MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CSRSP ABNORMAL	SA00055919	46	F	SA00055919
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00055919	**Account Information**			**Report Notes**
Collected 03/13/2013 13:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 05/15/2013 12:15				

Test	Flag	Results	Unit	Reference Value	Perform Site*
CASR Full Gene Analysis			REPORTED 03/19/2013 11:42		
CASR Full Gene Analysis					
Reason for Referral		No Info			MCR
Not provided. Test for the presence of a mutation in the CASR gene.					
Method		Method			MCR
DNA sequencing analysis was used to test for the presence of a mutation in all 6 coding exons of the CASR gene (GenBank accession number: NM_000388.2).					
Result		Positive 1			MCR
The following heterozygous sequence change was detected: Exon: 2 DNA change: c.2T>G Amino Acid change: p.M1R This alteration is known to be disease-associated.					
Interpretation		FHH positive			MCR
This result is consistent with a diagnosis of familial hypocalciuric hypercalcemia (FHH). Age appropriate screening and management options 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		LYNETTE HANSEN			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 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:

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