USEFUL FOR

- Second-tier testing in hematologic specimens when previous cytogenetic or FISH testing have detected an acquired chromosome abnormality of unknown significance
- Determining the size, precise breakpoints, gene content, and any unappreciated complexity of abnormalities detected by other methods such as conventional chromosome and FISH studies
- Providing important diagnostic, prognostic, and therapeutic information critical to proper patient management

GENETICS TEST INFORMATION

This testing is only appropriate in individuals with previously detected acquired chromosomal abnormalities. If previous testing was performed at another institution, supply a copy of those results. If sufficient information regarding the patient’s known chromosome abnormality is not made available, this testing will be cancelled.

CLINICAL INFORMATION

While many hematologic neoplasms (leukemias and lymphomas) have a subset of common or well-characterized acquired chromosome abnormalities, sometimes patients are found to have acquired chromosome abnormalities of uncertain significance. Further characterization of these abnormalities may lead to a better understanding of their pathogenicity and potentially lead to prognostic information or guide treatment and management of the patient.

Mate-pair sequencing is a next-generation sequencing technology that can aid in the further characterization of chromosome abnormalities by sequencing the entire genome and bioinformatically mapping short fragments of the genome to create a structural map of the genome. This technique enables the mapping of chromosome rearrangements to a resolution of approximately 2 kilobases or less, which allows for determination of genes at or near the breakpoints.

REFERENCE VALUES

An interpretative report will be provided.

ANALYTIC TIME

14 days

SPECIMEN REQUIRED

Submit only 1 of the following specimens:

**Type**
- Bone marrow

**Container/Tube**
- Green top (sodium heparin)

**Specimen Volume**
- 1–2 mL

**Type**
- Whole blood

**Container/Tube**
- Green top (sodium heparin)

**Specimen Volume**
- 7–10 mL

CONTENT AND VALUES SUBJECT TO CHANGE. SEE THE MAYO MEDICAL LABORATORIES TEST CATALOG FOR CURRENT INFORMATION.
INTERPRETATION

The interpretation describes the further characterization of the previously identified acquired abnormality. When possible, the interpretation will state how this finding might be associated with the hematologic process and any potential information on diagnosis, prognosis, and treatment options given the finding.

The continual discovery of novel structural rearrangements and published clinical reports means that the interpretation of any finding may evolve with increased scientific understanding.

Although the presence of a clonal abnormality usually indicates a neoplasia, in some situations it may reflect a benign or constitutional genetic change. If a genetic change is identified that is likely constitutional and clearly pathogenic, follow-up with a medical genetics consultation may be suggested.

The absence of an abnormal clone may be the result of specimen collection from a site that is not involved in the neoplasm or may indicate that the genetic abnormality is not detectable by this assay.

CLINICAL REFERENCE
