



Patient ID <b>SA00055106</b>	Patient Name <b>SAMPLEREPORT, TGF1</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>46</b>
Order Number <b>SA00055106</b>	Client Order Number <b>SA00055106</b>	Ordering Physician <b>UNKNOWN, PROVIDER</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>14 Mar 2013 22:49</b>		

## TGFBR1, Full Gene Sequence

### TGFBR1 Full Gene Sequence

#### TGFBR1 Result

MCR

A mutation was not detected in TGFBR1.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available from the lab upon request.

#### TGFBR1 Interpretation

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This result does not rule out the diagnosis of Loeys-Dietz syndrome (LDS) or other TGFBR1-associated syndrome. Some individuals with features of LDS or other TGFBR1-associated syndrome may have a TGFBR1 mutation that is not identified by the described testing methodology. In addition, some individuals with this phenotype may have a mutation in a gene other than TGFBR1, such as FBN1 or TGFBR2. If applicable, please contact the laboratory at 1-800-533-1710 or the on-line test catalog at [www.mayomedicallaboratories.com](http://www.mayomedicallaboratories.com) for information about how to order the test for FBN1, Full Gene Sequence (89308) and/or TGFBR2, Full Gene Sequence (89461).

#### 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.

#### ADDITIONAL INFORMATION

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 (GenBank accession number NM\_004612.2).

If the patient has had an allogeneic blood or marrow transplant or a recent (i.e. less than 6 weeks from time of sample collection) heterologous blood transfusion these results may be inaccurate due to the presence of donor DNA. Laboratory developed test.

#### TGFBR1 Reviewed by

MCR

Yvonne Philo

### TGFBR1 Full Gene Sequencing

MCR

Performed

**Received:** 14 Mar 2013 22:49

**Reported:** 13 Jun 2013 09:38

#### Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905