



**UNC**  
HEALTH CARE  
MEMORANDUM #182

**TO:** UNCHCS Attending Physicians, Housestaff, Clinical Nurse Coordinators, Department Heads and Supervisors

**FROM:** *HCV* Herbert C. Whinna, MD, PhD, Medical Director, McLendon Clinical Laboratories  
*EW* Eric Weimer, PhD, Director, Molecular Immunology Laboratory

**DATE:** December 17, 2018

**SUBJECT: Genetic Testing for Primary Immune Deficiencies (SCID, HLH, ALPS, Hyper IgE, Hyper IgM)**

Effective January 2, 2019, UNC McLendon Clinical Laboratories (MCL) Molecular Immunology Laboratory will offer genetic testing for several primary immune deficiencies including severe combined immunodeficiency (SCID), familial hemophagocytic lymphohistiocytosis (HLH) types 2-5, autoimmune lymphoproliferative syndrome (ALPS), hyper IgE, and hyper IgM.

EPIC Order Name	Test ID	TAT
Severe Combined Immunodeficiency (SCID)	LAB6745	14 days
Hemophagocytic Lymphohistiocytosis (HLH)	LAB6746	14 days
Hyper IgE (HIES)	LAB6747	14 days
Hyper IgM (HIGM)	LAB6748	14 days
Autoimmune Lymphoproliferative Syndrome (ALPS)	LAB6749	14 days

The genes tested for each order are shown below.

SCID			HLH		Hyper IgE	Hyper IgM	ALPS
ADA	ORAI1	RFXAP	AP3B1	STX11	STAT3	CD40LG	CASP8
CD3D	PNP	TAP1	BLOC1S6	STXBP2	GATA2	UNG	CASP10
CD3E	PTPRC	TAP1	CD27	UNC13D	DOCK8	AICDA	FADD
DCLRE1C	RAG1	CIITA	ITK	XIAP	CXCR4	CD40	FASLG
FOXN1	RAG2	RFX5	LYST		SPINK5		ITK
IL2RG	RMRP	TAPBP	MAGT1				KRAS
IL7R	STAT5B		PRF1				FAS
JAK3	STIM1		RAB27A				MAGT1
LIG4	TBX1		SH2D1A				NRAS
NHEJ1	ZAP70		SLC7A7				CTLA4

Testing may be ordered Monday – Friday using the EPIC Order Name or Test ID shown above. Specimen must be received in the laboratory by 1430 on Fridays.

Results will be available in EPIC under Laboratory Results>Molecular Pathology.

For questions or more information regarding this change, please contact Dr. Eric Weimer at 984-974-1451.