MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FAPKM	SA00046683	45	F	SA00046683
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00046683	**Account Information**			**Report Notes**
Collected 05/20/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/15/2012 12:07				

Test	Flag	Results	Unit	Reference Value	Perform Site*
FAP Known Mutation			REPORTED 07/13/2012 10:05		
Specimen		Blood			MCR
Specimen ID		1038192			MCR
Order Date		21 May 2012 14:58			MCR
Reason For Referral					MCR

Family history of familial adenomatous polyposis (FAP). Test for the presence of a mutation in the APC gene.

Method MCR

DNA sequence analysis was used to test for the presence of the c.453delA mutation in exon 4 of the APC gene. Testing was performed for this specific mutation because it was previously identified in an affected family member of this individual.

Result MCR

The following deletion was detected:
Exon: 4
DNA change: c.453delA
Amino Acid change: p.E152KfsX18 (Glu152LysfsX18)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of FAP for this individual. Appropriate screening procedures and/or prophylactic measures should be considered.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for FAP is available at Mayo Medical Laboratories by ordering FAPKM/83001 FAP Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

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.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Misinterpretation of results may occur if the information provided is inaccurate or incomplete.					
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.					
Reviewed By: Melody Elizabeth Kimball					MCR
Release Date		13 Jul 2012 10:00			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.

TEST SITE