MAYO CLINIC LABORATORIES

Test Definition: MISCF

Miscellaneous Studies Using Chromosome-Specific Probes, FISH

Overview

Useful For

Resolution of unusual or complex structural alterations, questionable mosaicism, and unbalanced chromosome abnormalities that cannot be resolved by chromosome or chromosomal microarray analysis

Identifying gain, loss, or rearrangement of chromosome regions using gene or locus-specific probes

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_PBCT	Probe, +2	No, (Bill Only)	No
_PADD	Probe, +1	No, (Bill Only)	No
_PB02	Probe, +2	No, (Bill Only)	No
_PB03	Probe, +3	No, (Bill Only)	No
_ML10	Metaphases, 1-9	No, (Bill Only)	No
_M30	Metaphases, >=10	No, (Bill Only)	No
_IL25	Interphases, <25	No, (Bill Only)	No
_I099	Interphases, 25-99	No, (Bill Only)	No
_I300	Interphases, >=100	No, (Bill Only)	No

Testing Algorithm

Consult with the laboratory before ordering this test.

This test includes a charge for the probe application, analysis, and professional interpretation of results for 1 probe set (2 individual fluorescence in situ hybridization probes). Additional charges will be incurred for all additional probe sets performed. If no cells are available for analysis, no analysis charges will be incurred.

Method Name

Fluorescence In Situ Hybridization (FISH)

NY State Available

No

Specimen

Specimen Type

Varies

Ordering Guidance

Consult with the laboratory before ordering this test.

The fluorescence in situ hybridization (FISH) probes to be analyzed must be specified on the request when ordering, otherwise test processing may be delayed in order to determine the intended analysis. If specific probes are not provided, this test may be canceled by the laboratory.

If testing bone marrow or blood samples for specific hematologic malignancies is desired, order HEMMF / Hematologic Specified FISH, Varies. If specific FISH probes for hematologic malignancies are ordered and a bone marrow or blood sample is received, this test will be canceled and automatically reordered by the laboratory as HEMMF / Hematologic Specified FISH, Varies.

Shipping Instructions

Advise Express Mail or equivalent if not on courier service.

Necessary Information

1. **A list of probes requested for analysis is required**.
2. 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. If this information is not provided, an appropriate indication for testing may be entered by Mayo Clinic Laboratories.
3. A pathology report may be requested by the laboratory to optimize testing and aid in the interpretation of results. Acceptable pathology reports include working drafts, preliminary pathology, or surgical pathology reports.

Specimen Required

Submit only 1 of the following specimens:

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20-25 mL
Collection Instructions:
1. Optimal timing for specimen collection is during 14 to 18 weeks of gestation, but specimens collected at other weeks of gestation are also accepted. Provide gestational age at the time of amniocentesis.
2. Discard the first 2 mL of amniotic fluid.
Additional Information:
1. Unavoidably, about 1% to 2% of mailed-in specimens are not viable.
2. Bloody specimens are undesirable.
3. If the specimen does not grow in culture, you will be notified within 7 days of receipt.
4. Results will be reported and telephoned or faxed if requested.

Specimen Type: Blood (only accepted for Congenital/Hereditary [nonhematologic] testing)
Container/Tube:
Preferred: Yellow top (ACD)
Acceptable: Green top (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.**
3. Other anticoagulants are not recommended and are harmful to the viability of the cells.

Specimen Type: Chorionic villi (CVS)
Supplies: CVS Media (RPMI) and Small Dish (T095)
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 to 25 mg
Collection Instructions:
1. Collect specimen by the transabdominal or transcervical method.
2. Transfer chorionic villi to a Petri dish containing transport medium (eg, CVS media (RPMI)).
3. Using a stereomicroscope and sterile forceps, assess the quality and quantity of the villi and remove any blood clots and maternal decidua.

Specimen Type: Lymph node
Supplies: Hank's Solution (T132)
Container/Tube: Sterile container with sterile Hank's balanced salt solution, Ringer's solution, or normal saline
Specimen Volume: 1 cm(3)

Specimen Type: Skin biopsy
Supplies: Hank's Solution (T132)
Container/Tube: Sterile container with sterile Hank's balanced salt solution, Ringer's solution, or normal saline
Specimen Volume: 1-cm(3) biopsy specimen of muscle/fascia from the thigh
Collection Instructions:
1. Wash biopsy site with an antiseptic soap.
2. Thoroughly rinse area with sterile water.
3. Do not use alcohol or iodine preparations.
4. A local anesthetic may be used.
5. Biopsy specimens are best taken by punch biopsy to include full thickness of dermis.

Specimen Type: Tissue
Preferred: Tissue block
Collection Instructions: Submit a formalin-fixed, paraffin-embedded tumor tissue block. Blocks prepared with alternative fixation methods may be acceptable; provide fixation method used.
Additional Information:
1. Paraffin embedded specimens can be from any anatomic location (skin, soft tissue, lymph node, etc).
2. Bone specimens that have been decalcified will be attempted for testing, but the success rate is approximately 50%.

Acceptable: Slides
Collection Instructions: For each probe set ordered, 4 consecutive, unstained, 5 micron-thick sections placed on positively charged slides. Include 1 hematoxylin and eosin-stained slide for the entire test order.

