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
- Improved detection rate of typical, non-random MDS abnormalities
- Detects deletions of chromosomes 5q, 7q, and 20q
- Detects monosomy 5 and 7, trisomy 8, and MLL/11q23 rearrangements
- 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.

Specimen Requirements:

Specimen must be bone marrow aspirate or biopsy

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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
MDS Panel by FISH

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

<table>
<thead>
<tr>
<th>Chrm</th>
<th>Region</th>
<th>Genes</th>
<th>Spot Count</th>
<th>Remarks</th>
</tr>
</thead>
<tbody>
<tr>
<td>5</td>
<td>5p15</td>
<td>hTERT</td>
<td>1R2G2A</td>
<td>del 5q31</td>
</tr>
<tr>
<td></td>
<td>5q31</td>
<td>EGR1</td>
<td>2R1G2A</td>
<td>del 5q33</td>
</tr>
<tr>
<td></td>
<td>5q33</td>
<td>RPS14</td>
<td>1R1G2A</td>
<td>del 5q31-q33</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>1R1G1A</td>
<td>Monosomy 5</td>
</tr>
<tr>
<td>7</td>
<td>7p11.1-q11.1</td>
<td>CEN 7</td>
<td>2R2G</td>
<td>del 7q31</td>
</tr>
<tr>
<td></td>
<td>7q31</td>
<td>D7S486</td>
<td>1R2G</td>
<td>Monosomy 7</td>
</tr>
<tr>
<td>8</td>
<td>8p11.1-q11.1</td>
<td>CEN 8</td>
<td>3A</td>
<td>Trisomy 8</td>
</tr>
<tr>
<td></td>
<td>20q12</td>
<td>D20S108</td>
<td>1R2G</td>
<td>del 20q12</td>
</tr>
<tr>
<td></td>
<td>20qter</td>
<td></td>
<td>2R1G</td>
<td>del 20qter</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>1R1G</td>
<td>del long arm of 20 or monosomy 20</td>
</tr>
<tr>
<td>11</td>
<td>11q23</td>
<td>MLL</td>
<td>1R1G1F</td>
<td>MLL gene rearrangement</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>1R1F</td>
<td>Partial MLL gene deletion</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>1F</td>
<td>MLL gene deletion</td>
</tr>
</tbody>
</table>

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:

<table>
<thead>
<tr>
<th>Risk Category</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Good Risk</td>
<td>Normal karyotype, isolated del(5q), isolated del(20q)</td>
</tr>
<tr>
<td>Poor Risk</td>
<td>Complex abnormalities, for example ≥3 abnormalities and abnormalities of chromosome 7</td>
</tr>
<tr>
<td>Intermediate Risk</td>
<td>All other abnormalities</td>
</tr>
</tbody>
</table>

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