Test Definition: MDSMF

Myelodysplastic Syndrome (MDS), Specified FISH, Varies

Overview

Useful For

Detecting recurrent common chromosome abnormalities associated with myelodysplastic syndromes (MDS) or other myeloid malignancies using **client-specified** probe set(s)

As an adjunct to conventional chromosome studies in patients with MDS

Evaluating specimens in which chromosome studies are unsuccessful

Identifying and tracking known chromosome abnormalities in patients with MDS and monitoring response to therapy

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
MDSMB	Probe, Each Additional (MDSMF)	No, (Bill Only)	No

Testing Algorithm

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 [FISH] probes). Additional charges will be incurred for all reflex, if requested, 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 performed using client-specified FISH probes and is not intended as a panel test. Specific probes must be listed in the probe request field. Reflex probes can be performed when appropriate if specified in the order request field.

When specified, any of the following probes will be performed:
inv(3) or t(3;3) or *GATA2::MECOM* fusion, request probe GATA2/MECOM
t(1;3)(p36;q21) or *GATA2::PRDM16* fusion, request probe PRDM16/GATA2
t(3q26.2;var) or 3q26.2 rearrangement, request probe MECOM break-apart
-5/5q-, request probe D5S630/EGR1
-7/7q-, request probe D7Z1/D7S486
+8, request probe D8Z2/MYC
-17/17p-, request probe TP53/D17Z1
-20/20q-, request probe D20S108/20qter

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
Varies

Ordering Guidance
This test is intended for instances when **limited** myelodysplastic syndrome (MDS) fluorescence in situ hybridization (FISH) probes are needed based on specific abnormalities or abnormalities identified in the diagnostic sample. **The FISH probes to be analyzed must be specified on the ordering request.** If targeted FISH probes are not included with this test order, test processing will be delayed and the test may be canceled and automatically reordered by the laboratory as MDSDF / Myelodysplastic Syndrome (MDS), Diagnostic FISH, Varies.

Chromosome analysis is recommended as first-tier testing; order either CHRBM / Chromosome Analysis, Hematologic Disorders, Bone Marrow, or CHRHB / Chromosome Analysis, Hematologic Disorders, Blood. This second-tier test should only be ordered if chromosome analysis is not successful, as it does not increase the sensitivity for detection of MDS for classic abnormalities (ie, -5/5q-, -7/7q-).

If a complete MDS FISH panel is preferred, order MDSDF / Myelodysplastic Syndrome (MDS), Diagnostic FISH, Varies.

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

Necessary Information
1. 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.
2. 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
Submit only 1 of the following specimens:

Preferred:
Specimen Type: Bone marrow
Container/Tube:
Preferred: Yellow top (ACD)
Acceptable: Green top (sodium heparin) or lavender top (EDTA)

Specimen Volume: 2 to 3 mL
Collection Instructions:
1. It is preferable to send the first aspirate from the bone marrow collection.
2. Invert several times to mix bone marrow.
3. Send bone marrow in original tube. **Do not aliquot.**

Acceptable:
Specimen Type: Whole blood
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 in original tube. **Do not aliquot.**

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

Specimen Minimum Volume
Bone marrow: 1 mL; Whole blood: 2 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
Varies	Ambient (preferred)		
	Refrigerated		

Clinical & Interpretive

Clinical Information
Myelodysplastic syndromes (MDS) primarily occur in the older adult population and have a yearly incidence of 30 in 100,000 in persons older than 70 years. These disorders are typically associated with a hypercellular bone marrow and low peripheral blood counts, and with significant morbidity and mortality. The eventual clinical outcome for patients with MDS relates to either bone marrow failure or transformation to acute myeloid leukemia. MDS can be either primary (de novo) or secondary (due to previous treatment with alkylating or etoposide chemotherapy, with or without radiation).

Cytogenetic studies can provide confirmatory evidence of clonality in MDS and can be used to provide clinical prognostic or diagnostic information. Clonal cytogenetic abnormalities are more frequently observed in cases of secondary MDS

(80% of patients) than in primary MDS (40%-60% of patients). The common chromosomal abnormalities associated with MDS include: inv(3), -5/5q-, -7/7q-, +8, and 20q-. These abnormalities can be observed singly or in concert.

Conventional chromosome analysis is the gold standard for identification of the common, recurrent chromosome abnormalities in MDS; however, some of the subtle rearrangements associated with secondary MDS can be missed.

Fluorescence in situ hybridization (FISH) analysis of nonproliferating (interphase) cells can be used to detect the common diagnostic and prognostic chromosome abnormalities observed in patients with MDS. When recurrent translocations or inversions are identified, FISH testing can also be used to track response to therapy.

Reference Values
An interpretive report will be provided.

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

The absence of an abnormal clone does not rule out the presence of a 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.

Fluorescence in situ hybridization (FISH) is not a substitute for conventional chromosome studies because the latter detects chromosome abnormalities associated with other hematological disorders that would go undetected in a targeted myelodysplastic syndromes FISH test.

Bone marrow is the preferred specimen type for this FISH test. If bone marrow is not available, a blood specimen may be used if there are neoplastic cells in the blood specimen (as verified by a hematopathologist).

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

Clinical Reference
1. Bernasconi P, Klersy C, Boni M, et al. World Health Organization classification in combination with cytogenetic markers improves the prognostic stratification of patients with de novo primary myelodysplastic syndromes. Br J Haematol. 2007;137(3):193-205
2. Swerdlow SH, Campo E, Harris NL, et al, eds. WHO Classification of Tumour of Haematopoietic and Lymphoid Tissues. 4th ed. IARC Press; 2017
3. 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

Performance

Method Description

This test is performed using commercially available and laboratory-developed probes. Deletion or monosomy of chromosomes 5, 7, 17, and 20, and trisomy of chromosome 8 are detected using enumeration strategy probe sets. Rearrangements involving *MECOM* are detected using a dual-color break-apart (BAP) strategy probe set. Dual-color, dual-fusion fluorescence in situ hybridization (D-FISH) strategy probe sets are used to detect inv(3) and t(1;3). For the 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

88271 x2, 88275 x1, 88291 x1- FISH Probe, Analysis, Interpretation; 1 probe set
88271 x2, 88275 x1 – FISH Probe, Analysis; each additional probe set (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MDSMF	MDS, Specified FISH	62367-8

Test Definition: MDSMF

Myelodysplastic Syndrome (MDS), Specified FISH, Varies

Result ID	Test Result Name	Result LOINC® Value
614289	Result Summary	50397-9
614290	Interpretation	69965-2
614291	Result Table	93356-4
614292	Result	62356-1
GC124	Reason for Referral	42349-1
GC125	Probes Requested	78040-3
GC126	Specimen	31208-2
614293	Source	31208-2
614294	Method	85069-3
614295	Additional Information	48767-8
614296	Disclaimer	62364-5
614297	Released By	18771-6