MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

TACIG

Transmembrane Activator and CAML Interactor (TACI) Gene, Known Mutation Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00055100	SAMPLEREPORT, TACIG		1966-06-10	F	46
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00055100	SA00055100	UNKNOWN, PROVIDER			
Account Information		**Collected**			
C7028846 DLMP Rochester		15 Mar 2013 03:42			

TACI Gene, Known Mutation

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Reason for Referral MCR

CVID

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

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. Only common polymorphisms are noted. TACI gene 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. Typically assessment of quantitative immunophenotyping of B cells (test #88800) is recommended, if not previously evaluated, to determine if class-switched memory B cells and other relevant B cell subsets are present, absent or decreased. B cell subset analysis is not diagnostic for CVID but may be helpful in determining prognosis and classification. In this patient, the reported absence of B cells precludes this analysis. Approximately 5% of CVID patients have <1% B cells, suggestive of a defect in early B cell differentiation.

ADDITIONAL INFORMATION

Laboratory developed test.

Reviewed By MCR

Yvonne Philo

TACI Known Mutation Sequencing

Sequencing MCR

Performed

Received: 15 Mar 2013 03:42 **Reported:** 13 Jun 2013 12:50

Performing Site Legend

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