

Laboratory Service Report

1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT, CHEKK	SA00064006	28	M	SA00064006
Ordering Phys CLIENT,CLIENT				DOB 06/15/1985
Client Order #	Account Information			Report Notes
SA00064006				
Collected	C7028846-DLMP Roch			
10/30/2013 00:00		SDSC 2 - Client Support		
Printed	Rochester, MN 55901			
12/24/2013 09:28				

Test Flag Results Unit Value Site*

CHEK2 Gene, Known Mutation

RECEIVED: 10/30/2013 13:28 **REPORTED:** 12/02/2013 13:00

Reason For Referral

MCR

MCR

Family history of breast cancer. Test for the presence of a mutation in the CMFK2 cane

mutation in the CHEK2 gene.

Result MCR

The following deletion was detected: DNA change: c.1100delC (g.29091857)

Amino acid change: p.T367MfsX15 (Thr367MetfsX15)

Classification: DELETERIOUS

Interpretation

This alteration is a known deleterious mutation.

Mutations in the CHEK2 gene have been identified in individuals with a suspected diagnosis of Li-Fraumeni syndrome/Li-Fraumeni-like syndrome. Therefore, this result may be contributing to a risk for cancer in this patient and/or family. In addition, individuals with a truncating mutation in CHEK2 may be at an increased risk for breast cancer (J Clin Oncol 2011 29:3747-3752). 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 is available at Mayo Medical Laboratories by ordering CHEKK/61479 CHEK2 Gene, 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. 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,CHEKK	10/30/2013 00:00	Final
Page 1 of 2		>> Continued on Next Page >>

^{*} Report times for Mayo performed tests are CST/CDT



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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.

MCR DNA sequence analysis was used to test for the presence of

the c.1100delC alteration in the CHEK2 gene (GenBank accession number, NM $_$ 007194; build GRCh37 (hg19)).

Specimen

Reviewed By Matthew John Ferber PhD

02 Dec 2013 12:58 Release Date MCR

* Performing Site:

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

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT, CHEKK	10/30/2013 00:00	Final
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