Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,HNPCC	SA00066729	47	F	SA00066729
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00066729	**Account Information**			**Report Notes**
Collected 04/06/2014 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 04/09/2014 08:37				

Test	Flag	Results	Unit	Reference Value	Perform Site*
HNPCC Screen					
RECEIVED: 04/07/2014 12:56 **REPORTED:** 04/08/2014 08:57					
Microsatellite Instability, Tumor					
Specimen		Tissue-Tumor			MCR
Specimen ID		1063066			MCR
Order Date		07 Apr 2014 15:32			MCR
Reason For Referral					MCR
Possible diagnosis of Hereditary Nonpolyposis Colon Cancer (HNPCC)/Lynch syndrome. Evaluate tumor tissue for evidence of defective DNA mismatch repair.					
Method					MCR
Immunohistochemical staining (IHC) is used to determine the presence or absence of protein expression for MLH1, MSH2, MSH6 and PMS2. Lymphocytes and normal epithelium exhibit strong nuclear staining and serve as positive internal controls for staining of these proteins.					
Microscopic examination was performed by a pathologist only to identify areas of normal and tumor for enrichment by macrodissection. A PCR based assay is used to test for tumor microsatellite instability (MSI) with the use of 5 mononucleotide repeat markers (BAT25, BAT26, Mono27, NR24, and NR21). The tumor tissue is classified as MSS/MSI-L (instability detected in 0 or 1 out of 5 markers), or MSI-H (instability in 2 or more of 5 markers tested).					
Results					MCR
Provided diagnosis: adenocarcinoma IHC: Normal expression of MLH1, MSH2, MSH6, and PMS2 MSI: MSS/MSI-L (instability observed informative markers)					
Interpretation					MCR
The combination of normal protein expression and an MSS/MSI-L phenotype suggests the presence of normal DNA mismatch repair function within the tumor. Thus, the likelihood that this individual has an inherited colon cancer syndrome due to defective DNA mismatch repair (HNPCC/Lynch syndrome) is reduced but not eliminated.					
However, these results do not rule out the possibility that this individual's tumor is due to an inherited defect in another gene not involved in DNA mismatch repair. A significant fraction of clinically defined HNPCC cases (30% or more) do not have defective DNA mismatch repair as the underlying genetic basis of their disease.					

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Additionally, we cannot rule out the possibility that this individual or family has HNPCC/Lynch syndrome because this tumor could represent a sporadic occurrence. If there is a strong personal or family history of HNPCC/Lynch syndrome related cancers for this patient or if this individual has multiple tumors, consider microsatellite instability (MSI) and immunohistochemical staining (IHC) on a different tumor to further evaluate the possible role of defective DNA mismatch repair for this individual or family.

Of note, the literature suggests that MSI analysis on neoadjuvant chemoradiated tumor specimens may influence MSI status and lead to an erroneous interpretation of results (Int J Radiat Oncol Biol Phys. 2007 68(5):1584).

Due to the sensitivity of the method being used, microsatellite instability cannot be reliably detected in samples containing less than 30% tumor DNA. Samples are routinely macrodissected to enrich for tumor cells, with those less than 30% rejected from further testing.

These data should be interpreted in the context of the histopathologic findings. A surgical pathology consult may be ordered separately.

A genetic consultation may be of benefit.

CAUTIONS:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. If results obtained do not match other clinical or laboratory findings, please contact the laboratory for possible interpretation. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Laboratory developed test.

Extraction Performed?		Yes.		MCR
Consultant		Heather Lynn Owen		MCR
Report Date		07 Apr 2014 15:36		MCR
MMR Protein, IHC Only, Tumor				
Specimen		Tissue-Tumor		MCR
Specimen ID		1063066		MCR
Order Date		07 Apr 2014 15:32		MCR
Reason For Referral				MCR

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Possible diagnosis of Hereditary Nonpolyposis Colon Cancer (HNPCC)/Lynch syndrome. Evaluate tissue for evidence of defective DNA mismatch repair.					
Method					MCR
Microscopic examination was performed by a pathologist only for immunohistochemical (IHC) interpretation. IHC is used to determine the presence or absence of protein expression for one or more of the following: MLH1, MSH2, MSH6, and PMS2. Lymphocytes and normal epithelium exhibit strong nuclear staining to serve as positive internal controls for staining of these proteins.					
MLH1 IHC		Performed			MCR
MSH2 IHC		Performed			MCR
MSH6 IHC		Performed			MCR
PMS2 IHC		Performed			MCR
Result					MCR
Provided diagnosis: adenocarcinoma IHC: Normal expression of MLH1, MSH2, MSH6, and PMS2					
Interpretation					MCR
The results of the IHC analysis suggest the presence of normal DNA mismatch repair function within the tumor. Thus, the likelihood that this individual has an inherited colon cancer syndrome due to defective DNA mismatch repair (HNPCC/Lynch syndrome) is reduced but not eliminated. These results reduce but do not completely rule out the possibility of defective DNA mismatch repair with in the tumor because approximately 5% of cases with defective mismatch repair do not show absence of protein expression by IHC. These results also do not exclude the possibility that this individual's tumor is due to an inherited defect in another gene not involved in DNA mismatch repair. A significant fraction of clinically defined HNPCC cases (30% or more) do not have defective DNA mismatch repair as the underlying genetic basis of their disease. Additionally, we cannot rule out the possibility that this individual or family has HNPCC/Lynch syndrome because this tumor could represent a sporadic occurrence. If there is a strong personal or family history of HNPCC/Lynch syndrome related cancers for this individual or if this individual has multiple tumors, consider microsatellite instability (MSI) testing on this tumor or a different tumor to further evaluate the possible role of defective DNA mismatch repair					

Performing Site Legend on Last Page of Report

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for this individual or family.

These data should be interpreted in the context of the histopathologic findings. A surgical pathology consult may be ordered separately.

A genetic consultation may be of benefit.

CAUTIONS:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. If results obtained do not match other clinical or laboratory findings, please contact the laboratory for possible interpretation. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Laboratory developed test.

Reviewed By:	Heather Lynn Owen	MCR
Release Date	07 Apr 2014 15:36	MCR

* Performing Site:

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