MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
RVTEST,DMGLM502	RVDMGLM502	34	F	RVDMGLM502

Ordering Phys		DOB
		12/21/1980

Client Order #	Account Information	Report Notes
RVDMGLM502	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901	
Collected 04/27/2015 21:56		
Printed 06/11/2015 14:34		

Test	Flag	Results	Unit	Reference Value	Perform Site*
DMD/BMD Deletion/Duplication					
Result Summary		POSITIVE			MCR
Result					MCR

The following deletion was identified:
Exon(s): 3-16

Interpretation MCR

This result indicates that this individual is a carrier of Duchenne Muscular Dystrophy (DMD). This assumes that the entire exon associated with the affected probes is deleted. In addition, studies suggest that female carriers of DMD may be at risk for developing symptoms related to the disease.

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

A genetic consultation may be of benefit.

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

Gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in the DMD gene (GenBank accession number NM_004006; build GRCh37 (hg19)).
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 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. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Laboratory developed test.

Performing Site Legend on Last Page of Report

RVTEST,DMGLM502	04/27/2015 21:56	Final
		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
PDF Report available at: https://test.mmlaccess.com/Reports/C7028846-Jm6iG38o3a.ashx					
Specimen		WB Whole Blood			MCR
Released By		See Below			MCR
Result: D. Brian Dawson, Ph.D.					

RECEIVED: 04/27/2015 21:56 **REPORTED:** 04/29/2015 14:31

* 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.

MAYO CLINIC
Mayo Medical Laboratories

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, DMGLM502 **MEDICAL RECORD # (PATIENT ID)** RVDMGLM502

DOB	12/21/1980	**CLIENT ID/WARD**	7028846	**ORDER #**	D327000935
SEX	Female	**CLIENT/NAME WARD**	DLMP Rochester		
CLIENT MRN	RVDMGLM502	**CITY, ST, ZIP**	Rochester	**DATE COLLECTED**	4/27/2015 9:56 PM
REQUESTED BY	ROBERT ELLSWORTH WHAREN		MN 55901	**DATE RECEIVED**	4/27/2015 9:56 PM
				DATE REPORTED	4/29/2015 2:31 PM

DMD/BMD Deletion/Duplication

Result Summary:
POSITIVE

Result:
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Exon(s): 3-16

Interpretation:
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Specimen:
WB Whole Blood

Method:
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Released By:
D. Brian Dawson, Ph.D.

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