Overview

Useful For

Assisting in the classification and follow-up of certain malignant hematological disorders when bone marrow is not available

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 includes a charge for cell culture of fresh specimens and professional interpretation of results. Analysis charges will be incurred for total work performed, and generally include 2 banded karyograms and the analysis of 20 metaphase cells. 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.

This test is not appropriate for detecting constitutional/congenital chromosome abnormalities. If this test is ordered with a reason for referral indicating a concern for a constitutional/congenital chromosome abnormality, the test will be cancelled and CHRCB / Chromosome Analysis, Congenital Disorders, Blood will be added and performed as the appropriate test.

If this test is ordered and the laboratory is informed that the patient is on a COG protocol, this test will be canceled and automatically reordered by the laboratory as COGBL / Chromosome Analysis, Hematologic Disorders, Childrenâ€™s Oncology Group Enrollment Testing, Blood.

Special Instructions

- Laboratory Screening Tests for Suspected Multiple Myeloma

Method Name

Cell Culture without Mitogens* followed by Chromosome Analysis*

*In addition to the cell culture without mitogens, a CpG stimulated culture will be added and 10 additional cells will be analyzed for any specimen received from a patient age 30 or older with a reason for referral of chronic lymphocytic leukemia (CLL), small lymphocytic lymphoma (SLL), lymphocytosis, Waldenstrom macroglobulinemia, or when CLLF / Chronic Lymphocytic Leukemia (CLL), FISH is ordered concurrently.

NY State Available

Yes

Specimen

Specimen Type

Whole blood

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

Container/Tube: Green top (sodium heparin)

Specimen Volume:5-10 mL

Collection Instructions:

1. Invert several times to mix blood.

2. Other anticoagulants are not recommended and are harmful to the viability of the cells.

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

Forms

If not ordering electronically, complete, print, and send a Hematopathology/Cytogenetics Test Request (T726) with the specimen.

Specimen Minimum Volume

3 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
Whole blood	Ambient (preferred)		
	Refrigerated		

Clinical and Interpretive

Clinical Information

Chromosomal abnormalities play a central role in the pathogenesis, diagnosis, and monitoring of treatment of many hematologic disorders. Whenever possible, it is best to do chromosome studies for neoplastic hematologic disorders on bone marrow. Bone marrow studies are more sensitive and the chances of finding metaphases are about 95%, compared with only a 60% chance for blood studies. When it is not possible to collect bone marrow, chromosome studies on blood may be useful.

When blood cells are cultured in a medium without mitogens, the observation of any chromosomally abnormal clone

may be consistent with a neoplastic process.

Conventional chromosome studies of B-cell disorders are not always successful because B-lymphocytes do not proliferate well in cell culture. The agent CpG 7909 (CpG) is a synthetic oligodeoxynucleotide that binds to the Toll-like receptor 9 (TLR9) present on B cells, causing B-cell activation. In the laboratory setting, CpG may be used as a mitogen to stimulate B-cells in patient specimens, thus allowing identification of chromosome abnormalities. CpG stimulation reveals an abnormal karyotype in approximately 80% of patients with of chronic lymphocytic leukemia (CLL), and the karyotype is complex in 20% to 25% of cases. Several studies have reported that increased genetic complexity revealed by CpG-stimulated chromosome studies confers a less favorable time to first treatment, treatment response, and overall survival.

See Laboratory Screening Tests for Suspected Multiple Myeloma in Special Instructions.

Reference Values

An interpretative report will be provided.

Interpretation

The presence of an abnormal clone usually indicates a malignant neoplastic process.

The absence of an apparent abnormal clone in blood may result from a lack of circulating abnormal cells and not from an absence of disease.

On rare occasions, the presence of an abnormality may be associated with a congenital abnormality and, thus, not related to a malignant process. When this situation is suspected, follow-up with a medical genetics consultation is recommended.

Cautions

We recommend consultation with personnel from the Cytogenetics Laboratory when considering blood studies for hematologic disorders.

Bone marrow specimens are preferred over peripheral blood specimens for the diagnosis of neoplastic hematologic disorders. When peripheral blood must be used, FISH studies may detect some disorders better than conventional chromosome studies.

FISH studies will detect chromosome anomalies with prognostic significance much more often than conventional chromosome studies for:

-Chronic lymphocytic leukemia (CLL)

-Plasma cell proliferative disorders (PCPDs) such as multiple myeloma

-FISH studies also may be superior for other hematological disorders when compared to conventional chromosome studies utilizing blood specimens.

This test is not useful for the following reasons and disorders: multiple miscarriages, infertility, pregnancy loss, multiple congenital anomalies, developmental delay, Down syndrome, Turner syndrome, premature ovarian failure, amenorrhea, ambiguous genitalia, and other congenital abnormalities. The appropriate test for these situations is CHRCB / Chromosome Analysis, Congenital Disorders, Blood.

Interfering factors:

Technical:

-Cell lysis caused by forcing blood quickly through the needle at collection

-Use of an improper anticoagulant (sodium heparin is best) or improperly mixing the blood with the anticoagulant

-Excessive transport time

-Exposure of the specimen to temperature

Biological:

-Abnormalities missed due to sampling error

-Subtle structural chromosome abnormalities may be missed occasionally

-Neoplastic cells not dividing or not circulating in the bloodstream

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

A cell count is performed to establish a plating volume. Based on the cell count, a corresponding volume of blood is added to 2 culture flasks containing culture medium and incubated for 24 to 48 hours at 37 degrees C. In the harvest process, the cells are exposed to colcemid and hypotonic solution, and are fixed with glacial acetic acid and methanol. 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 abnormality and to permit systematic interpretation of the anomalies.(Unpublished Mayo method)

When a specimen is received from a patient age 30 or older with a reason for referral of chronic lymphocytic leukemia (CLL), small lymphocytic lymphoma (SLL), lymphocytosis, or Waldenstrom macroglobulinemia, a CpG-stimulated culture will be added and 10 additional cells will be analyzed. Additional metaphases may be analyzed from the unstimulated or CpG-stimulated cell cultures if necessary to provide an accurate interpretation. All metaphases 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 abnormalities.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

9 to 11 days

Specimen Retention Time

Three 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 US Food and Drug Administration.

CPT Code Information

88237, 88291- Tissue culture for neoplastic disorders; bone marrow, blood, Interpretation and report

88264 w/ modifier 52-Chromosome analysis with less than 20 cells (if appropriate)

88264-Chromosome analysis with 20 to 25 cells (if appropriate)

88264,88285- Chromosome analysis with greater than 25 cells (if appropriate)

88283-Additional specialized banding technique (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
CHRHB	Chromosomes, Hematologic, Blood	62348-8

Result ID	Test Result Name	Result LOINC Value
52391	Result Summary	50397-9
52393	Interpretation	69965-2
52392	Result	82939-0
CG778	Reason for Referral	42349-1
52394	Specimen	31208-2
52395	Source	31208-2

Result ID	Test Result Name	Result LOINC Value
52397	Method	85069-3
52396	Banding Method	62359-5
54633	Additional Information	48767-8
52398	Released By	18771-6