# Test Definition: MDSMF

Myelodysplastic Syndrome (MDS), Specified
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 client specified probes

Evaluating specimens in which standard cytogenetic analysis is unsuccessful

### 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 one probe set (2 individual fluorescence in situ hybridization probes). Additional charges will be incurred for all additional probe sets performed.

If the patient is being treated for known abnormalities, indicate the abnormality and which probes should be used.

**When specified**, any of the following probes will be performed:
inv(3) or t(3;3), RPN1/MECOM
t(1;3)(p36;q21), PRDM16/RPN1
t(3;21)(q26.2;q22), MECOM/RUNX1
-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.

### Method Name

Fluorescence In Situ Hybridization (FISH)

### NY State Available

Yes

## Specimen

### Specimen Type
Varies

### Ordering Guidance
This test uses targeted myelodysplastic syndrome (MDS) fluorescence in situ hybridization (FISH) probes to evaluate specific abnormalities or abnormalities identified in the diagnostic sample. The FISH probes to be analyzed must be specified on the request, otherwise test processing may be delayed in order to determine the intended analysis. If specific probes are not included with this test order, the test may be canceled and automatically reordered by the laboratory as MDSDF / Myelodysplastic Syndrome (MDS), Diagnostic FISH, Varies.

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

At diagnosis, conventional cytogenetic studies should be performed. For more information see CHRBM / Chromosome Analysis, Hematologic Disorders, Bone Marrow.

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

### Necessary Information
1. **A list of probes requested for analysis is required.** Probes available for this test are listed in the Testing Algorithm section.
2. 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.
3. A pathology and/or flow cytometry report may be requested by the laboratory to optimize testing and aid in interpretation of results.

### Specimen Required
**Preferred**
**Specimen Type:** Bone marrow
**Container/Tube:**
**Preferred:** Yellow top (ACD)
**Acceptable:** Green top (heparin) or lavender top (EDTA)
**Specimen Volume:** 2-3 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.

**Acceptable**
**Specimen Type:** Blood
**Container/Tube:**

**Preferred:** Yellow top (ACD)
**Acceptable:** Green top (heparin) or lavender top (EDTA)
**Specimen Volume:** 6 mL
**Collection Instructions:** Invert several times to mix blood.

### 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.

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.

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

### 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 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 July;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 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 |

| 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 |