Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,PTENS	SA00058904	67	M	SA00058904
Ordering Phys CLIENT,CLIENT				**DOB** 06/15/1945
Client Order # SA00058904	**Account Information**			**Report Notes**
Collected 06/11/2013 08:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/27/2013 12:47				

Test	Flag	Results	Unit	Reference Value	Perform Site*

PTEN Gene, Full Gene Analysis
RECEIVED: 06/11/2013 09:33 **REPORTED:** 06/12/2013 10:02

Reason For Referral MCR

Possible diagnosis of PTEN hamartoma tumor syndrome (PHTS).
Test for the presence of a mutation in the PTEN gene.

Result MCR

The following sequence change was detected:
Exon: 8
DNA change: c.1003C>T
Amino acid change: p.R335X (Arg335X)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of PTEN Hamartoma Tumor syndrome (PHTS). The clinical spectrum of PHTS is broad and includes Cowden syndrome, Bannayan-Riley-Ruvalcaba syndrome, Proteus syndrome and Proteus-like syndrome. Therefore, clinical correlation is recommended to determine the appropriate screening procedures and/or prophylactic measures for this individual.

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 PTENK/61488, PTEN 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.

Rare polymorphisms exist that could lead to false negative

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,PTENS	SA00058904	67	M	SA00058904
Ordering Phys CLIENT,CLIENT				**DOB** 06/15/1945
Client Order # SA00058904	**Account Information**			**Report Notes**
Collected 06/11/2013 08:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/27/2013 12:47				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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					MCR
Bi-directional sequence analysis was performed to test for the presence of a mutation in the promoter (c.-1250_c.1), all coding regions and intron/exon boundaries of the PTEN gene. Array comparative genomic hybridization (aCGH) is used to test for the presence of large deletions and duplications. Mutation nomenclature is based on GenBank accession number; NM_000314.4					
Array Billed?		Yes.			MCR
See COLDB, Hereditary Colon Cancer CGH Array, for billing information.					
Specimen		Blood			MCR
Reviewed By					MCR
Matthew John Ferber PhD					
Release Date		12 Jun 2013 09:59			MCR

* Performing Site:

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