MDS Panel by FISH

Why use MDS Panel by FISH for your patient?

Fluorescence in situ hybridization (FISH) testing is utilized to detect genetic changes associated with the diagnosis and prognosis of patients with myelodysplastic syndromes (MDS). MDS FISH testing is beneficial to perform with classic cytogenetic testing (also performed at Sterling Pathology) for initial diagnosis, but FISH may be performed solely for continual monitoring of disease for the following reasons:

- Important diagnostic and prognostic indicators[1]
- Improved detection rate of typical, non-random MDS abnormalities[2]
- Detects deletions of chromosomes 5q, 7q, and 20q
- Detects monosomy 5 and 7[3], trisomy 8[4], and MLL/11q23 rearrangements[5]
- 48 hour turnaround time

In addition to FISH for MDS:

- Cytogenetic analysis is an important complementary tool in testing for MDS abnormalities, as some abnormalities are better discovered by cytogenetics[1]

Specimen Requirements:

- Specimen must be bone marrow aspirate or biopsy

1 Steensma, D. P., and A. F. List. "Genetic Testing in the Myelodysplastic Syndromes: Molecular Insights Into Hematologic Diversity." *Mayo Clinic Proceedings* 80.5 (2005): 681-98.

2 W, Gordon D. "Cytogenetic and FISH Studies in Myelodysplasia, Acute Myeloid Leukemia, Chronic Lymphocytic Leukemia and Lymphoma." *International Journal of Hematology* 76.2 (2002): 65-74

3 Brizard F, Brizard A, Guilhot F, Tanzer J, Berger R. Detection of monosomy 7 and trisomies 8 and 11 in myelodysplastic disorders by interphase fluorescent in situ hybridization: comparison with acute non-lymphocytic leukemias. *Leukemia*. 1994;8:1005-1011.

4 Beyer V, Castagne C, Muhlematter D, et al. Systematic screening at diagnosis of -5/del(5)(q31), -7, or chromosome 8 aneuploidy by interphase fluorescence in situ hybridization in 110 acute myelocytic leukemia and highrisk myelodysplastic syndrome patients: concordances and discrepancies with conventional cytogenetics. *Cancer Genet Cytogenet*. 2004;152:29-41.

5 Andreasson P, Johansson B, Billstrom R, Garwicz S, Mitelman F, Hoglund M. Fluorescence in situ hybridization analyses of hematologic malignancies reveal frequent cytogenetically unrecognized 12p rearrangements. *Leukemia*. 1998;12:390-400.

Company Overview

Sterling Pathology provides the latest testing technologies specializing in the monitoring and diagnosis of hematopoietic diseases. Sterling Pathology is dedicated to providing the best diagnostic hematopathology services to meet the needs of our hematology and oncology physicians and their patients. We offer a continuum of diagnostic, prognostic, and predictive testing services in anatomic morphology, molecular genetics, cytogenetics, flow cytometry, FISH, and immunohistochemistry.

Expertise

- Board-Certified pathologists with hematopathology subspecialty expertise
- Board-Certified geneticists with cytogenetic subspecialty expertise
- Access to hematopathologist and geneticist for peer-to-peer telephone consultations
- Academic clinical case review

Service

- Unmatched industry-leading turn-around time
- Personalized service from your local Account Executives
- Dedicated customer service care team

Quality

- CAP-accredited, CLIA and state licensed testing facility
- Expanded comprehensive test menu through strategic alliances
- Dedicated logistic staff to manage specimen transport

Report Delivery

- Standardized reporting with full-color photomicrographs
- Reports available via mail, facsimile, remote print, or EMR interface
- WebPortal with 24/7 access to patient reports

Third Party Billing

- Sterling Pathology will bill Medicare, Medicaid and all private insurance providers
- Sterling Pathology will bill all secondary and supplementary insurance providers

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Genetic regions analyzed: Deletions of chromosomes 5q, 7q and 20q; monosomy 5 and 7, trisomy 8 and MLL gene deletions/rearrangements.

5	5p15	hTERT	A	Spot Count	2R2G2A	1R2G2A	del 5q31
	5q31	EGR1	R			2R1G2A	del 5q33
	5q33	RPS14	G			1R1G2A	del 5q31-q33
						1R1G1A	Monsomy 5
7	7p11.1-q11.1	CEN 7	G	Spot Count	2R2G	1R2G	del 7q31
	7q31	D7S486	R			1R1G	Monosomy 7
8	8p11.1-q11.1	CEN 8	A	Spot Count	2R2G2A	3A	Trisomy 8
20	20q12	D20S108	R			1R2G	del 20q12
	20qter	20qter	G			2R1G	del 20qter
						1R1G	del long arm of 20 or monosomy 20
11	11q23	MLL	R&G	Break-apart	2F	1R1G1F	MLL gene rearrangement
						1R1F	Partial MLL gene deletion
						1F	MLL gene deletion

MDS is a heterogeneous group of disorders where patients normally present with cytopenias of unknown origin. The precise etiology of MDS is unknown and often involves multiple factors. The diagnosis and treatment is also difficult at best. FISH and cytogenetic testing are the preferred methods for assisting in the diagnosis of MDS.

Treatment usually involves transfusions, chemotherapy and, in some cases, stem cell transplantation.

Indication of Risk

There are three risk categories identified in Myelodysplastic Syndromes[6] as follows:

Good Risk	Normal karyotype, isolated del(5q), isolated del(20q)
Poor Risk	Complex abnormalities, for example ≥3 abnormalities and abnormalities of chromosome 7
Intermediate Risk	All other abnormalities

Each of the chromosomes which harbor important indicators of risk (5, 7and 20) are included in the MDS FISH panel analysis, as noted above.

6 Swerdlow, Steven H. *WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues*. Lyon, France: International Agency for Research on Cancer, 2008. Page 93. Print.