MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,VHLSP A	SA00060516	47	F	SA00060516
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00060516	**Account Information**			**Report Notes**
Collected 07/31/2013 00:00	C7002670-MML Unknown Client Account 3050 Superior Drive NW Rochester, MN 55901			
Printed 08/12/2013 14:51				

Test	Flag	Results	Unit	Reference Value	Perform Site*
VHL Deletion Detection		Performed			MCR

RECEIVED: 08/01/2013 15:04 **REPORTED:** 08/01/2013 15:05

VHL Full Gene Analysis

RECEIVED: 08/01/2013 14:16 **REPORTED:** 08/01/2013 15:05

VHL Full Gene Analysis

Reason For Referral		Clinical features	MCR
Patient has clinical features suggestive of Von Hippel-Lindau (VHL) syndrome. Test for the presence of mutation(s) within the VHL gene.			
Method		Sequencing and MLPA	MCR
Fluorescent DNA sequence analysis was used to test for the presence of mutations in the 3 exons and exon-intron boundaries of the VHL gene. Additionally, multiplex ligation dependent probe amplification (MLPA) was used to test for the presence of large deletions in the VHL gene. This method utilizes probes for all 3 exons of the VHL gene.			
Result		Positive	MCR
This individual was shown to have a heterozygous large deletion involving the following exon(s) in the VHL gene: 1 through 3 (i.e. heterozygous deletion of all exons of the VHL gene).			
Interpretation		Positive for VHL	MCR
This result is consistent with a diagnosis of VHL syndrome. Therefore, this individual is at risk for developing symptoms associated with VHL. Appropriate screening procedures should be considered. Since a mutation has been identified in the VHL gene in this individual, genetic testing for this specific mutation in other family members is recommended. Please contact the Endocrine Laboratory at 1-800-533-1710 or the online test catalog at mayomedicallaboratories.com for information about how to order VHL Known Mutation (89084/VHLKP) for sequencing mutations, or VHL Deletion Detection (89211/VHLD) for large deletion mutations.			
Comment		GC consult	MCR
A genetic consultation may be of benefit.			
Reviewed By		Dee Feighner	MCR
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. Test results should be interpreted in the context of clinical findings, family history, and other laboratory			

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. A list of common polymorphisms identified for this patient is available from the Endocrine Laboratory upon request. Rarely, individuals may have a mutation or deletion in the gene(s) tested that is not identified by the described testing methodology. In addition, the phenotype observed in the individual tested here may be due to a variant in a gene not analyzed by this test. Laboratory developed test.					
VHL Gene Sequencing					
Sequencing		Performed			MCR

* Performing Site:

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