

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,MLHKM	Patient ID SA00050429	Age 60	Gender M	Order # SA00050429
Ordering Phys		·		DOB 06/15/1952
Client Order # SA00050429	Account Information			Report Notes
Collected 11/06/2012	C7028846-DLMP ROC 3050 SUPERIOR DRI	VE		
Printed 11/20/2012 09:07	ROCHESTER,MN 559	901		

Test	Flag	Results	Unit	Reference Value	Perform Site
H1 Large Del/Dup			REPORTED	11/06/2012 13:24	
Result Analysis has been complete	ed. Refer to	the MLH1 Known			MCF
Mutation for results and i	nterpretation				
Specimen		Blood			MCF
Reviewed By:					MCF
Melody Elizabeth Kimball Release Date		06 Nov 2012 13	3:23		MCF
.H1 Known Mutation Reason For Referral			REPORTED	11/19/2012 18:53	MCF
Family history of Lynch Sy a mutation in the MLH1 ger		for the presen	nce of		
Result The following sequence cha	inge was detec	ted:			MCR
DNA change: c.1459C>T					
Amino acid change: p.R487	(Arg487X)				
Classification: DELETERIOU	JS				
Interpretation This alteration is a known					MCF

This result is consistent with a diagnosis of Lynch Syndrome. Appropriate screening procedures and/or prophylactic measures should be considered.

Identification of a mutation for this individual means that predictive testing can be performed for at risk family members.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

CAUTIONS:

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.

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,MLHKM	11/06/2012	Final
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^{*} Report times for Mayo performed tests are CST/CDT

MCR

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Test Flag Results Unit Value Site*

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.

Laboratory developed test.

Method

hod

DNA sequence analysis was used to test for the presence of

the c.1459C>T (p.R487X) mutation in exon 13 of the MLH1 gene. Testing was performed for this specific mutation because it was previously identified in an affected family member of this individual. Mutation nomenclature is based on GenBank accession number, NM_000249.

SpecimenBloodReviewed By:D Brian Dawson PhDRelease Date19 Nov 2012 18:52

* Performing Site:

MCR Mayo Clinic Laboratories - Rochester Main Campus Lab Director: Franklin R. Cockerill, III, M.D.

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