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,	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 application of the first probe set (2 FISH probes) and professional interpretation of results. Additional charges will be incurred of all reflex probes performed. Analysis charges will be incurred based on the number of cells analyzed per probe set. 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

Specimen Required

Provide a reason for referral 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.

Advise Express Mail or equivalent if not on courier service.

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. Place the tubes in a Styrofoam container (T329).

2. Fill remaining space with packing material.

3. Unavoidably, about 1% to 2% of mailed-in specimens are not viable.

4. Bloody specimens are undesirable.

5. If the specimen does not grow in culture, you will be notified within 7 days of receipt.

6. Results will be reported and also telephoned or faxed, if requested.

Specimen Type: Blood

Container/Tube: Green top (sodium heparin)

Specimen Volume: 5 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.

Specimen Type: Bone marrow

Container/Tube: Green top (sodium heparin)

Specimen Volume: 1-2 mL

Collection Instructions:

1. Invert several times to mix bone marrow.

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

Specimen Type: Chorionic villi

Container/Tube: 15-mL tube containing 15 mL of transport media

Specimen Volume: 20-30 mg

Collection Instructions:

1. Collect specimen by the transabdominal or transcervical method.

2. Transfer the chorionic villi to a Petri dish containing transport medium.

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

Container/Tube: Sterile container with sterile Hank's balanced salt solution (T132), Ringer's solution, or normal saline.

Specimen Volume: 1 cm(3)

Specimen Type: Skin biopsy

Container/Tube: Sterile container with sterile Hank's balanced salt solution (T132), Ringer's solution, or normal saline.

Specimen Volume: 4-mm diameter

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 block or slide

Preferred: Formalin-fixed, paraffin-embedded tumor tissue block and 1 hematoxylin and eosin (H and E)-stained slide.

Acceptable: Four consecutive, unstained, 5-micron thick sections placed on positively charged slides and 1 H and E-stained slide.

Specimen Type: Tumor

Container/Tube: Sterile container with sterile Hank's balanced salt solution (T132), Ringer's solution, or normal saline.

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

Specimen Minimum Volume

Amniotic Fluid: 5 mL/Blood: 2 mL/Bone Marrow: 1 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 and 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.

FISH 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 and assess level of mosaicism.

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 U.S. 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

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;148:544-550

4. Wiktor A, Van Dyke DL: FISH analysis helps identify low-level mosaicism in Ullrich-Turner syndrome patients. Genet Med 2004;6:132-135

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) and Time(s) Test Performed

Samples processed Monday through Sunday. Results reported Monday through Friday; 8 a.m. to 5 p.m. CST.

Analytic Time

9 days

Maximum Laboratory Time

10 days

Specimen Retention Time

Amniotic Fl. (remaining supernatant/whole fluid aliquots): Discarded 14 days after report. Blood: 4 weeks. Products of Conception (identifiable fetal tissue): Cremated quarterly after results reported. All Other Specimens: Discarded when results reported.

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 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 U.S. Food and Drug Administration.

CPT Code Information

88271x2, 88291 Ã¢â‚¬â€œ DNA probe, each (first probe set), Interpretation and report

88271x2 Ã¢â‚¬â€œ DNA probe, each; each additional probe set (if appropriate)

88271x1 Ã¢â‚¬â€œ DNA probe, each; coverage for sets containing 3 probes (if appropriate)

88271x2 Ã¢â‚¬â€œ DNA probe, each; coverage for sets containing 4 probes (if appropriate)

88271x3 Ã¢â‚¬â€œ 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 Ã¢â‚¬â€œ Interphase in situ hybridization, <25 cells, each probe set (if appropriate)

88274 Ã¢â‚¬â€œ Interphase in situ hybridization, 25 to 99 cells, each probe set (if appropriate)

88275 Ã¢â‚¬â€œ 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	51991-8

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	49549-9
55028	Additional Information	48767-8
53829	Disclaimer	62364-5
52170	Released By	18771-6