Overview

Useful For

Assisting in the diagnosis and classification of certain malignant hematological disorders

Evaluating the prognosis of patients with certain malignant hematologic disorders

Monitoring effects of treatment

Monitoring patients in remission

Reflex Tests

Test ID	Reporting Name	Available Separately	Always Performed
_ML20	Metaphases, 1-19	No, (Bill Only)	No
_M25	Metaphases, 20-25	No, (Bill Only)	No
_MG25	Metaphases, >25	No, (Bill Only)	No
_STAC	Ag-Nor/CBL Stain	No, (Bill Only)	No

Testing Algorithm

This test only includes a charge for professional interpretation of results and does not include charges for analysis.

Analysis charges will be incurred for total work performed, and generally include 2 banded karyograms and the analysis of 20 or more metaphase cells for this test. If no metaphase cells are available for analysis, no analysis charges will be incurred. If additional analysis work is required, additional charges may be incurred. See the Method Description for specific details.

The following algorithms are available in Special Instructions:

-Acute Promyelocytic Leukemia: Guideline to Diagnosis and Follow-up

-Bone Marrow Staging for Known or Suspected Malignant Lymphoma Algorithm

-Laboratory Screening Tests for Suspected Multiple Myeloma

-Myelodysplastic Syndrome: Guideline to Diagnosis and Follow-up

-Myeloproliferative Neoplasm: A Diagnostic Approach to Bone Marrow Evaluation

Special Instructions

- Laboratory Screening Tests for Suspected Multiple Myeloma
- Myeloproliferative Neoplasm: A Diagnostic Approach to Bone Marrow Evaluation
- Bone Marrow Staging for Known or Suspected Malignant Lymphoma Algorithm
- Myelodysplastic Syndrome: Guideline to Diagnosis and Follow-up
- Acute Promyelocytic Leukemia: Guideline to Diagnosis and Follow-up
- Informed Consent for Genetic Testing (Spanish)

Method Name

Chromosome Analysis on Fixed Cells

NY State Available

Yes

Specimen

Specimen Type

Varies

Necessary Information

A pathology and/or flow cytometry report may be requested by the Genomics Laboratory to optimize testing and aid in interpretation of results.

Specimen Required

Provide a reason for referral and specimen type with each specimen. The laboratory will notreject testing if this information is not provided, but appropriate testing and interpretation may be compromised or delayed.

Specimen Volume: 2 mL

Additional Information: Advise Express Mail or equivalent if not on courier service.

Forms

New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

Specimen Minimum Volume

1 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Refrigerated		

Clinical and Interpretive

Clinical Information

Chromosomal abnormalities play a central role in the pathogenesis, diagnosis, and treatment monitoring of many hematologic disorders. Cytogenetic studies on bone marrow may be helpful in many malignant hematologic disorders as the observation of a chromosomally abnormal clone may be consistent with a neoplastic process.

Certain chromosome abnormalities may help classify a malignancy. As examples, the Philadelphia (Ph) chromosome, also referred to as der(22)t(9;22)(q34;q11.2), is usually indicative of chronic myeloid leukemia (CML) or acute leukemia, t(8;21)(q22;q22) defines a specific subset of patients with acute myeloid leukemia, and t(8;14)(q24.1;q32) is associated with Burkitt lymphoma.

Cytogenetic studies are also used to monitor patients with hematologic neoplasia and may identify disease progression, such as the onset of blast crisis in CML, which is often characterized by trisomy 8, isochromosome 17q, and multiple Ph chromosomes.

Reference Values

An interpretative report will be provided.

Interpretation

To ensure the best interpretation, it is important to provide some clinical information to verify the appropriate type of cytogenetic study is performed.

The following factors are important when interpreting the results:

-Although the presence of an abnormal clone usually indicates a malignant neoplastic process, in rare situations, the clone may reflect a benign condition.

-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 disorder is caused by submicroscopic abnormalities that cannot be identified by chromosome analysis.

-On rare occasions, the presence of an abnormality may be associated with a congenital abnormality that is not related to a malignant neoplastic process. Follow-up with a medical genetics consultation is recommended.

-On occasion, bone marrow chromosome studies are unsuccessful. If clinical information has been provided, we may have a FISH study option that could be performed.

Cautions

In some cases, FISH studies may detect some disorders better than conventional chromosome studies:

Interfering factors

Technical:

-Not processing the bone marrow as indicated before shipping the specimen

-Not sending the first aspirate from the patient's bone marrow draw

Biological:

-Abnormalities missed due to sampling error

-Subtle structural chromosome abnormalities may be missed occasionally

-Neoplastic cells not dividing

Clinical Reference

1. Dewald GW, Ketterling RP, Wyatt WA, Stupca PJ: Cytogenetic studies in neoplastic hematologic disorders. In Clinical Laboratory Medicine. Second edition. Edited by KD McClatchey. Baltimore, Williams and Wilkens, 2002, pp 658-685

2. Rigolin GM, Cibien F, Martinelli S, et al: Chromosome aberrations detected by conventional karyotyping using novel mitogens in chronic lymphocytic leukemia with "normal" FISH: correlations with clinicobiological parameters. Blood 2012 Mar 8;119(10):2310-2313

Performance

Method Description

Metaphase cells are dropped onto microscope slides and are stained by G-banding. Other staining methods are employed as needed. Twenty metaphases are usually examined. If a clone is suspected, but not confirmed within 20 metaphases, 30 metaphases will be analyzed. Minimal evidence for the presence of an abnormal clone is defined as 2 or more metaphases with the same structural abnormality or chromosome gain (trisomy), or 3 or more metaphases lacking the same chromosome. All cells analyzed are captured using a computerized imaging system, and 1 or more karyograms from each clone are prepared to document the type of abnormality and to permit systematic interpretation of the anomalies.(College of American Pathologists. Accessed April 14, 2016 Available at http://www.cap.org)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

10 to 11 days

Specimen Retention Time

4 weeks

Performing Laboratory Location

Rochester

Fees and 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 Regional Manager. 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. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

88291

88264 w/modifier 52 (if appropriate)

88264 (if appropriate)

88264, 88285 (if appropriate)

88283 (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
CHFXH	Chromosomes, Hematol Fixed Cells	In Process

Result ID	Test Result Name	Result LOINC Value
38487	Result Summary	50397-9
38488	Interpretation	69965-2
38489	Result	62356-1
38490	Reason for Referral	42349-1
38491	Specimen	31208-2
38492	Source	31208-2
38493	Method	49549-9
38494	Banding Method	62359-5
38495	Additional Information	48767-8
38496	Released By	18771-6