MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

ALPS

Alpha Beta Double-Negative T Cells for Autoimmune Lymphoproliferative Syndrome

Patient ID	Patient Name		Birth Date	Gender	Age
SA00057233	SAMPLEREPORT, ALPS		1986-06-10	F	26
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00057233	SA00057233	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		06 May 2013 00:00			

ALPS Screen

%alpha/beta-TCR DNT — MCR

⚠ 6.5 % CD3 T cells — High

Reference Value <3.0

alpha/beta-TCR DNT — MCR

⚠ 49 cells/mcL — High

Reference Value <35

% TCR+DNT B220+ — MCR

⚠ 1.2 % CD3 T cells — High

Reference Value <0.3

Absolute TCR+DNT B220+ — MCR

⚠ 9 cells/mcL — High

Reference Value <6

Interpretation — [1] MCR

Increased frequency (%) and absolute count of abTCR+ DNT cells and these cells are positive for B220 expression. This result appears to support a diagnosis of Autoimmune Lymphoproliferative Syndrome (ALPS). However, the flow cytometric assay quantifying the % and absolute counts of alpha-beta TCR+ DNT cells and DNT B220+ T cells is to be used only as a screening assay for ALPS and cannot be considered a confirmatory diagnostic test.

Abnormal results should be confirmed by correlating clinical history along with in vitro assessment of apoptosis defects (available at Cincinnati Childrens' Hospital) and/or genetic testing for the relevant gene mutations (ALPS Type 0 (homozygous germline FAS mutations), Type 1a (heterozygous germline FAS mutations), Type 1m (somatic FAS mutations), Type 1b (germline FASLG - Fas ligand mutations), Type 11a (germline CASP10 - caspase 10 mutations), Type III (unknown genetic defect). Genetic testing is available at Cincinnati Childrens Hospital or Gene Dx).

Unable to provide further interpretation without clinical history. Clinical correlation recommended.

ADDITIONAL INFORMATION

New reference values will be used for the ALPS assay. In addition, a marker - B220 that improves the specificity of the assay has been included along with the evaluation of alpha-beta TCR+ DNT cells. Changes in reporting effective 05/28/2008.

Received: 07 May 2013 11:13 **Reported:** 07 May 2013 11:37

Laboratory Notes

[1] Analyte Specific Reagent: This test was developed and its performance characteristics determined by Mayo Clinic. It has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

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