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**Microsatellite Instability**

**↑CPT**

83890 (x2); 84311 (x2); 83907 (x2); 83900 (x4); 83901 (x4); 83909 (x4); 83912

**↑Synonyms**

Mismatch Repair Defect; MSI; RER

**↑Test Includes**

Testing of 5 loci; BAT25, BAT26, D2S123, D5S346, D17S250.

**↑Laboratory**

Molecular Pathology

**↑Availability**

Monday-Friday, 0700-1700

**↑Turnaround Time**

3-14 days

**↑Specimen**

1 normal tissue and 1 tumor tissue. Normal tissue may be substituted by a blood sample.

**↑Volume**

Each tissue sample must be  $\geq 25$  mg. If blood is submitted, 4 mL is required.

## ↑ Container

Tissue may be paraffin-embedded or fresh frozen. Blood must be collected in a 4 mL EDTA tube.

## ↑ Storage Instructions

Fresh frozen tissue must remain frozen at all times. Paraffin-embedded tissue may remain at room temperature indefinitely. Blood may be stored at room temperature up to 48 hours.

## ↑ Causes for Rejection

Fresh frozen tissue allowed to thaw; specimen inadequately identified

## ↑ Reference Range

No MSI observed

## ↑ Use

Microsatellite instability is caused by failure of the DNA mismatch repair (MMR) system to correct errors that occur during the replication of DNA. The resulting errors in critical cell growth genes, such as oncogenes and tumor suppressor genes, can promote the malignant transformation process and thus the MMR defect can be a hallmark of certain tumors. This has been most extensively worked out in the case of hereditary nonpolyposis colorectal cancer (HNPCC), but can be found in a variety of other tumor types as well. A convenient way to detect the MMR defect is by observing errors in replication of microsatellite short tandem repeats in tumor DNA, a phenomenon called microsatellite instability (MSI). Presence of MSI in a colonic adenocarcinoma may suggest a familial cancer syndrome (though some sporadic colon cancers are also MSI-positive), and may also assist in deciding management options.

## ↑ Limitations

The finding of MSI by itself does not absolutely define the tumor as genetic in origin. This can be done by testing of the MMR genes themselves for mutations, or by examining their protein products.

## ↑ Methodology

Amplification by Polymerase Chain Reaction (PCR) of 5 microsatellite short tandem repeats and detection of these alleles by electrophoresis and sizing on an automated fluorescence detector

## **↑Additional Information**

While still a subject of debate, most authorities do not consider MSI analysis to be a genetic test requiring informed consent; however, those ordering the test should be aware of its potential genetic implications for the patient and other family members.

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