



Test Catalog

Diagnostic. Prognostic. Predictive. Predisposition.



MLH1 Promoter Methylation Analysis

Alternative Name

MLH1 Methylation

Methodology

Molecular

Test Description

This assay is performed on tumor tissue to detect hypermethylation of the MLH1 gene promoter. Bisulfite modification of tumor DNA and real-time PCR are used to quantify CpG methylation within the promoter. Percentage of methylated DNA (compared to total DNA) is reported for positive results. Analysis should be considered in combination with IHC, BRAF, and/or MSI.

Clinical Significance

MLH1 promoter methylation analysis is useful to distinguish sporadic from inherited colorectal and endometrial cancers in tumors that are MLH1-deficient by IHC staining and/or have high levels of microsatellite instability (MSI-H). The majority of MSI in sporadic cases of these tumors is caused by MLH1 promoter hypermethylation, while hypermethylation is rare in inherited cases. MLH1 promoter methylation analysis results should be considered with other clinical risk factors in determination of likelihood of HNPCC/Lynch Syndrome.

Specimen Requirements

- **FFPE solid tumor tissue:** Paraffin block is preferred. Alternatively, send 1 H&E slide plus 5-10 unstained slides cut at 5 or more microns. Please use positively-charged slides and 10% NBF fixative. Do not use zinc fixatives.

Storage & Transportation

Use cold pack for transport, making sure cold pack is not in direct contact with specimen.

CPT Code(s)*

81288

New York Approved

Yes

Level of Service

Global

Turnaround Time

10 days

*The CPT codes provided with our test descriptions are based on AMA guidelines and are for informational purposes only. Correct CPT coding is the sole responsibility of the billing party.

Please direct any questions regarding coding to the payor being billed.

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Committed to research as the means to improve patient care, we provide Pharma Services for pharmaceutical companies, in vitro diagnostic manufacturers, and academic scientist-clinicians. We promote joint publications with our client physicians. NeoGenomics welcomes your inquiries for collaborations. Please contact us for more information.

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