Test Definition: MFCDF

Myeloma, High Risk with Reflex Probes,
Diagnostic FISH Evaluation, Fixed Cell Pellet

Overview

Useful For

Detecting, at diagnosis, recurrent high-risk common chromosome abnormalities associated with multiple myeloma or other plasma cell proliferative disorders, when fresh bone marrow is unavailable, using a laboratory-designated probe set algorithm

Evaluating specimens in which the bone marrow is past 96 hours from collection

This test **should not be used** to track the progression of disease

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
MFCDB	Probe, Each Additional (MFCDF)	No, (Bill Only)	No
_STR1	Comp Analysis using STR (Bill only)	No, (Bill only)	No
_STR2	Add'l comp analysis w/STR (Bill Only)	No, (Bill only)	No

Testing Algorithm

This test includes a charge for probe application, analysis, and professional interpretation of results for 3 probe sets (6 individual fluorescence in situ hybridization [FISH] probes). Additional charges will be incurred for all reflex or additional probe sets 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.

This test is designed for diagnostic bone marrow specimens from patients with multiple myeloma, or other plasma cell proliferative disorders, when either a fixed cell pellet or a bone marrow sample exceeding 96 hours post-collection is available. Best results are obtained when the bone marrow demonstrates at least 20% involvement by a plasma cell proliferative disorder.

The high-risk myeloma FISH panel includes testing for the following abnormalities using the FISH probes listed:
1p deletion/1q gain, CDKN2C/1q22 probe set
t(14q32;var) or *IGH* rearrangement, IGH break-apart probe set
-17/17p-, TP53/D17Z1 probe set

If an *IGH* rearrangement is identified, appropriate reflex testing will be performed in an attempt to identify the translocation partner using the FISH probes listed:
t(4;14)(p16.3;q32) *IGH::FGFR3* fusion, FGFR3/IGH probe set
t(11;14)(q13;q32) or *IGH::CCND1* fusion, CCND1/IGH probe set
t(14;16)(q32;q23) *IGH::MAF* fusion, IGH/MAF probe set
t(14;20)(q32;q12) *IGH::MAFB* fusion, IGH/MAFB probe set

Appropriate ancillary probes may be performed at consultant discretion to render comprehensive assessment. Any additional probes will have the results included within the final report and will be performed at an additional charge.

Method Name
Fluorescence In Situ Hybridization (FISH)

NY State Available
Yes

Specimen

Specimen Type
Fixed Cell Pellet Bone Marrow

Ordering Guidance
For the most complete genetic evaluation on fresh bone marrow specimens, order MSMRT/ Mayo Algorithmic Approach for Stratification of Myeloma and Risk-Adapted Therapy Report, Bone Marrow.

For evaluation of high-risk abnormalities, with reflex probes, on fresh bone marrow specimens that will be received within 96 hours of collection, order PCPDS / Plasma Cell Proliferative Disorder, High Risk with Reflex Probes, Diagnostic FISH Evaluation, Bone Marrow. If the specimen received for this test is within 96 hours of collection, this test will be canceled and automatically reordered by the laboratory as PCPDS.

For testing paraffin-embedded tissue specimens from patients with a plasma cell disorder, order PLASF / Plasma Cell Proliferative Disorder, FISH, Tissue. If the specimen received for this test is paraffin-embedded, this test will be canceled and automatically reordered by the laboratory as PLASF.

Fresh bone marrow is only acceptable specimen for this test if the specimen will be received 96 hours or more post-collection.

Shipping Instructions
Advise Express Mail or equivalent if not on courier service.

Necessary Information

1. A list of probes requested for analysis is required if select probes are necessary or if the patient is being tracked for known abnormalities. Probes available for this test are listed in the Testing Algorithm section.
2. A reason for testing must be provided. If this information is not provided, an appropriate indication for testing may be entered by Mayo Clinic Laboratories.
3. A flow cytometry and/or a bone marrow pathology report 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.

Specimen Required

Container/Tube: Sterile container
Specimen Volume: 1 Fixed cell pellet
Collection Instructions: Place specimen in a sterile container with a 3:1 methanol:glacial acetic acid (or similar) fixative.

