MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CSRKP ABNORMAL	SA00056256	46	F	SA00056256
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00056256	**Account Information**			**Report Notes**
Collected 03/27/2013	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 05/15/2013 12:18				

Test	Flag	Results	Unit	Reference Value	Perform Site*
CASR Known Mutation			REPORTED 03/28/2013 10:54		
CASR Known Mutation					
Reason for Referral		Familial mutation			MCR
A calcium sensing receptor (CASR) gene mutation was previously identified in an affected family member. Test for the presence of the following mutation in the CASR gene: Exon 2, DNA change c.141A>C , Amino Acid change p.K47N					
Method		Method			MCR
Fluorescent DNA sequence analysis was used to test for the presence of a specific alteration in the CASR gene (GenBank accession number: NM_000388.2), which was previously identified in an affected family member of this individual.					
Result		Positive 2			MCR
The following heterozygous sequence changes were detected: Exon:2 DNA change: c.141A>C Amino Acid change: p.K47N This alteration is known to be disease-associated. Exon: 3 DNA change: c.382T>C Amino Acid change: p.F128L This alteration is known to be disease-associated.					
Interpretation		Positive			MCR
This result suggests that this individual is at risk for developing symptoms associated with abnormal CASR function. Appropriate screening and measures should be considered. Since a CASR mutation(s) has been identified, genetic testing of at risk family members is possible.					
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					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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.

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.