RUNX1-RUNX1T1 (*AML1-ETO*) t(8;21) Quantitative

Indications for Ordering

Detect and quantitate *RUNX1-RUNX1T1* fusions arising from t(8;21) in acute myeloid leukemia (AML)

Test Description

Quantitative reverse transcription polymerase chain reaction (PCR)

- Whole blood, bone marrow (BM)

Tests to Consider

Primary test

RUNX1-RUNX1T1 (AML1-ETO) t(8;21) Detection, Quantitative 2010138

- Can be used for minimal residual disease monitoring

Related tests

Acute Myeloid Leukemia Panel by FISH 2011132

- Diagnosis, prognosis, and monitoring of AML
- Includes

Probe Target	Gene(s)/Unique Sequence
t(15;17)(q24;q21)	*PML-RARA*
t(8;21)(q22;q22)	*RUNX1T1-RUNX1* (*ETO-AML1*)
inv(16)(p13.3q22)	*CBFB*
11q23	*KMT2A (MLL)*
inv(3) or t(3;3)	*RPN1-MECOM (EVI1)*
del(5)(q31)	*EGR1*
del(7)(q31)/-7	D7S486

Chromosome Analysis, Bone Marrow 2002292

- Diagnosis, prognosis, and monitoring of AML

Chromosome Analysis, Bone Marrow with Reflex to Genomic Microarray 2007130

- Diagnosis, prognosis, and monitoring of AML
- If chromosome analysis is "normal" or "no growth," then genomic microarray testing will be added

Cytogenomic SNP Microarray – Oncology 2006325

- Preferred test for fresh specimens at time of diagnosis for detecting prognostically important genomic abnormalities in leukemias/lymphomas and solid tumors involving
  - Loss/gain of DNA
  - Loss of heterozygosity (LOH)
- Monitor disease progression and response to therapy

Chromosome FISH, Interphase 2002298

- Specific FISH probe for t(8;21) must be requested

Myeloid Malignancies Mutation Panel by Next Generation Sequencing 2011117

- Assess for single gene mutations, including substitutions and insertions and deletions that may have diagnostic, prognostic, and/or therapeutic significance

KIT Mutations in AML by Fragment Analysis and Sequencing 2002437

- Prognostication in core-binding factor-related (CBF) AML (NCCN, 2011)

Disease Overview

Incidence – t(8;21) AML represents 5-7% of de novo adult AMLs and 11-13% of pediatric AMLs

Treatment issues

- Identification of t(8;21) AML may influence therapy and survival
- Use of FISH or PCR for identification of this translocation is important in treatment decisions
  - Quantitative test can be used in monitoring
- *KIT* mutation testing is also important since the presence of the mutation is associated with a worse outcome

Test Interpretation

Sensitivity/specificity

- Analytical sensitivity – 1 cell with t(8;21) in 1,000 normal cells
- Analytical specificity – 100%

Results

- Detected – *RUNX1-RUNX1T1* transcripts identified and quantitated
- Not detected – No *RUNX1-RUNX1T1* transcripts detected

Limitations

- "Not detected" does not exclude the possibility of *RUNX1-RUNX1T1* transcripts below the test limit of detection
- BM samples preferred for maximum sensitivity
- Poor RNA yield and/or quality due to sample age or hypocellularity will negatively impact the test