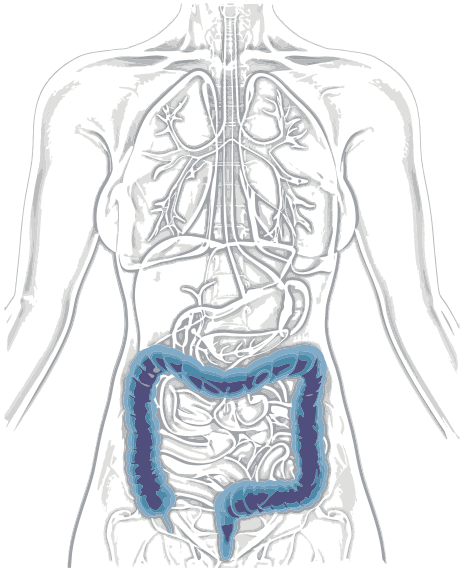

Colon Cancer and Lynch Syndrome

- Universal screening of all colorectal cancer (CRC) specimens (NCCN 2015; ASCO 2014) and endometrial cancer specimens (ASCO 2014; ESMO 2013;NCCN 2015) is recommended.
- Lynch syndrome (LS) is an autosomal dominant inherited cancer syndrome that predisposes to colorectal, endometrial, gastric, ovarian, upper urinary tract, and other cancers.
- The presence of mismatch repair (MMR) deficiency helps identify patients at risk for LS; however, MMR also occurs in ~15% of sporadic colorectal cancers.
- Differentiating colorectal tumors with MMR deficiency due to a sporadic somatic event from colorectal tumors with MMR deficiency due to a LS germline mutation is important.

www.aruplab.com/testing/colon-cancer

ARUP LABORATORIES

www.aruplab.com

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Salt Lake City, UT 84108-1221
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keyword: colon cancer

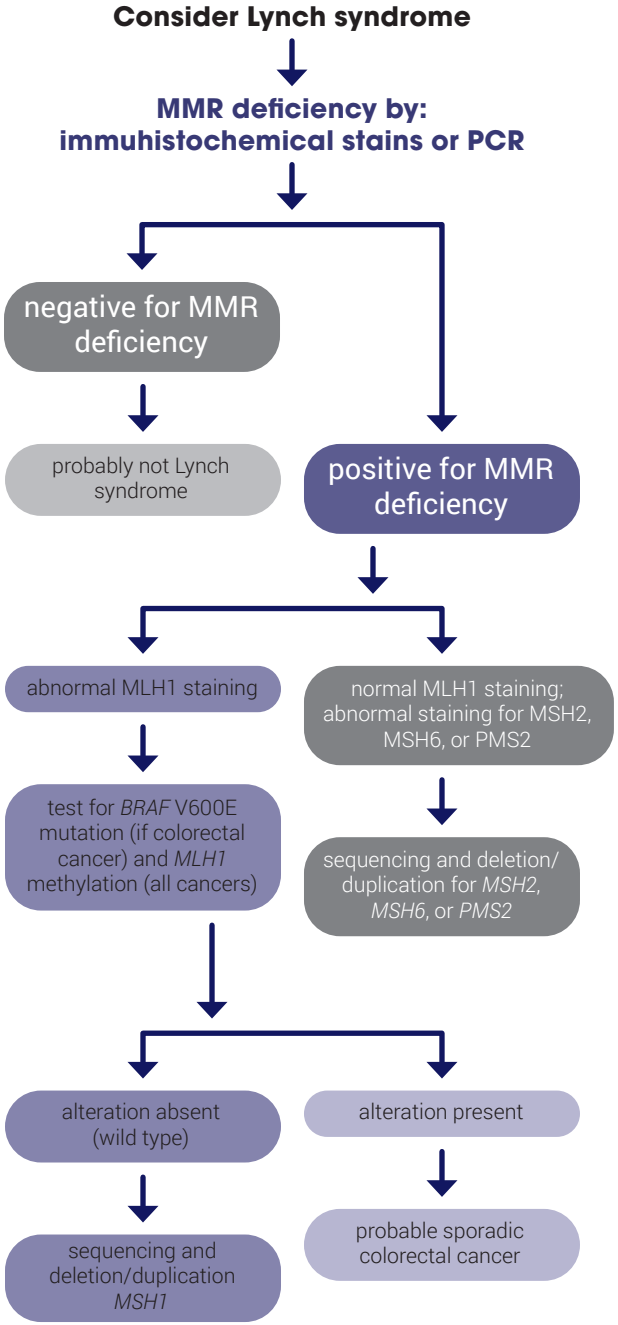
A nonprofit enterprise of the University of Utah and its Department of Pathology

BD-CS-033, Rev 2, May 2018

Colon Cancer and Lynch Syndrome

testing at ARUP Laboratories

ARUP LABORATORIES



ARUP Test Menu

Mismatch Repair by Immunohistochemistry (0049302)
First-line screening test for Lynch syndrome; directs additional molecular diagnostic testing for Lynch syndrome. Includes MLH1, MSH2, MSH6, and PMS2.

