Test Definition: MDSDF

Myelodysplastic Syndrome (MDS), Diagnostic FISH, Varies

Overview

Useful For

Detecting a neoplastic clone associated with the common chromosome abnormalities seen in patients with myelodysplastic syndromes or other myeloid malignancies using a laboratory-designated probe set algorithm

Evaluating specimens in which standard cytogenetic analysis is unsuccessful

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 probes). Additional charges will be incurred for all reflex or additional probe sets performed.

Panel includes testing for the following abnormalities using the probes listed:
-inv(3) or t(3;3), RPN1/MECOM
-5/5q-, D5S630/EGR1
-7/7q-, D7S486/D7Z1
+8, D8Z2/MYC
-17p-, TP53/D17Z1
-20/20q-, 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.

In the absence of *RPN1::MEC*OM fusion, when an extra MECOM signal is identified*,* reflex testing using the MECOM/RUNX1 probe set will be considered at the laboratory's discretion to identify a potential t(3;21)(q26.2;q22) rearrangement. Laboratory discretion may be influenced by available karyotype results.

In the absence of *RPN1::MECOM* fusion, when an extra RPN1 signal is identified, reflex testing using the PRDM16/RPN1 probe set will be considered at the laboratory's discretion to identify a potential t(1;3)(p36;q21) rearrangement. Laboratory discretion may be influenced by available karyotype results.

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.

This test is intended for instances when the entire MDS fluorescence in situ hybridization (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 AMLAF / Acute Myeloid Leukemia (AML), FISH, Adult, Varies or AMLPF / Acute Myeloid Leukemia (AML), FISH, Pediatric, Varies, it will be canceled and reordered as MDSMF to avoid duplicate FISH probe testing.

At follow-up, targeted MDS FISH probes can be evaluated based on the abnormalities identified in the diagnostic study. Order MDSMF / Myelodysplastic Syndrome (MDS), Specified FISH, Varies and request specific probes or abnormalities.

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

Necessary Information
1. 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.
2. A pathology and/or flow cytometry report may be requested, if not received, by the laboratory to optimize testing and aid in interpretation of results.

Specimen Required

Submit only 1 of the following specimens:

Preferred:
Specimen Type: Bone marrow
Container/Tube:
Preferred: Yellow top (ACD)
Acceptable: Green top (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: Blood
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 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

Blood: 2 mL
Bone Marrow: 1 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 of age. 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.

Bone marrow is the preferred specimen type for this fluorescence in situ hybridization 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).

Supportive Data

Each probe was independently tested and verified on unstimulated peripheral blood and bone marrow specimens. Normal cutoffs were calculated based on the results of 25 normal specimens. Each probe set was evaluated to confirm the probe set detected the abnormality it was designed to detect.

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 May;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 Jul;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, trisomy of chromosome 8, and deletion or rearrangement of chromosomes 17 and 20 are detected using enumeration strategy probes. Dual-color, dual-fusion fluorescence in situ hybridization (D-FISH) strategy probe sets are used to detect inv(3), t(3;21), and t(1;3). For the enumeration probe sets, 100 interphase nuclei are scored; 200 interphase nuclei are scored when D-FISH probes are used. 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 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