BCR-ABL1 (BCR::ABL1) Qualitative and Quantitative Testing

Last Literature Review: December 2023 Last Update: February 2024

BCR::ABL1 (*BCR-ABL1*) testing is recommended for patients with either chronic myeloid leukemia (CML), a hematopoietic stem cell disease, or acute lymphoblastic leukemia (ALL), an aggressive type of leukemia of either B- or T-lineage immature lymphoid cells.[1,2] These tests use reverse transcription polymerase chain reaction (RT-PCR) to identify and/or quantify the fusion transcript present.

In CML, *BCR::ABL1* fusion gene identification and quantification are used for diagnosis and ongoing therapeutic monitoring.[1] In ALL, *BCR::ABL1* fusion identification and quantification are used for risk stratification and treatment decisions.[2] *BCR::ABL1* usion quantification is used for minimal residual disease (MRD) assessment of Philadelphia chromosome-positive (Ph+) ALL.[2] A qualitative test with reflex to quantitation should be performed in the diagnostic workup to identify the appropriate quantitative test for future use. Repeat qualitative testing should not be performed for therapeutic monitoring as it is less sensitive.

Refer to the following ARUP Consult topics for more disease information and summaries of the testing strategies for conditions associated with *BCR::ABL1*:

- Chronic Myeloid Leukemia - CML
- Acute Lymphoblastic Leukemia - ALL

Genetics

Gene

BCR::ABL1 (*BCR-ABL1*) fusion gene

Test Interpretation

BCR-ABL1, Major (p210), Quantitative

Transcripts Quantified

e13a2 or e14a2 transcripts of the *BCR::ABL1* major (p210) fusion form

Analytic Sensitivity

Limit of detection: 0.0032% IS

Limitation

Does not detect p190, p230, or rare variants of p210 forms

BCR-ABL1, Minor (p190), Quantitative

Transcript Quantified

e1a2 transcript of the *BCR::ABL1* minor (p190) fusion form

Featured ARUP Testing

Diagnostic Qualitative BCR-ABL1 Assay with Reflex to p190 or p210 Quantitative Assays 3005839

Method: Reverse Transcription Polymerase Chain Reaction

- Recommended initial diagnostic test for CML or ALL when the *BCR::ABL1* fusion form is unknown (no previous *BCR::ABL1* molecular testing performed) or unclear
- If the qualitative test is positive for the common fusion transcripts of p210 (major breakpoint) or p190 (minor breakpoint), then the corresponding quantitative test is performed.
- This test should not be reordered once a fusion transcript has been identified for disease monitoring; individual quantitative tests should be ordered instead.

Quantitative Detection of BCR-ABL1, Major Form (p210) 3005840

Method: Reverse Transcription Polymerase Chain Reaction

- Appropriate for monitoring CML or Ph+ ALL once a p210 fusion transcript has been identified
- Quantifies the *BCR::ABL1* major (p210) fusion form (e13a2 or e14a2 transcripts), which is present in almost all cases of CML and in a subset of ALL cases

Quantitative Detection of BCR-ABL1, Minor Form (p190) 3016968

Method: Quantitative Reverse Transcription Polymerase Chain Reaction

- Appropriate for monitoring Ph+ ALL or CML once a p190 fusion transcript has been identified
- Quantifies the *BCR::ABL1* p190 fusion form (e1a2 transcript)

Analytic Sensitivity

Limit of quantitation: 5 x 10-5 *BCR-ABL1*/*ABL1* transcripts

Limit of detection: Between 10 and 20 x 10-6 *BCR-ABL1*/*ABL1* transcripts

Limitation

Does not detect p230, p210, or other rare fusion forms

References

1. National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology: chronic myeloid leukemia. Version 2.2024. Updated Dec 2023; accessed Jan 2024.

2. National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology: acute lymphoblastic leukemia. Version 3.2023. Updated Oct 2023; accessed Jan 2024.

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