correlated with clinical and pathologic findings.

In addition to the main transcript variants described above, rare occurrences of both CML and Ph+ ALL can have alternative break-fusion events resulting in unusual *BCR-ABL1* transcript types. Examples include e6-a2 and *BCR* exon fusions to *ABL1* exon a3 (eg, e13-a3, e14-a3, or e1-a3). In addition to detecting common *BCR*::*ABL1* mRNA transcripts, this assay also can identify these rarer *BCR*::*ABL1* transcript variants and is, therefore, a comprehensive screen for both usual and uncommon *BCR*::*ABL1* gene fusions in hematopoietic malignancies. Given the nature of genetic events in tumors, however, this assay will not identify extremely rare and unexpected *BCR*::*ABL1* events involving other exons (eg, case report level) and is, therefore, not absolutely specific but is predicted to detect more than 99.5% of *BCR*::*ABL1* events. Therefore, it is recommended that for diagnosis, RT-PCR plus a second method (eg, *BCR*::*ABL1* fluorescence in situ hybridization or cytogenetics) should be used. However, this RT-PCR assay is invaluable at diagnosis for identifying the precise *BCR*::*ABL1* mRNA type (eg, for future quantitative assay disease monitoring), which cannot be done by complementary methods.

This assay is intended as a qualitative method, providing information on the presence (and specific mRNA type) or absence of the *BCR*::*ABL1* mRNA. Results from this test can be used to determine the correct subsequent assay for monitoring of transcript levels following therapy (eg, BCRAB / *BCR*::*ABL1*, p210, mRNA Detection, Reverse Transcription-PCR (RT-PCR), Quantitative, Monitoring Chronic Myeloid Leukemia (CML), Varies; BA190 / *BCR*::*ABL1*, p190, mRNA Detection, Reverse Transcription-PCR (RT-PCR), Quantitative, Monitoring Assay, Varies, or BARQ / *BCR*::*ABL1*, Rare Fusion Monitoring, Quantitative, Varies). Because the assay is analytically sensitive, it compensates for situations such as partially degraded RNA quality or low cell number, but it is not intended for quantitative or monitoring purposes.

### Reference Values

A qualitative result is provided that indicates the presence or absence of *BCR*::*ABL1* messenger RNA. When positive, the fusion variant is also reported.

### Interpretation

An interpretive report will be provided.

When positive, the test identifies the specific messenger RNA fusion variant present to guide selection of an appropriate monitoring assay.
Monitoring is available for common p210 or p190 fusion variant detected.
-Common fusion variants detected: e13-a2 or e14-a2 (p210), e1-a2 (p190), and e6-a2 (p205*)
-Rare fusion variants detected: e13-a3 (p210), e14-a3 (p210), e1-a3 (p190), e19-a2 (p230)
-Potential rare fusions detected: e12-a3, e19-a3
*This is formerly observed as the e6-a2 (p185) fusion form.

### Cautions

No significant cautionary statements

### 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. Tefferi A. The classic myeloproliferative neoplasms: Chronic myelogenous leukemia, polycythemia vera, essential thrombocythemia, and primary myelofibrosis. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019. Accessed September 10, 2026. Available at https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225078035

## Performance

### Method Description

Total RNA is extracted from the patient's blood or bone marrow at the time of diagnosis and messenger RNA (mRNA) is

reverse transcribed into complementary DNA (cDNA). The cDNA is then subjected to polymerase chain reaction (PCR) using 4 separate multiplex reactions. A qualitative result, which will include the relative ratio of target translocation mRNA to control *GUSB* gene mRNA, will be provided. Although this method employs a quantitative PCR platform, the results can be used to evaluate the relative expression levels of the translocation mRNA relative to control mRNA, thus, providing an improved measure of RNA quality in the assay. Reporting of results will be qualitative; either *BCR*::*ABL1* mRNA positive/detected (with transcript type) or negative/not detected.(Unpublished Mayo method)

### PDF Report
No

### Day(s) Performed
Monday through Saturday

### Report Available
5 to 10 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
81206
81207
81208

### LOINC® Information

| Test ID | Test Order Name | Order LOINC® Value |
|---|---|---|
| BADX | BCR/ABL1, RNA-Qual, Diagnostic | 55135-8 |

| Result ID | Test Result Name | Result LOINC® Value |
|---|---|---|

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|---|---|---|
| MP001 | Specimen Type | 31208-2 |
| 19783 | Interpretation | 69047-9 |
| 39466 | Diagnostic BCR/ABL1 Result | No LOINC Needed |