

Patient Name RVTEST,DMGLM290	Patient ID RVDMGLM290	Age 34	Gender F	Order # RVDMGLM290
Ordering Phys				DOB 12/21/1980
Client Order # RVDMGLM290	Account Information			Report Notes
Collected 04/07/2015 12:55	C7028846-DLMP Rochester SDSC 2 - Client Support			
Printed 07/02/2015 09:29	Rochester, MN 55901			

Test	Flag	Results	Unit	Reference Value	Perform Site*
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Familial Mutation, Targeted Testing

Result Summary	POSITIVE	MCR
Result		MCR

The following heterozygous alteration was identified:

Amino Acid change: p.C609Y (Cys609Tyr)

DNA change: c.1826G>A (g.43609070)

Classification: PATHOGENIC

Interpretation

MCR

The c.1826G>A (p.C609Y) alteration is a known pathogenic mutation.

This result is consistent with a diagnosis of multiple endocrine neoplasia type 2A (MEN 2A)/familial medullary thyroid carcinoma (FMTC) for this individual. Appropriate surveillance procedures and/or treatment strategies should be considered.

Since a mutation has been identified, genetic testing of at risk family members could be considered.

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.

-----ADDITIONAL INFORMATION-----

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen.

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.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone Marrow transplants from allogenic donors will

Performing Site Legend on Last Page of Report

Patient Name RVTEST,DMGLM290	Collection Date and Time 04/07/2015 12:55	Report Status Final
Page 1 of 2		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

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interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. These tools are updated regularly, therefore changes to these algorithms may result in different predictions for a given alteration. However, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Laboratory developed test.
 PDF Report available at:

<https://test.mmlaccess.com/Reports/C7028846-i3LoHiJQBZ.ashx>

Specimen	WB Whole Blood	MCR
Method		MCR
	DNA sequence analysis was used to test for the presence of the p.C609Y (c.1826G>A; g.43609070) alteration in the RET gene (GenBank accession number NM_020975; build GRCh37 (hg19)).	
Released By	ANDE RUMILLA	MCR

RECEIVED: 04/07/2015 12:55 **REPORTED:** 04/17/2015 04:55

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: William G. Morice, II, M.D., Ph.D.
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Patient Name RVTEST,DMGLM290	Collection Date and Time 04/07/2015 12:55	Report Status Final
Page 2 of 2		** End of Report **

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Performing Site:

Mayo Clinic Laboratories - Rochester Main Campus
200 First Street SW, Rochester MN 55905
William G. Morice, II, M.D., Ph.D. Lab Director
Phone: 800-533-1710
<http://www.mayomedicallaboratories.com>

RVTEST, DMGLM290

MEDICAL RECORD # (PATIENT ID) RVDMLM290

DOB	12/21/1980	CLIENT ID/WARD	7028846	ORDER #	D307000599
SEX	Female	CLIENT/NAME WARD	DLMP Rochester		
CLIENT MRN	RVDMLM290	CITY, ST, ZIP	Rochester MN 55901	DATE COLLECTED	4/7/2015 12:55 PM
REQUESTED BY	ROBERT ELLSWORTH WHAREN			DATE RECEIVED	4/7/2015 12:55 PM
				DATE REPORTED	4/17/2015 4:55 AM

Familial Mutation, Targeted Testing

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Released By:

ANDE RUMILLA

QA TESTING