



Patient ID SA00057240	Patient Name SAMPLEREPORT, XHIM	Birth Date 1986-06-10	Gender M	Age 26
Order Number SA00057240	Client Order Number SA00057240	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 06 May 2013 00:00		

X-Linked Hyper IgM Syndrome, B

Interpretation

1 MCR

There is normal activation of CD4 T cells with CD69 expression. There is complete absence of CD40L (CD154) expression on activated T cells and also there is no function of the ligand (absent binding of ligand to soluble receptor - CD40-mulg) as the protein is absent from the cell surface. This result is consistent with a diagnosis of X-linked Hyper IgM syndrome (CD40L deficiency) for this male patient. Recommend genetic testing for identification of the pathogenic mutation in the CD40LG gene. Also, recommend carrier testing for mother and other female relatives as appropriate. Carrier testing may be performed with this flow cytometry assay.

CD40 Ligand Expression

MCR



Absent

Reference Value
Present

CD40mulg (Function)

MCR



Absent

Reference Value
Present

Received: 07 May 2013 11:16

Reported: 07 May 2013 12:35

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