

# Microsatellite Instability Testing (MSI) in Colon Cancer

## Clinical Indication and Relevance

Please refer to the following references.

1. GeneTests Web “Hereditary Non-Polyposis Colon Cancer”  
<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=hnpcc>
2. Genetic testing for colon cancer: Joint statement of the American College of Medical Genetics and American Society of Human Genetics  
[http://www.acmg.net/StaticContent/StaticPages/Colon\\_Cancer.pdf](http://www.acmg.net/StaticContent/StaticPages/Colon_Cancer.pdf)

## Methodology

DNA from paired normal and tumor tissues is analyzed by multiplex PCR and capillary electrophoresis for five mononucleotide repeat markers (NR-21, BAT-26, BAT-25, NR-24, and MONO-27). Allelic profiles from paired tissues are compared to determine the MSI status.

## Sensitivity

It is recommended that the tumor sample be obtained from a resection in which more than 70% malignant cells are present.

## Turn-around Time

Five to seven working days

## Sample Requirements

Testing requires BOTH normal colorectal tissue and colorectal tumor tissue for each patient. Please submit samples together with these forms: 1) Request for Molecular Diagnostic Studies Form; 2) Supplemental Form for Submitting Slides and/or Tissues.

## Collect

### Recommended specimen type:

- Paraffin-embedded tissue blocks: one normal colorectal tissue block AND one colorectal tumor tissue block. The pathologist should choose a tumor block with >70% carcinoma, and a block containing normal tissue from the same patient. Otherwise, one block that contains both normal and tumor tissue is acceptable.

### Alternative specimen types:

- Unstained colorectal tissue slides: if tissue blocks are unavailable, five to seven unstained slides representing each of normal and tumor tissue are acceptable. Please submit unstained slides with one H&E stained slide.
- Peripheral blood (PB): only if normal colorectal tissue is not available, please submit one tube of peripheral blood (EDTA or ACD) TOGETHER with one tumor tissue block. Note: Peripheral blood is only an acceptable replacement for normal tissue. In addition, tumor block or slides are also required. Testing cannot be performed solely on peripheral blood.

## Transport

- Formalin-fixed paraffin-embedded (FFPE) tissue blocks or slides can be delivered at room temperature.
- If peripheral blood (PB) collected, deliver blood sample immediately at 2-8°C (wet ice or cold packs). Do not freeze.

## Stability

Blood samples: ambient - 8 hours; refrigerated - 48 hours.

## Unacceptable Samples

- Serum or plasma; frozen PB; clotted blood; severely hemolyzed blood samples
- Tissue fixed in Zenker's, B5, or Bouin's fixatives
- Tissue blocks with no evidence of tumor
- Biopsy specimens (the assay has not been validated for biopsy specimens)
- Adenoma tissue should not be tested

## CPT Code(s)

81301: Microsatellite instability analysis of markers for mismatch repair deficiency includes comparison of neoplastic and normal tissue

G0452-26: Molecular pathology procedure; physician interpretation and report