| **Patient Name** | **Patient ID** | **Age** | **Gender** | **Order #** |
|---|---|---|---|---|
| SAMPLEREPORT,VHLKP A | SA00060514 | 47 | F | SA00060514 |
| **Ordering Phys** CLIENT,CLIENT | | | | **DOB** 06/10/1966 |
| **Client Order #** SA00060514 | **Account Information** | | | **Report Notes** |
| **Collected** 07/31/2013 00:00 | C7028846-DLMP Rochester<br>3050 Superior Drive<br>Rochester, MN 55901 | | | |
| **Printed** 08/02/2013 11:21 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **VHL Known Mutation** | | | | | |
| **RECEIVED:** 08/01/2013 14:15 **REPORTED:** 08/01/2013 15:05 | | | | | |
| VHL Known Mutation | | | | | |
| Reason For Referral | | Familial mutation | | | MCR |
| A VHL gene mutation was previously identified in an affected family member. Test for the presence of the following mutation in the VHL gene: Exon 1, c.246C>T, p.Arg82Arg (p.R82R) | | | | | |
| Method | | Familial mutation | | | MCR |
| Fluorescent DNA sequence analysis was used to test for the presence of a specific mutation in the VHL gene, which was previously identified in an affected family member of this individual. | | | | | |
| Result | | Positive | | | MCR |
| This individual was shown to be heterozygous for the familial mutation: Exon 1, c.246C>T, p.Arg82Arg (p.R82R) | | | | | |
| Interpretation | | Positive | | | MCR |
| The above mutation was previously identified in a family member affected with Von Hippel-Lindau (VHL) syndrome. The presence of this mutation in the individual tested here is consistent with a diagnosis of VHL syndrome for this individual. 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. | | | | | |
| 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 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 | | | | | |

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|---|---|---|---|---|---|
| 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 Known Mutation Sequencing | | | | | |
| Sequencing | | Performed | | | MCR |

* Performing Site:

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