MAYO CLINIC LABORATORIES

Test Definition: CHRHB

Chromosome Analysis, Hematologic Disorders, Blood

Overview

Useful For

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

This test is **not useful for** congenital disorders.

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, which generally includes 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.

In addition to the cell culture, a CpG-stimulated culture will be added and 10 additional cells will be analyzed for any specimen received from a patient 30 years or older with a reason for testing of chronic lymphocytic leukemia (CLL), small lymphocytic lymphoma, lymphocytosis, Waldenstrom macroglobulinemia, or any CLL fluorescence in situ hybridization test previously ordered or is ordered concurrently.

Special Instructions

- Multiple Myeloma: Laboratory Screening

Method Name

Cell Culture without Mitogens followed by Chromosome Analysis

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Ordering Guidance

This test is not appropriate for detecting constitutional/congenital chromosome abnormalities. If this test is ordered with a reason for testing indicating a concern for a constitutional/congenital chromosome abnormality, the test will be canceled 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 Children's Oncology Group (COG) protocol, this test will be canceled and automatically reordered by the laboratory as COGBL / Chromosome Analysis, Hematologic Disorders, Children's Oncology Group Enrollment Testing, Blood.

If this test is ordered concurrently with MDSDF / Myelodysplastic Syndrome (MDS), Diagnostic FISH, Varies, FISH testing will be held pending the results of this chromosome test. If the chromosome results are complete and informative, MDSDF will be canceled. MDS FISH testing is a second-tier test and should only be ordered if chromosome analysis is not successful, as it does not increase the sensitivity for detection of myelodysplastic syndrome (MDS) for classic abnormalities (ie, -5/5q-, -7/7q-). If a complete chromosome study is not achieved (<20 metaphases), MDS FISH testing will proceed. If an ambiguous abnormality (may include nonclonal abnormality or unresolved structural abnormality) is observed and targeted MDS probes could be useful in characterizing the abnormality, MDSDF test will be canceled and reordered with appropriate probes as MDSMF / Myelodysplastic Syndrome (MDS), Specified FISH, Varies.(1)

Consultation with personnel from the Cytogenetics Laboratory is recommended when considering blood studies for hematologic disorders. Call 800-533-1710 and ask for the Cytogenetics Genetic Counselor on call.

Shipping Instructions

Advise Express Mail or equivalent if not on courier service.

Necessary Information

1. A reason for testing should be submitted with each specimen. The laboratory will not reject testing if this information is not provided, but appropriate testing and interpretation may be compromised or delayed.
2. A pathology and/or flow cytometry report may be requested by the laboratory to optimize testing and aid in interpretation of results.
3. If a patient has received an opposite sex bone marrow transplant prior to specimen collection, note this information on the request.

Specimen Required

Container/Tube:
Preferred: Yellow top (ACD)
Acceptable: Green top (sodium heparin) or lavender top (EDTA)
Specimen Volume: 6 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not** aliquot.

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 & 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 chronic lymphocytic leukemia, 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.

For more information see Multiple Myeloma: Laboratory Screening.

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 that is not related to a malignant process. When this situation is suspected, consultation with a Clinical Geneticist is recommended.

Cautions

Interfering factors:
Technical:
-Cell lysis caused by forcing blood quickly through the needle at collection
-Use of an improper anticoagulant or improperly mixing the blood with the anticoagulant
-Clotted blood specimen
-Excessive transport time
-Exposure of the specimen to extreme temperature

Biological:
-Abnormalities missed due to sampling error
-Subtle structural chromosome abnormalities may not be detected by conventional chromosome analysis
-Neoplastic cells not dividing or not circulating in the bloodstream

Clinical Reference

1. He R, Wiktor AE, Durnick DK, et al. Bone marrow conventional karyotyping and fluorescence in situ hybridization: Defining an effective utilization strategy for evaluation of myelodysplastic syndromes. Am J Clin Pathol. 2016;146(1):86-94. doi:10.1093/ajcp/aqw077
2. Swerdlow et al. WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. IARC Press:Lyon, 2017
3. Dewald GW, Ketterling RP, Wyatt WA, Stupca PJ. Cytogenetic studies in neoplastic hematologic disorders. In: McClatchey KD, ed. Clinical Laboratory Medicine. 2nd ed. Williams and Wilkens; 2002:658-685
4. 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;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 then fixed with glacial acetic acid and methanol. Metaphase cells are applied to microscope slides and 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 abnormalities

When a specimen is received from a patient 30 years or older with a reason for testing noted as chronic lymphocytic leukemia, small lymphocytic lymphoma, lymphocytosis, or Waldenstrom macroglobulinemia, a CpG-stimulated culture will be added and 10 additional cells 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.(Arsham MS, Barch MJ, Lawce HJ. eds. The AGT Cytogenetics Laboratory Manual. 4th ed. Wiley-Blackwell, 2017)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
9 to 11 days

Specimen Retention Time
3 weeks

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

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
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 Definition: CHRHB

Chromosome Analysis, Hematologic Disorders, Blood

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
52397	Method	85069-3
52396	Banding Method	62359-5
54633	Additional Information	48767-8
52398	Released By	18771-6