MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,HNPCC	SA00046796	45	F	SA00046796
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00046796	**Account Information**			**Report Notes**
Collected 05/24/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/15/2012 14:52				

Test	Flag	Results	Unit	Reference Value	Perform Site*
HNPCC Screen			REPORTED 07/13/2012 10:26		
Microsatellite Instability, Tumor					
Specimen		Tissue-Tumor			MCR
Specimen ID		1038229			MCR
Order Date		29 May 2012 08:46			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.

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

Tumor type: colon adenocarcinoma
IHC: Loss of MSH2 and MSH6; Retention of MLH1 and PMS2
MSI: MSI-H (instability observed in 5 of 5 informative markers)

Interpretation MCR

These results indicate loss of normal DNA mismatch repair function within the tumor. Loss of MSH2 and MSH6 expression is frequently associated with the presence of a germline (heritable) mutation in MSH2 and less frequently in MSH6. Thus, this individual and other family members are at increased risk for having an inherited colon cancer syndrome due to defective DNA mismatch repair (HNPCC/Lynch syndrome).

It is important to note that these results do not distinguish between somatic and germline mutations. Germline testing of MSH2 (Mayo test code 83016) and MSH6 (Mayo test code 83723) on an additional blood sample may help to distinguish between these two possibilities and provide the opportunity for predictive testing for at risk family members.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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.					
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.					
For research use only.					
Extraction Performed?		Yes			MCR
Consultant					MCR
Melody Elizabeth Kimball					
Report Date		13 Jul 2012 10:23			MCR
MMR Protein, IHC Only, Tumor					
Specimen		Tissue-Tumor			MCR
Specimen ID		1038229			MCR
Order Date		29 May 2012 08:46			MCR
MLH1 IHC		Performed			MCR
MSH2 IHC		Performed			MCR
MSH6 IHC		Performed			MCR
PMS2 IHC		Performed			MCR
Result					MCR
Analysis has been completed. Refer to the Microsatellite Instability, Tumor for results and interpretation.					
Reviewed By:					MCR
Melody Elizabeth Kimball					
Release Date		13 Jul 2012 10:23			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.