

## **Laboratory Service Report**

### 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #	
SAMPLEREPORT,TGF2	SA00055117	46	F	SA00055117	
Ordering Phys				DOB	
UNKNOWN,PROVIDER				06/10/1966	
Client Order #	Account Information	Account Information			
SA00055117					
Collected	C7028846-DLMP Roch	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
03/14/2013 22:52	•				
Printed	Rochester, MN 55901				
06/13/2013 09:44					

Test Flag Results Unit Value Site\*

TGFBR2 Full Gene Sequence

**RECEIVED:** 03/14/2013 22:52 **REPORTED:** 06/13/2013 09:36

TGFBR2 Full Gene Sequence

MCR

One copy of the following mutation was detected in TGFBR2: Exon 7, nucleotide c. 1589\_1590delCA, amino acid p. Thr530SerfsX10. This deletion creates a frame shift starting at codon Thr530. The new reading frame ends in a STOP codon 9 positions downstream.

TGFBR2 Interpretation

MCR

This TGFBR2 mutation is consistent with the Loeys-Dietz features observed in this individual. Appropriate surveillance and management strategies should be considered.

Since a mutation has been identified in the TGFBR2 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 on-line test catalog at www.mayomedicallaboratories.com for information about how to order the test for TGFBR2 Gene, Known Mutation (89462). Please refer to family number when ordering testing on family members of this individual.

Fluorescent DNA sequence analysis was used to test for the presence of mutations in all 7 exons and exon-intron boundaries of the TGFBR2 gene, isoform B (GenBank accession number NM $\_003242.5$ ). Exon 2 from TGFBR2 isoform A, and exon 5 from TGFBR2 isoform C, were also sequenced along with their exon-intron boundaries.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is 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.

#### \*\*\*Performing Site Legend on Last Page of Report\*\*\*

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,TGF2	03/14/2013 22:52	Final
Page 1 of 2		>> Continued on Next Page >>

<sup>\*</sup> Report times for Mayo performed tests are CST/CDT



# **Laboratory Service Report**

1-800-533-1710

Patient Name SAMPLEREPORT,TGF2	Patient ID SA00055117	<b>Age</b> 46	Gender F	<b>Order #</b> SA00055117	
Ordering Phys UNKNOWN,PROVIDER				<b>DOB</b> 06/10/1966	
Client Order # SA00055117	Account Information	Account Information  C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901		Report Notes	
<b>Collected</b> 03/14/2013 22:52	3050 Superior Drive				
<b>Printed</b> 06/13/2013 09:44	Rochester, MN 55901				

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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.

Laboratory developed test.

TGFBR2 Reviewed by TGFBR2 Full Gene Sequencing

Yvonne Philo Performed MCR MCR

* Performing Site:	:
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	MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:	

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,TGF2	03/14/2013 22:52	Final
Page 2 of 2		** End of Report **

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