

1-800-533-1710

<b>PATIENT NAME</b> TESTING, TGF1		<b>PATIENT NUMBER</b> L3MRNG9165187		<b>AGE</b> 34	<b>SEX</b> F	<b>ACCESSION #</b> G9165187
<b>ORDERING PHYSICIAN</b>			<b>CLIENT ORDER #</b>		<b>ACCOUNT #</b> LIAISONS	
<b>COLLECTION</b> 02/22/11 01:34 P	<b>RECEIVED</b> 02/22/11 01:34 P	<b>REPORT PRINTED</b> 03/18/11 10:02 A		<b>SPECIMEN INFORMATION</b> DATE OF BIRTH:		
<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>				
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
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**DNA Extraction, NPL** **REPORTED: 02/22/11 01:49 P**

Specimen Blood MCR

Specimen ID 1035203 MCR

Order Date 22 Feb 2011 13:42 MCR

Comment MCR

Genomic DNA has been extracted.

Reviewed By MCR

Michelle Dawn Fjerstad

Release Date 22 Feb 2011 13:46 MCR

**TGFBR1 Full Gene Sequence** **REPORTED: 02/25/11 02:13 P**

**TGFBR1 Result** MCR

This individual was shown to have one copy of the following mutation in TGFBR1:  
Exon 4, c.722C>T,p.S241L

**TGFBR1 Interpretation** MCR

This result is consistent with a diagnosis of Loeys-Dietz syndrome.  
Since a mutation has been identified in the TGFBR1 gene in this individual, genetic testing for this specific mutation in other family members is recommended. Please contact the laboratory at 1-800-533-1710 or the online test catalog at [www.mayomedicallaboratories.com](http://www.mayomedicallaboratories.com) for information about how to order testing for a specific familial mutation (89460, "TGFBR1 Gene, Known Mutation").

**TGFBR1 Reviewed By** MCR

John L. Black, M.D.  
Laboratory developed test.  
Fluorescent DNA sequence analysis was used to test for the presence of mutations in all 9 exons and exon-intron boundaries of the TGFBR1 gene.  
A genetic consultation may be of benefit.  
A list of common polymorphisms identified for this patient is

\* Perform Site Legend on last page of report

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available from the lab upon request.

**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.

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 Full Gene  
Sequencing Result**

**Performed**

**MCR**

\* PERFORMING SITE

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