



Patient ID <b>SA00055123</b>	Patient Name <b>SAMPLEREPOR, TGFK1</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>46</b>
Order Number <b>SA00055123</b>	Client Order Number <b>SA00055123</b>	Ordering Physician <b>UNKNOWN, PROVIDER</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>14 Mar 2013 22:55</b>		

## TGFBR1 Gene, Known Mutation

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#### TGFBR1 Result

MCR

One copy of the TGFBR1 Exon 4, nucleotide c.782G>A, amino acid p.Gly261Glu (p.G261E) familial mutation was detected in this individual.

#### TGFBR1 Interpretation

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The TGFBR1 p.G261E mutation was previously identified in a family member with features of Loeys-Dietz syndrome (LDS). The presence of this mutation, therefore, suggests that this individual is at risk for development and/or exacerbation of features of LDS. Appropriate surveillance procedures and/or management strategies should be considered.

#### 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 22:55

**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