Specimen Type: Tumor
Supplies: Hank's Solution (T132)
Container/Tube: Sterile container with sterile Hank's balanced salt solution, Ringer's solution, or normal saline

Specimen Volume: 0.5 to 3 cm(3) or larger

Specimen Minimum Volume
Amniotic fluid: 5 mL; Blood: 2 mL; Chorionic villi: 5 mg; Lymph node: 0.5 cm(3); Solid tumor: 0.5 cm(3)

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 & Interpretive

Clinical Information
Conventional cytogenetic studies can identify the presence of chromosome abnormalities and most mosaic conditions. In approximately 2% of these chromosomally abnormal cases, the genetic makeup of the chromosome abnormality can be identified, but not completely characterized, by conventional techniques alone. For malignant disorders, the proportion of specimens with unresolvable chromosome abnormalities is much higher. Chromosomal microarray analysis (CMA) can detect copy number gain or loss of a chromosomal region but cannot identify the mechanism.

Fluorescence in situ hybridization using gene-specific probes and various probe strategies can help characterize chromosome abnormalities. This includes abnormalities that cannot be accurately characterized by chromosome analysis or CMA (such as unusual structural alterations) and unbalanced chromosome abnormalities (such as deletions, duplications, and translocations). Scoring large numbers of interphase nuclei can more accurately establish the frequency of chromosome abnormalities.

Reference Values
An interpretive report will be provided.

Interpretation
An interpretive report will be provided.

Cautions
This test should not be ordered without prior consultation and approval by the laboratory.

This test is not approved by the US Food and Drug Administration, and it is best used as an adjunct to existing clinical and pathologic information.

Clinical Reference
1. Remstein ED, Dogan A, Einerson RR, et al: The incidence and anatomic site specificity of chromosomal translocations in primary extranodal marginal zone B-cell lymphoma of mucosa-associated lymphoid tissue (MALT lymphoma) in North

America. Am J Surg Pathol. 2006 Dec;30(12):1546-1553
2. Fonseca R, Blood E, Rue M, et al: Clinical and biologic implications of recurrent genomic aberrations in myeloma. Blood. 2003 Jun 1;101(11):4569-4575
3. Van Dyke DL, Shanafelt TD, Call TG, et al: A comprehensive evaluation of the prognostic significance of 13q deletions in patients with B-chronic lymphocytic leukaemia. Br J Haematol. 2010 Feb;148(4):544-550
4. Wiktor A, Van Dyke DL: FISH analysis helps identify low-level mosaicism in Ullrich-Turner syndrome patients. Genet Med. 2004 May-Jun;6(3):132-135
5. Swerdlow SH, Campo E, Harris NL, eds, et al: WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. IARC; 2017
6. American College of Medical Genetics and Genomics (ACMG): Technical Standards and Guidelines for Clinical Genetics Laboratories. ACMG; 2008. Updated January 2018. Accessed April 28, 2023. Available at www.acmg.net/PDFLibrary/Standards-Guidelines-Cytogenetics.pdf

Performance

Method Description
This test is performed using commercially available and laboratory-developed probes. Depending on the indication for testing, analysis of metaphase cells or interphase nuclei is performed. Two technologists analyze each probe set, and all results are reported indicating presence, absence, or rearrangement of the gene region being interrogated. If interphase nuclei are scored, the results are expressed as the percent abnormal nuclei.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
9 to 10 days

Specimen Retention Time
Amniotic fluid (remaining supernatant/whole fluid aliquots): 14 days after report; Blood: 4 weeks; Products of Conception (identifiable fetal tissue): Cremated quarterly after results reported; All other specimens: Not retained

Performing Laboratory Location
Rochester

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 using an analyte specific reagent. Its performance characteristics were 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

88271x2, 88291-DNA probe, each (first probe set), Interpretation and report
88271x2-DNA probe, each; each additional probe set (if appropriate)
88271x1-DNA probe, each; coverage for sets containing 3 probes (if appropriate)
88271x2-DNA probe, each; coverage for sets containing 4 probes (if appropriate)
88271x3-DNA probe, each; coverage for sets containing 5 probes (if appropriate)
88273 w/modifier 52-Chromosomal in situ hybridization, less than 10 cells (if appropriate)
88273-Chromosomal in situ hybridization, 10-30 cells (if appropriate)
88274 w/modifier 52-Interphase in situ hybridization, <25 cells, each probe set (if appropriate)
88274-Interphase in situ hybridization, 25 to 99 cells, each probe set (if appropriate)
88275-Interphase in situ hybridization, 100 to 300 cells, each probe set (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MISCF	Miscellaneous Studies, FISH	62367-8

Result ID	Test Result Name	Result LOINC® Value
52163	Result Summary	50397-9
52165	Interpretation	69965-2
52164	Result Table	93356-4
54586	Result	62356-1
CG746	Reason for Referral	42349-1
CG943	Specimen	31208-2
52167	Source	31208-2
52168	Tissue ID	80398-1
52169	Method	85069-3
55028	Additional Information	48767-8
52170	Released By	18771-6
53829	Disclaimer	62364-5