MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,TGF2	SA00055117	46	F	SA00055117
Ordering Phys UNKNOWN,PROVIDER				**DOB** 06/10/1966
Client Order # SA00055117	**Account Information**			**Report Notes**
Collected 03/14/2013 22:52	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/13/2013 09:56				

Test	Flag	Results	Unit	Reference Value	Perform 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

* Report times for Mayo performed tests are CST/CDT

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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		Yvonne Philo			MCR
TGFBR2 Full Gene Sequencing		Performed			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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