

Patient ID SA00055117	Patient Name SAMPLEREP, TGF2	Birth Date 1966-06-10	Gender F	Age 46
Order Number SA00055117	Client Order Number SA00055117	Ordering Physician UNKNOWN, PROVIDER	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Mar 2013 22:52		

TGFBR2 Full Gene Sequence

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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

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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.

ADDITIONAL INFORMATION

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.

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.

TGFBR2 Reviewed by

MCR

Yvonne Philo

TGFBR2 Full Gene Sequencing

MCR

Performed

Received: 14 Mar 2013 22:52

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