MAYO CLINIC LABORATORIES

Test Definition: T821Q

RUNX1::RUNX1T1 Translocation (8;21), Minimal Residual Disease Monitoring, Quantitative, Varies

Overview

Useful For

Detection of *RUNX1*::*RUNX1T1* gene fusion in acute myeloid leukemia patients at the time of diagnosis

Minimal residual disease monitoring during the clinical and therapeutic course of these patients

Highlights

This test is a highly sensitive quantitative assay for the detection of translocation t(8;21)(q22;q22); *RUNX1::RUNX1T1* gene fusion in acute myeloid leukemia patients, at the time of diagnosis as well as minimal residual disease monitoring during the clinical and therapeutic course of these patients.

Method Name

Quantitative Real-Time Reverse Transcription Polymerase Chain Reaction (qRT-PCR)

NY State Available

Yes

Specimen

Specimen Type

Varies

Shipping Instructions

1. Refrigerated specimens must arrive within 5 days of collection, and ambient specimens must arrive within 3 days of collection.

2. Collect and package specimen as close to shipping time as possible.

Necessary Information

The following information is required:

1. Pertinent clinical history

2. Date of collection

3. Specimen source (blood or bone marrow)

Specimen Required

Submit only 1 of the following specimens:

Specimen Type: Blood

Container/Tube: Lavender top (EDTA) or yellow top (ACD-B)

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 aspirate
Container/Tube: Lavender top (EDTA) or yellow top (ACD-B)
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 an Hematopathology/Cytogenetics Test Request (T726) with the specimen.

Specimen Minimum Volume

Peripheral blood: 8 mL; Bone marrow: 2 mL

Reject Due To

Gross hemolysis	Reject
Bone marrow core biopsies	Reject
Heparin sample	Reject
Paraffin-embedded bone marrow clots	Reject
Slides	Reject
Paraffin shavings	Reject
Moderately to severely clotted	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container

Varies	Refrigerated (preferred)	5 days	
	Ambient	72 hours	

Clinical & Interpretive

Clinical Information

RUNX1::RUNX1T1 minimal residual disease (MRD) monitoring in patients with acute myeloid leukemia (AML) with translocation t(8;21)(q22;q22) is useful for evaluating disease response after therapy and identifying individuals with increased risk of relapse. Quantitative real-time reverse transcription polymerase chain reaction testing in neoplasms with known clonal genetic markers can achieve highly sensitive detection of neoplastic cells in blood or bone marrow samples. It is one of the most mature technologies available for this purpose. In this assay, translocation of chromosome 8q22 to 21q22 resulting in fusion of two genes *RUNX1* and *RUNX1T1* will be evaluated. Quantitative results will provide physicians with an accurate and precise measurement of disease burden to guide patient intervention decisions. This assay can be used for post-therapy MRD monitoring as well as detection of *RUNX1::RUNX1T1* fusion in AML patients at the time of diagnosis.

Reference Values

An interpretive report will be provided.

Interpretation

The assay is reported in the form of a normalized ratio of RUNX1::RUNX1T1 fusion transcript to the control gene ABL1 expressed as a percentage, which is an estimate of the level of RUNX1::RUNX1T1 fusion RNA present in the specimen, expressed in relation to the level of RNA from an internal control gene (ABL1). The normalized ratio has no units but is directly related to the level of RUNX1::RUNX1T1 detected (ie, larger numbers indicate higher relative levels of RUNX1::RUNX1T1 and smaller numbers indicate lower levels). A relative expression value minimizes variability in the RNA levels and cell numbers measured in separate specimens tested at different times. The precision of the quantitative assay is excellent, but interassay variability can occur such that result changes should not be considered significant if 2 single measurements differ by less than 0.5 log. More critical results, such as a change in the status of positivity or 1 log or greater increase between 2 positive samples should be repeated on a separate specimen with appropriate time interval to verify the result.

Cautions

The limit of detection for this test is 0.01%. Monitoring should be performed using the same method and laboratory for each subsequent specimen.

Clinical Reference

1. Corbacioglu A, Scholl C, Schlenk RF, et al. Prognostic impact of minimal residual disease in RUNX1-RUNX1T1-positive acute myeloid leukemia. J Clin Oncol. 2010;28(23):3724-3729. doi:10.1200/JCO.2010.28.6468
2. Dohner H, Estey E, Grimwade D, et al. Diagnosis and management of AML in adults: 2017 ELN recommendations from an international expert panel. Blood. 2017;129(4):424-447. doi:10.1182/blood-2016-08-733196
3. Tallman MS, Wang ES, Altman JK, et al. Acute Myeloid Leukemia, Version 3. 2019, NCCN Clinical Practice Guidelines in Oncology. J Natl Compr Canc Netw. 2019;17(6):721-749. doi:10.6004/jnccn.2019.0028

4. Schuurhuis GJ, Heuser M, Freeman S, et al. Minimal/measurable residual disease in AML: a consensus document from the European LeukemiaNet MRD Working Party. Blood. 2018;131(12):1275-1291. doi:10.1182/blood-2017-09-801498
5. Jourdan E, Boissel N, Chevret S, et al. Prospective evaluation of gene mutations and minimal residual disease in patients with core binding factor acute myeloid leukemia. Blood. 2013;121(12):2213-2223. doi:10.1182/blood-2012-10-462879
6. Lane S, Saal R, Mollee P, et al. A > or =1 log rise in RQ-PCR transcript levels defines molecular relapse in core binding factor acute myeloid leukemia and predicts subsequent morphologic relapse. Leuk Lymphoma. 2008;49(3):517-523. doi:10.1080/10428190701817266
7. Yin JA, O'Brien MA, Hills RK, et al. Minimal residual disease monitoring by quantitative RT-PCR in core binding factor AML allows risk stratification and predicts relapse: results of the United Kingdom MRC AML-15 trial. Blood. 2012;120(14):2826-2835. doi:10.1182/blood-2012-06-435669

Performance

Method Description

Total RNA is extracted from blood or bone marrow and reverse transcribed to generate complementary DNA. Quantitative real-time polymerase chain reaction is performed, and the data analyzed using dedicated software for relative quantification with calibrator normalization. Results are provided as a normalized relative value of *RUNX1*::*RUNX1T1/ABL1* messenger RNA transcripts with a reproducible analytical sensitivity of 0.01%.(Unpublished Mayo method)

The normalized ratio is a relative quantification calculation as follows:

$$\text{Normalized ratio} = \frac{\textit{RUNX1}::\textit{RUNX1T1}\ (\text{sample})/\textit{ABL1}\ (\text{sample})}{\textit{RUNX1}::\textit{RUNX1T1}\ (\text{run calibrator})/\textit{ABL1}\ (\text{run calibrator})}$$

PDF Report

Supplemental

Day(s) Performed

Monday through Saturday

Report Available

4 to 8 days

Specimen Retention Time

Blood/Bone marrow: 2 weeks; Extracted RNA 3 months

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Test Definition: T821Q

RUNX1::RUNX1T1 Translocation (8;21), Minimal Residual Disease Monitoring, Quantitative, Varies

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

81401

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
T821Q	RUNX1/RUNX1T1, t(8;21), Quant, V	72207-4

Result ID	Test Result Name	Result LOINC® Value
MP061	Specimen Type	31208-2
615906	Interpretation	69047-9
616034	Signing Pathologist	18771-6