

Patient ID SAMPLE	Patient Name PATIENT, TEST	Birth Date 1989-08-04	Sex M	Age 35
Order Number SAMPLE	Client Order Number SAMPLE	Ordering Physician SAMPLE	Report Notes Sample Report	
Account Information SAMPLE		Collected 22 Sep 2024 14:37		

X-Linked Hyper IgM Syndrome, B

Hyper IgM Functional



Absent

Abn

CD40mulg (Function)



Absent

Abn

SDL

Reference Value
Present

SDL

Reference Value
Present

Interpretation

1 SDL

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. The 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.

Received: 23 Sep 2024 14:32

Reported: 23 Sep 2024 14:37

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	Lab Director	CLIA Certificate
SDL	Mayo Clinic Laboratories - Rochester Superior Drive	3050 Superior Drive NW, Rochester MN 55905	Nikola Baumann Ph.D.	24D1040592