Forms

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

Reject Due To

Fresh tissue	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Fixed Cell Pellet Bone Marrow	Ambient (preferred)		
	Refrigerated		

Clinical & Interpretive

Clinical Information

Multiple myeloma is a hematologic neoplasm that generally originates in the bone marrow and develops from malignant plasma cells. There are 4 main categories of plasma cell proliferative disorders: monoclonal gammopathy of undetermined significance (MGUS), monoclonal immunoglobulin deposition diseases (amyloidosis), plasmacytoma, and multiple myeloma. MGUS, which occurs in 3% to 4% of individuals older than 50 years, represents the identification of an asymptomatic monoclonal protein, yet approximately 1% per year will progress to multiple myeloma. Amyloidosis represents a rare group of deposition disorders including primary amyloidosis vs. light chain and heavy chain disease. Plasmacytomas represent isolated collections of bone or extramedullary plasma cells with a risk for development of multiple myeloma. Generalized bone pain, anemia, limb numbness, or weakness, symptoms of hypercalcemia, and recurrent infections are all symptoms that may indicate multiple myeloma.

As myeloma progresses, the malignant plasma cells interfere with normal blood product formation in the bone marrow resulting in anemia and leukopenia. Myeloma also causes an overstimulation of osteoclasts, causing excessive

breakdown of bone tissue without the normal corresponding bone formation. These bone lesions are seen in approximately 66% of myeloma patients. In advanced disease, bone loss may reach a degree where the patient suffers fractures easily.

Multiple myeloma is increasingly recognized as a disease characterized by marked cytogenetic, molecular, and proliferative heterogeneity. This heterogeneity is manifested clinically by varying degrees of disease aggressiveness. Multiple myeloma patients with more aggressive disease experience suboptimal responses to some therapeutic approaches; therefore, identifying these patients is critically important for selecting appropriate treatment options.

Reference Values

An interpretive report will be provided.

Interpretation

A neoplastic clone is detected when the percentage of cells with an abnormality exceeds the normal reference range for any given probe set.

The absence of an abnormal clone does not rule out the presence of a plasma cell clone or another neoplastic disorder.

Cautions

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.

If no fluorescence in situ hybridization (FISH) signals are observed post-hybridization, the case will be released indicating a lack of FISH results.

Clinical Reference

1. WHO Classification of Tumours Editorial Board, eds. Haematolymphoid Tumours. 5th ed. IARC Press; 2024:603-630. WHO Classification of Tumours. Vol 11
2. Arber D., Borowitz, Cook J, et al. The International Consensus Classification of Myeloid and Lymphoid Neoplasms. Wolters Kluwer; 2025:384-396
3. Kumar SK, Rajkumar SV. The multiple myelomas-current concepts in cytogenetic classification and therapy. Nat Rev Clin Oncol. 2018;15(7):409-421. doi:10.1038/s41571-018-0018-y
4. Lu X, Andersen EF, Banerjee R, et al. Guidelines for the testing and reporting of cytogenetic results for risk stratification of multiple myeloma: a report of the Cancer Genomics Consortium Plasma Cell Neoplasm Working Group. Blood Cancer J. 2025;15(1):86
5. Gagnon MF, Midthun SM, Fangel JA, et al. Superior detection rate of plasma cell FISH using FACS-FISH. Am J Clin Pathol. 2024;161(1):60-70. doi:10.1093/ajcp/aqad108

Performance

Method Description

This test is performed using commercially available and laboratory-developed probes. Deletion of the *TP53* locus from chromosome 17 or monosomy 17 and deletion of the *CDKN2C* locus or gain of the 1q22 locus are detected using

enumeration strategy probes. Rearrangements involving *IGH* are detected using dual-color break-apart (BAP) strategy probes. Dual-color, dual-fusion fluorescence in situ hybridization (D-FISH) strategy probe sets are used when a rearrangements of the *IGH* gene is detected. For enumeration and BAP strategy probe sets, 100 interphase nuclei are scored; 200 interphase nuclei are scored when D-FISH probes are used. All results are expressed as the percent abnormal nuclei.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
7 to 10 days

Specimen Retention Time
4 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
88271x6, 88275x3, 88291 x1-FISH Probe, Analysis, Interpretation; 3 probe sets
88271x2, 88275x1-FISH Probe, Analysis; each additional probe set (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MFCDF	Myeloma Fixed Cell, High Risk, FISH	In Process

Result ID	Test Result Name	Result LOINC® Value
614300	Result Summary	50397-9

614301	Interpretation	69965-2
614302	Result Table	93356-4
614303	Result	62356-1
GC128	Reason for Referral	42349-1
614304	Specimen	31208-2
614306	Method	85069-3
614307	Additional Information	48767-8
614308	Disclaimer	62364-5
614309	Released By	18771-6
614305	Source	31208-2