

Laboratory Service Report

1-800-533-1710

MCR

Patient Name SAMPLEREPORT,TGFK1	Patient ID SA00055120	Age 46	Gender F	Order # SA00055120
Ordering Phys UNKNOWN,PROVIDER			•	DOB 06/10/1966
Client Order # SA00055120	Account Information			Report Notes
Collected 03/14/2013 22:53	C7028846-DLMP Rochester 3050 Superior Drive			
Printed 06/13/2013 09:47	Rochester, MN 55901			

Reference Perform
Test Flag Results Unit Value Site*

TGFBR1 Gene, Known Mutation

RECEIVED: 03/14/2013 22:53 **REPORTED:** 06/13/2013 09:36

TGFBR1 Gene, Known Mutation

TGFBR1 Result MCR

The TGFBR1 Exon 9, nucleotide c. 1459C>T, amino acid p. $Arg487Trp\ (p.\ R487W)$ familial mutation was not detected in this individual.

TGFBR1 Interpretation

The TGFBR1 p.R487W mutation was previously identified in a family member affected with Loeys-Dietz syndrome (LDS). Since this mutation was not detected in this individual, this suggests that this patient is at no greater risk than someone in the general population for having LDS. Fluorescent DNA sequence analysis was used to test for the presence of a specific mutation in the TGFBR1 gene (GenBank accession number NM_004612.2), 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.

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. Laboratory developed test.

TGFBR1 Reviewed by Yvonne Philo MCR
TGFBR1 Known Mutation Sequencing Performed MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus	Lab Director:	
IVICK	200 First St SW Rochester, MN 55905	Lab Director.	

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,TGFK1	03/14/2013 22:53	Final
Page 1 of 1		** End of Report **

^{*} Report times for Mayo performed tests are CST/CDT