

TEST UPDATE

MICROSATELLITE INSTABILITY BY PCR

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This change will take effect **September 2, 2014**

OVERVIEW AND CLINICAL UTILITY:

PathGroup is pleased to announce the availability of Microsatellite Instability (MSI) analysis by PCR (MSI). Microsatellite instability is a result of a deficiency in a specific type of DNA repair capability called Mismatch Repair (MMR). Cells deficient in MMR have a reduced capability of repairing certain types of damage that naturally occur in their DNA, specifically mismatched bases. This deficiency is apparent in the replication of microsatellite sequences. Such microsatellite sequences are typically replicated and maintain a constant number of repeats. In MMR-deficient cells, the number of repeats can increase or decrease. Tumors that display MMR show a change in the size of microsatellite sequences from those seen in non-neoplastic tissue and are said to display MSI.

MSI is detected in about 15 to 20% of all colorectal cancers; ~3% of these are associated with Lynch syndrome and the other 12 to 17% are caused by sporadic, acquired hypermethylation of the promoter of the *MLH1* gene, which occurs in tumors with the CpG island methylator phenotype. Colorectal tumors with MSI have distinctive features, including a tendency to arise in the proximal colon, lymphocytic infiltrate, and a poorly differentiated, mucinous or signet ring appearance. They have a slightly better prognosis than colorectal tumors without MSI and do not have the same response to chemotherapeutics. Discovery of MSI in colorectal tumors has increased awareness of the diversity of colorectal cancers and implications for specialized management of patients.

TEST METHODOLOGY:

Genomic DNA is isolated from the tumor and normal areas of the specimen. Microsatellite markers NR-21, BAT-26, BAT-25, NR-24 and MONO-27 are amplified by PCR from both the tumor and normal areas independently, followed by size fractionation via capillary electrophoresis. Normal results and tumor results are compared to detect changes in microsatellite sizes.

ORDERING:

| TEST | TEST NAME | CPT | LOINC |
|------|-----------------------------------|--------------|--------|
| MSI | Microsatellite Instability by PCR | 81301, 88381 | T00818 |

NORMAL AND REFERENCE RANGE:

| TEST | | RESULT |
|------|--|----------|
| MSI | No Microsatellite Instability Detected | Normal |
| | Low Microsatellite Instability Detected | Abnormal |
| | High Microsatellite Instability Detected | Abnormal |

SPECIMEN COLLECTION AND STORAGE:

| IF ORDERING: | |
|--------------|---|
| → | FFPE: Requires a minimum of 25 mm ² of tumor tissue containing a minimum of 20% malignant cells. Also requires a minimum of 25 mm ² of normal tissue (can be from the same specimen). |
| → | Tissue blocks or 7 to 10 unstained, 7 micron sections on uncharged slides are acceptable |
| → | Store and transport at room temperature. |

TEST PERFORMED: Wednesdays

TURNAROUND TIME: 8-10 days

For further questions, please contact Client Services at 615-562-9300 or 1-888-474-5227