Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,HCCD	SA00056045	38	M	SA00056045
Ordering Phys CLIENT,CLIENT				**DOB** 03/16/1975
Client Order # SA00056045	**Account Information**			**Report Notes**
Collected 03/21/2013 13:48	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 04/16/2013 15:07				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Hereditary Colon Cancer Del/Dup			REPORTED 03/26/2013 14:03		
Reason for Referral					MCR
Test for the presence of large deletions and duplications in the genes included in the Hereditary Colon Cancer Deletion/Duplication Panel.					
Result					MCR
No large genomic deletions or duplications were identified.					
Interpretation					MCR
This result does not rule out involvement of the genes evaluated in this panel. We predict that some individuals who have a single-gene associated risk for colon cancer may have a mutation in one of these genes that is not identified by the methods described.					
This assay does not rule out the presence of other mutations within these genes or within other genes that are associated with colorectal and other cancers.					
A genetic consultation may be of benefit.					
Caution					MCR
CLINICAL CORRELATIONS Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.					
If testing was performed because of a family history colon cancer or colon polyps, it is often useful to first test an affected family member. Identification of a specific gene mutation in this family would lead to more direct testing of at risk individuals.					
TECHNICAL LIMITATIONS Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.					
Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.					
EVALUATION TOOLS Multiple in-silico evaluation tools were used to assist in					

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the interpretation of these results. These tools are updated regularly, therefore changes to these algorithms may result in different predictions for a given alteration. Additionally, the predictability of these tools for the determination of pathogenicity is currently unvalidated.

RECLASSIFICATION OF VARIANTS - POLICY
All detected alterations are evaluated according to ACMG recommendations (Genet Med 2008:10(4):294-300). Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. At this time, it is not standard practice for the laboratory to systematically review LIKELY DELETERIOUS alterations or VARIANTS OF UNCERTAIN SIGNIFICANCE that are detected and reported. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

TEST CLASSIFICATION
Laboratory developed test.

Method MCR

Array comparative genomic hybridization (aCGH) was used to test for the presence of large deletions and/or duplications in the APC, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MLH3, MSH2, MSH6, TP53, PTEN, SCG5, SMAD4, and STK11 genes. Gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in the PMS2 gene.

Mutation nomenclature is based on GenBank accession numbers listed below.

GENE	GENBANK#
APC	NM 000038.5
AXIN2	NM 004655.3
BMPR1A	NM 004329.2
CDH1	NM 004360.3
CHEK2	NM 007194.3
EPCAM	NM 002354
MLH1	NM 000249.3
MLH3	NM 000014.8
MSH2	NM 000251.1
MSH6	NM 000179.2
TP53	NM 000546.4

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Test	Flag	Results	Unit	Reference Value	Perform Site*
PMS2 NM 000535.5					
PTEN NM 000314.4					
SCG5 NM 001144757.1					
SMAD4 NM 005359.5					
STK11 NM 000455.4					
Specimen		Blood			MCR
Reviewed By					MCR
Matthew John Ferber PhD					
Release Date		26 Mar 2013 13:59			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

* Report times for Mayo performed tests are CST/CDT