

1-800-533-1710

PATIENT NAME			PATIENT NU	MBER		AGE	SEX	ACCESSION #
TESTING, KM			L3MRNG91	65196		34	F	G9165196
ORDERING PHYSICIAN			CLIENT ORD	ER#				ACCOUNT # LIAISONS
COLLECTION RECEIVED		REPORT PRI	REPORT PRINTED SPECIMEN INFORMA		TION			
02/23/11 08:25 A	02/23/11 0	8:25 A	03/18/11	10:04 A	DATE OF BIRTH:			
DATE TIME	DATE	TIME	DATE	TIME				
Test Client								
Attn: Mayo Liaisons								
200 First Street SW								
Rochester, MN 55905								
507-284-8202								

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TEST REQUESTED LO REF RANGE PERFORM SITE *

DNA Extraction, NPL REPORTED: 02/23/11 08:33 A

 Specimen
 Blood
 MCR

 Specimen ID
 1035206
 MCR

 Order Date
 23 Feb 2011 08:30
 MCR

 Comment
 MCR

Genomic DNA has been extracted.

Reviewed By MCR

Michelle Dawn Fjerstad

Release Date 23 Feb 2011 08:31 MCR

TGFBR1 Gene, Known Mutation REPORTED: 02/25/11 02:14 P

TGFBR1 Result MCR

This individual was shown to have one copy of the following familial mutation in the TGFBR1 gene:

Exon 9, c.1459C>T,p.R487W

TGFBR1 Interpretation MCR

The above mutation within the TGFBR1 gene was previously identified in a family member affected with Loeys-Dietz syndrome (LDS). The presence of this mutation, therefore, is consistent with a diagnosis of LDS in this individual.

TGFBR1 Reviewed By Loralie J. Langman, MCR

Ph.D.

Laboratory developed test.

Fluorescent DNA sequence analysis was used to test for the presence of a specific mutation in the TGFBR1 gene, which was previously identified in an affected family member of this individual.

A genetic consultation may be of benefit.

CAUTIONS:

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.

* Perform Site Legend on last page of report

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, KM	Final	02/23/11 08:25 A



1-800-533-1710

PATIENT N	NAME			PATIENT NUM	IBER		AGE	SEX	ACCESSION #
TESTING, KM			L3MRNG9165196			34	F	G9165196	
ORDERING	G PHYSICIAN			CLIENT ORDE	R#				ACCOUNT # LIAISONS
COLLECT	COLLECTION RECEIVED		REPORT PRINTED SPECIMEN INFORMA		TION				
02/23/11	08:25 A			03/18/11	10:04 A	DATE OF BIRTH:			
DATE	TIME	DATE	TIME	DATE	TIME				
Test Clie	ent								
Attn: Ma	ayo Liaisons								
200 Firs	t Street SW								
Rochester, MN 55905									
507-284	-8202								

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TEST REQUESTED LO REF RANGE PERFORM SITE *

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.

TGFBR1 Known Performed MCR

Mutation Sequencing

* PERFORMING SITE

MCR Mayo Clinic Dpt of Lab Med & Pathology
Lab Director: Franklin R. Cockerill, III, M.D.
200 First Street SW Rochester, MN 55905

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, KM	Final	02/23/11 08:25 A