Myelodysplastic Syndrome: Guideline to Diagnosis and Follow-up

Clinical suspicion for a myelodysplastic syndrome (MDS)

Bone marrow testing begins with:
- 70016 / Hematopathology Consultation, Wet Tissue
- CHRBM / Chromosome Analysis Hematologic Disorders, Bone Marrow

Bone marrow morphology: MDS?

YES

Diagnosis: MDS

Chromosomes:
- ≥20 metaphases and
- Resolved karyotype

MDS FISH studies not indicated

Apply results for a MDS prognostic assessment

EQUIVOCAL

Genetic anomalies detected (absence of definitive morphologic features of MDS)?

YES

The genetic anomalies are of uncertain significance and cannot be used as an unequivocal, definitive finding for MDS

NO

No definite diagnosis of MDS can be made

NO

Chromosomes:
- <20 metaphases or
- Unresolved karyotype or
- One abnormal metaphase

MDS FISH studies not indicated

Follow-up evaluation of previously diagnosed MDS

Bone marrow testing begins with:
- 70016 / Hematopathology Consultation, Wet Tissue
- CHRBM / Chromosome Analysis, Hematologic Disorders, Bone Marrow

If still chronic phase and previous studies showed a specific, FISH-detectable genetic anomaly*, then can also perform: MDSF / Myelodysplastic Syndrome (MDS), FISH (with specific FISH probe)

If the bone marrow shows progression to acute leukemia, then proceed with acute leukemia work-up and evaluation

Chromosomes:
- <20 metaphases or
- Unresolved karyotype or
- One abnormal metaphase

Chromosomes:
- ≥20 metaphases and
- Resolved karyotype

MDS FISH studies not indicated

Perform: MDSF / Myelodysplastic Syndrome (MDS), FISH**

Apply results for a MDS prognostic assessment

YES

If reflex testing for myelodysplasia-associated mutations is desired, consider ordering NGSHM / OncoHeme Next Generation Sequencing (NGS), Hematologic Neoplasms.

NO

EQUIVOCAL

* A MDS FISH study with a specific probe but without chromosome analysis may be sufficient in a follow-up bone marrow study for a previously diagnosed MDS with specified genetic anomaly.

** MDS FISH does not increase the detection of MDS if chromosome analysis is successful and ≥20 metaphases are analyzed. Thus, MDS FISH studies should be ordered at the discretion of the cytogeneticist if <20 metaphases are identified, if there is an unresolved karyotype, or if only 1 abnormal metaphase is identified.