

Patient Name RVTEST,DMGLM501	Patient ID RVDMGLM501	Age 34	Gender F	Order # RVDMGLM501
Ordering Phys				DOB 12/21/1980
Client Order # RVDMGLM501	Account Information C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			Report Notes
Collected 04/27/2015 21:53				
Printed 06/11/2015 14:33				

Test	Flag	Results	Unit	Reference Value	Perform Site*
DMD/BMD Deletion/Duplication					
Result Summary		NEGATIVE			MCR
Result		No deletions or duplications were identified.			MCR
Interpretation		This result reduces the likelihood, but does not rule out positive carrier status for Duchenne Muscular Dystrophy (DMD) or Becker Muscular Dystrophy (BMD). This assay does not exclude the presence of non-deletion and duplication types of mutations, such as point mutations. Additionally, there is an increased risk for germline mosaicism in women whose only affected family member is their son. For these women other offspring would still be at risk for inheriting a dystrophin mutation. Therefore, this result should be interpreted in the context of clinical presentation and results of other laboratory tests. If testing was performed due to a family history of DMD or BMD, genetic testing of an affected individual should be considered. Identification of the mutation in the family would allow for more accurate risk assessment of at risk individuals. 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			MCR

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
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RVTEST,DMGLM501	04/27/2015 21:53	Final
Page 1 of 2		>> 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*
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.					
PDF Report available at:					
https://test.mmlaccess.com/Reports/C7028846-kBdmrIIIoJ.ashx					
Specimen		WB Whole Blood			MCR
Released By		See Below			MCR
Result: D. Brian Dawson, Ph.D.					
RECEIVED: 04/27/2015 21:53 REPORTED: 04/29/2015 14:28					

* 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,DMGLM501	Collection Date and Time 04/27/2015 21:53	Report Status Final
Page 2 of 2		** End of Report **

* Report times for Mayo performed tests are CST/CDT

RVTEST, DMGLM501**MEDICAL RECORD # (PATIENT ID) RVDMLM501**

DOB	12/21/1980	CLIENT ID/WARD	7028846	ORDER #	D327000934
SEX	Female	CLIENT/NAME WARD	DLMP Rochester		
CLIENT MRN	RVDMLM501	CITY, ST, ZIP	Rochester	DATE COLLECTED	4/27/2015 9:53 PM
REQUESTED BY	ROBERT ELLSWORTH WHAREN	MN	55901	DATE RECEIVED	4/27/2015 9:53 PM
				DATE REPORTED	4/29/2015 2:28 PM

DMD/BMD Deletion/Duplication**Result Summary:**

NEGATIVE

Result:

No deletions or duplications were identified.

Interpretation:

This result reduces the likelihood, but does not rule out positive carrier status for Duchenne Muscular Dystrophy (DMD) or Becker Muscular Dystrophy (BMD).

This assay does not exclude the presence of non-deletion and duplication types of mutations, such as point mutations. Additionally, there is an increased risk for germline mosaicism in women whose only affected family member is their son. For these women other offspring would still be at risk for inheriting a dystrophin mutation. Therefore, this result should be interpreted in the context of clinical presentation and results of other laboratory tests.

If testing was performed due to a family history of DMD or BMD, genetic testing of an affected individual should be considered. Identification of the mutation in the family would allow for more accurate risk assessment of at risk individuals.

A genetic consultation may be of benefit.

Specimen:

WB Whole Blood

Method:

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)).

Disclaimer:

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.

PATIENT NAME: RVTEST, DMGLM501

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The collected, received, and reported dates and times on the report are in the time zone of the performing location.

DOB	12/21/1980	CLIENT ID/WARD	7028846	ORDER #	D327000934
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CLIENT MRN	RVDMLM501	CITY, ST, ZIP	Rochester	DATE COLLECTED	4/27/2015 9:53 PM
REQUESTED BY	ROBERT ELLSWORTH WHAREN	MN	55901	DATE RECEIVED	4/27/2015 9:53 PM
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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.

Released By:

D. Brian Dawson, Ph.D.

QA TESTING