MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

TACIF

Transmembrane Activator and CAML Interactor (TACI) Gene, Full Gene Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
0000181056	SAMPLEREPORT, TACIF		1966-06-10	F	46
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
0000181056	0000181056	,			
Account Information		**Collected**			
C7028846 DLMP Rochester		15 Mar 2013 03:41			

TACI, Full Gene Sequence

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Reason for Referral

MCR

CVID, considering BMT

Method

MCR

Fluorescent DNA sequence analysis was used to test for the presence of mutations in the 5 exons, exon-intron boundaries, and the 5' and 3' UTR of the TACI gene.

Result

MCR

No clinically significant mutations or variations were identified in this patient in any of the 5 coding exons or intron-exon boundaries of the TNFRSF13B (TACI) gene.

Interpretation

MCR

No clinically significant mutations or variations were identified in this patient in any of the 5 coding exons or intron-exon boundaries of the TNFRSF13B (TACI) gene. TACI mutations have been reported to account for 8–15% of CVID cases depending on the study population assessed. The majority of TACI mutations are sporadic though some familial mutations (autosomal dominant and autosomal recessive) have been reported. Present evidence seems to suggest that homozygous TACI mutations are associated with CVID and/or selective IgA deficiency, while the clinical significance of heterozygous mutations remains controversial, though these have also been reported in some CVID and IgA-deficient patients. The underlying genetic defect or molecular basis has not been identified in the majority (75–80%) of CVID patients. Therefore, it may be reasonable to regard most cases of CVID as oligogenic or polygenic, i.e., the phenotype results from the contribution of more than a single gene defect. More recent evidence (Orange et al, J Allergy Clinical Immunology, 2011, 127: 1360–1367) suggests that copy number variations (CNV) - duplications and/or deletions are present in a large subset of CVID patients.

Signaling through the TACI receptor has been shown to be important for IgA class-switching, especially in the gut. Therefore, IgAD has been reported to be associated with certain TNFRSF13B gene mutations. However, since selective IgA deficiency (sIgAD) is relatively common in the general population (incidence approximately 1:200 to 1:1000), the etiology underlying sIgAD is likely multifactorial and variable in individuals.

ADDITIONAL INFORMATION

Laboratory developed test.

Reviewed By

MCR

Yvonne Philo

TACI Gene Sequencing

MCR

Performed

Received: 15 Mar 2013 03:41 **Reported:** 13 Jun 2013 12:47

Performing Site Legend

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