Patient ID	Patient Name		Birth Date	Gender	Age
SA00055120	SAMPLEREPORT, TGFK1		1966-06-10	F	46
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00055120	SA00055120	UNKNOWN, PROVIDER			
Account Information		**Collected**			
C7028846 DLMP Rochester		14 Mar 2013 22:53			

TGFBR1 Gene, Known Mutation

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

MCR

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.

ADDITIONAL INFORMATION

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.

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 Known Mutation Sequencing

MCR

Performed

Received: 14 Mar 2013 22:53 **Reported:** 13 Jun 2013 09:36

Performing Site Legend

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