Mismatch Repair by Immunohistochemistry with Reflex to *BRAF* Codon 600 Mutation and *MLH1* Promoter Methylation (2002327)
Preferred screening test for Lynch syndrome in individuals with colorectal cancer.

Mismatch Repair by Immunohistochemistry with Reflex to *MLH1* Promoter Methylation (2005270)
Preferred reflex screening test for Lynch syndrome in non-colorectal cancer tumors (e.g., endometrial carcinoma).

***BRAF* Codon 600 Mutation Detection with Reflex to *MLH1* Promoter Methylation (0051750)**
Recommended reflex test for differentiating between Lynch syndrome and sporadic colorectal cancer in tumors showing loss of *MLH1*; if no *BRAF* mutation is detected, *MLH1* promoter methylation is evaluated.

***BRAF* Codon 600 Mutation Detection by Pyrosequencing (2002498)**
Used to evaluate suspected Lynch syndrome and predict effectiveness of therapies targeting the EGFR pathway.

***MLH1* Promoter Methylation, Paraffin (2002499)**
Distinguishes between Lynch syndrome and sporadic tumors with loss of *MLH1*.

Microsatellite Instability (MSI), HNPCC/Lynch Syndrome by PCR—Tumor and Normal Tissue (0051740)
First-line screening test for Lynch syndrome; directs additional molecular diagnostic testing for Lynch syndrome.

HNPCC/Lynch Syndrome Deletion/Duplication (2001728)
Second-tier test that requires approval from ARUP genetic counselor; call (800) 242-2787, ext. 2141, before ordering.

HNPCC/Lynch Syndrome (*MLH1*) Sequencing and Deletion/ Duplication (0051650)
Detects germline *MLH1* mutations; used in MMR-deficient carcinoma with suggestive IHC (loss of MLH1 and PMS2 proteins), absence of *BRAF* codon 600 mutation, and normal *MLH1* methylation studies.

HNPCC/Lynch Syndrome (*MSH2*) Sequencing and Deletion/ Duplication (0051654)
Detects germline *MSH2* mutations; used in MMR-deficient carcinoma with suggestive IHC (loss of MSH2 and MSH6 proteins); detects large *MSH2* deletions and three prime *EPCAM* deletions.

HNPCC/Lynch Syndrome (*MSH6*) Sequencing and Deletion/ Duplication (0051656)
Detects germline *MSH6* mutations; used in MMR-deficient carcinoma with suggestive IHC (isolated loss of MSH6 protein).

HNPCC/Lynch Syndrome (*PMS2*) Sequencing and Deletion/ Duplication (0051737)
Detects germline *PMS2* mutations; used in MMR-deficient carcinoma with suggestive IHC (isolated loss of PMS2 protein).

Gastrointestinal Hereditary Cancer, Sequencing and Deletion/ Duplication, 16 genes (2013449)
Confirms a diagnosis of hereditary gastrointestinal (GI) cancer in individuals with a personal or family history of GI cancer and/ or polyposis.

Cancer Panel, Hereditary, Sequencing and Deletion/Duplication, 47 Genes (2012032)
Confirms diagnosis of a hereditary cancer syndrome in an individual with a personal or family history that could be consistent with features of more than one cancer syndrome.

Familial Mutation, Targeted Sequencing (2001961)
Detects a mutation previously identified in a family member; consultation with a genetics counselor is advised.

***BRAF* Codon 600 Mutation Detection by Pyrosequencing (2002498)**
Evaluates suspected Lynch syndrome; used to predict effectiveness of therapies targeting the EGFR pathway.

***KRAS* Mutation Detection (0040248)**
Predicts response to anti-EGFR and MAPK pathway therapies in a variety of malignancies (e.g., colorectal and lung cancer).

***NRAS* Mutation Detection by Pyrosequencing (2003123)**
Predicts response to anti-EGFR and MAPK pathway therapies in a variety of malignancies (e.g., melanoma and colorectal cancer).

Colon Cancer Gene Panel, Somatic (2011616)
Indicated for individuals with metastatic colorectal cancer to guide treatment with anti-EGFR monoclonal antibodies (i.e., cetuximab and panitumumab); detects mutations in *BRAF*, *KRAS*, *NRAS*, extended *KRAS*, and *PIK3CA*.

Solid Tumor Mutation Panel by Next-Generation Sequencing (2007991)
Aids in therapeutic decisions for solid tumor cancers; does not detect translocations.

Epi proColon (2013906)
The Epi proColon test is indicated to screen adults of either sex, 50 years or older, defined as average risk for CRC, who have been offered and have a history of not completing CRC screening. Tests that are available and recommended in the USPSTF 2008 CRC screening guidelines should be offered and declined prior to offering the Epi proColon test.

Occult Blood, Fecal by Immunoassay (2007190)

Sample type: FFPE tissue | Sample type: whole blood | Sample type: plasma | Sample type: stool

For more information and educational opportunities, visit:

www.aruplab.com/testing/colon-cancer