Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,HCCD	SA00056046	60	F	SA00056046
Ordering Phys CLIENT,CLIENT				**DOB** 10/25/1952
Client Order # SA00056046	**Account Information**			**Report Notes**
Collected 03/21/2013 13:49	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 14:02		
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
The following deletion was detected: Gene: TP53 Exon(s): 1-11 Classification: DELETERIOUS					
Interpretation					MCR
The deletion of exon(s) 1-11 in TP53 is a known deleterious mutation. This result is consistent with a diagnosis of Li-Fraumeni syndrome for this individual. Appropriate screening procedures and/or prophylactic measures should be considered. For information regarding the full tumor spectrum and other associated phenotypes, see GeneReviews for this specific gene (http://www.ncbi.nlm.nih.gov/sites/GeneTests/). Since a mutation has been identified in the TP53 gene, testing of at risk family members is possible. Mutation-specific testing is available at Mayo Medical Laboratories. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test. 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					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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 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					

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Test		Flag	Results	Unit	Reference Value	Perform Site*
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					
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 14:00			MCR

* Performing Site:

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

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