Myelodysplastic Syndrome: Guideline to Diagnosis and Follow-up

Clinical suspicion for a myelodysplastic syndrome (MDS)

Bone marrow testing begins with:
- HPWET / Hematopathology Consultation, MML Embed
- HPCUT / Hematopathology Consultation, Client Embed
- 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

NO

EQUIVOCAL

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

MDS FISH studies not indicated

NO

Chromosomes:
- <20 metaphases and
- Resolved karyotype

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

YES

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

- The genetic anomalies are of uncertain significance and cannot be used as an unequivocal, definitive finding for MDS
- No definite diagnosis of MDS can be made

Follow-up evaluation of previously diagnosed MDS

Bone marrow testing begins with:
- HPWET / Hematopathology Consultation, MML Embed
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- 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

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

* 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.