Test Definition: MDSDF

Myelodysplastic Syndrome (MDS), Diagnostic FISH, Varies

Overview

Useful For

Detecting recurrent common chromosome abnormalities associated with myelodysplastic syndromes or other myeloid malignancies using a panel

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
MDSDB	Probe, Each Additional (MDSDF)	No, (Bill Only)	No

Testing Algorithm

This test includes a charge for the probe application, analysis, and professional interpretation of results for 6 probe sets (12 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 performed as panel testing only using the following analysis algorithm.

The panel includes testing for the following abnormalities using the probes listed:
inv(3) or t(3;3) or *GATA2::MECOM* fusion, GATA2/MECOM probe set
-5/5q-, D5S630/EGR1 probe set
-7/7q-, D7Z1/D7S486
+8, D8Z2/MYC probe set
-17/17p-, TP53/D17Z1 probe set
-20/20q-, D20S108/20qter probe set

Appropriate ancillary probes may be performed at consultant discretion to render comprehensive assessment. Any additional probes used will have the results included within the final report and will be performed at an additional charge. In the following situations, additional (reflex) testing may be performed at the laboratory's discretion and may be influenced by available karyotype results or other FISH testing.

In the absence of *GATA2::MECOM* fusion, when an extra GATA2 signal is identified, testing using the PRDM16/GATA2 probe set may be performed to identify a potential t(1;3)(p36;q21).

In the absence of *GATA2::MECOM* fusion, when an extra MECOM signal is identified, testing using the break-apart MECOM probe set may be performed to identify a potential variant translocation involving *MECOM*, t(3;var)(q26.2;?).

Method Name
Fluorescence In Situ Hybridization (FISH)

NY State Available
Yes

Specimen

Specimen Type
Varies

Ordering Guidance
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 myelodysplastic syndrome (MDS) for classic abnormalities (ie, -5/5q-, -7/7q-). If this test is ordered concurrently with a chromosomal study (CHRBM or CHRHB), testing will be held pending the results of the chromosome test. If the chromosome results are complete and informative, this test will be canceled. If the chromosome results are complete and normal, this test will be canceled. If a complete chromosome study is not achieved (<20 metaphases), this test 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, this test will be canceled and reordered with appropriate probes as MDSMF / Myelodysplastic Syndrome (MDS), Specified FISH, Varies.

This test **should not be used** to screen for residual MDS. If the patient is being treated for known abnormalities, MDSMF / Myelodysplastic Syndrome (MDS), Specified FISH, Varies is the more appropriate test order.

If targeted MDS fluorescence in situ hybridization (FISH) probes are preferred, order MDSMF / Myelodysplastic Syndrome (MDS), Specified FISH and request specific probes for targeted abnormalities.

This test is intended for instances when the entire MDS FISH panel is needed as a second-tier test. If limited MDS FISH probes are preferred, order MDSMF.

If this test is ordered in conjunction with AMLFA / Adult Acute Myeloid Leukemia Panel, FISH, Varies or AMLFP / Pediatric Acute Myeloid Leukemia Panel, FISH, Varies, it will be canceled and reordered as MDSMF to avoid duplicate FISH probe testing.

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. In addition, t(1;3) and t(3;21) are more frequently associated with secondary MDS.

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.

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 panel 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 Tumours of Haematopoietic and Lymphoid Tissues. 4th ed. IARC Press; 2017. WHO Classification of Tumours. Vol 2
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.

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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 x 12, 88275 x 6, 88291-FISH Probe, Analysis, Interpretation; 6 probe sets
88271 x 2, 88275-FISH Probe, Analysis; each additional probe set (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MDSDF	MDS, Diagnostic FISH	62367-8

Result ID	Test Result Name	Result LOINC® Value
614278	Result Summary	50397-9
614279	Interpretation	69965-2
614280	Result Table	93356-4
614281	Result	62356-1
GC121	Reason for Referral	42349-1
GC122	Specimen	31208-2
614282	Source	31208-2
614283	Method	85069-3
614284	Additional Information	48767-8
614285	Disclaimer	62364-5
614286	Released By	18771-6