

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,TACIG	Patient ID SA00055100	Age 46	Gender F	Order # SA00055100	
Ordering Phys UNKNOWN,PROVIDER		·		DOB 06/10/1966	
Client Order # SA00055100	Account Information			Report Notes	
Collected 03/15/2013 03:42	C7028846-DLMP Rock 3050 Superior Drive	nester			
Printed 06/13/2013 15:38	Rochester, MN 55901				

Test Flag Results Unit Value Site*

TACI Gene, Known Mutation

RECEIVED: 03/15/2013 03:42 **REPORTED:** 06/13/2013 12:50

TACI Gene, Known Mutation

Reason for Referral

CVID Method

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.

Interpretation

MCR MCR

MCR

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. Laboratory developed test.

Performing Site Legend on Last Page of Report

	Collection Date and Time 03/15/2013 03:42	Report Status Final
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Reviewed By		Yvonne Philo			MCR
TACI Known Mutation Sequencing Sequencing		Performed			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
	200 First St SW Rochester MN 55905	· ·

	Collection Date and Time 03/15/2013 03:42	Report Status Final
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^{*} Report times for Mayo performed tests are CST/CDT