

CLINICAL IMMUNODIAGNOSTIC AND RESEARCH LAB

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A Jeffrey Modell Diagnostic Center for Primary Immunodeficiency

TEST DESCRIPTION OVERVIEW

TEST NAME	DESCRIPTION	TEST CODE	CPT CODE(S)	ANTIGENS INVESTIGATED			
				<u>T-Subsets</u>	<u>B-Subsets</u>	<u>NK-Subsets</u>	<u>Other</u>
ABSOLUTE AT4	To monitor patient's individual CD4 and CD8 T cell populations. Panel is routinely used in determining therapeutic considerations for HIV+ patients.	AT4	88184-TC 88185 x4-TC 86359-TC 88187	CD3 CD4 CD8			CD14 CD45
AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME	Evaluates the presence of TCR alpha/beta positive –CD4 and CD8 double negative T cells, referred to as TCRab-DNTcs.	AILYMP	88184-TC 88185 x13-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 B220 TCRab TCRgd	CD19 CD21 CD27 IgD	CD16 CD56	CD14 CD45
BRUTON'S TYROSINE KINASE	Evaluates the presence of Bruton's Tyrosine Kinase (BTK) expression in monocytes and B cells.	BTK	88184-TC 88185 x8-TC 86355-TC 86359-TC 88187	CD3 CD4 CD8	CD19 BTK	CD16 CD56	CD14 CD45
COMMON VARIABLE IMMUNODEFICIENCY	Evaluates lymphocyte subpopulations, T cell memory and activation status, and B cells naïve/memory/switched memory status based on IgD and CD27 expression.	CVID	88184-TC 88185 x13-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 CD45RA CD45RO	CD19 CD21 CD27 IgD	CD16 CD56	CD14 CD45 HLA-DR
CYTOKINE-IBD (PBMC ISOLATION)	Determines the ability of IL-10 to inhibit LPS induced IL-6 cytokine production, a STAT3 dependent event. Useful to test IL10 receptor function and STAT3 function (HyperIgE)	CYTIBD	86353 x4-TC 83520-TC				IL-6
FUNCTIONAL ASPLENIA/HOWELL-JOLLY BOBY DETECTION	Screening test to aid in the diagnosis of splenic dysfunction by analyzing mature erythrocytes to detect the presence of micronuclei (Howell-Jolly Bodies) by staining with propidium iodide	FAHJB	88184-TC 88185x2-TC 88187				CD45 CD61 CD71
CYTOTOXICITY/APOPTOSIS (PBMC ISOLATION)	Evaluates NK cell cytotoxicity by measuring apoptosis of tumor cells. Abnormal in hemophagocytic lymphohistiocytosis.	CYTAPO	86352-TC 88184-TC 88185x3-TC 88187	CD3		CD56 CD16 CD107a	
HYPER IGM	Evaluates the expression of CD40L on activated CD4 cells, and CD40 expression. Can be used to evaluate carrier status.	HIGM	88184-TC 88185 x10-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 CD154 CD40-Fc	CD19 CD40	CD16 CD56	CD14 CD15
MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASES	Evaluates expression of INF gamma and IL12 receptors (CD119 and CD212), as well as responsiveness of cells to IFN gamma and IL12 by measuring phospho-STAT1 and phospho-STAT4, respectively. We also measure LPS/IFNγ induced IL12 production.	MSMD	86353-TC 88184-TC 88185 x15-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 CD45RA CD45RO	CD19	CD16 CD56 CD212	CD14 CD45 HLA-DR CD119 IL-12 pSTAT1 pSTAT4

NEUTROPHIL PHENOTYPE and FUNCTION (formerly NEUTROPHIL OXIDATIVE BURST, NEUOXB)	Evaluates the ability of neutrophils to generate an oxidative burst using dihydrorhodamine (DHR). Patients with chronic granulomatous disease (CGD) are defective in this response. We also evaluate PMN surface marker expression for CD15 and CD18.	NPF	88184-TC 88185x2-TC 88187				CD15 CD18 CD45
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PERFORIN GRANZYME	Determines the presence of intracellular perforin and granzyme B within natural killer cells.	PERGRA	88184-TC 88185 x9-TC 86355-TC 86359-TC 88188	<u>T-Subsets</u> CD3 CD4 CD8	<u>B-Subsets</u> CD19	<u>NK-Subsets</u> CD56 CD16	<u>Other</u> CD14 CD45 Granzyme B Perforin
PRIMARY IMMUNODEFICIENCY 1	Evaluates lymphocyte subpopulations(T, B, NK)	PID1	88184-TC 88185 x7-TC 86355-TC 86359-TC 88187	CD3 CD4 CD8	CD19	CD16 CD56	CD14 CD45
PRIMARY IMMUNODEFICIENCY 2	Evaluates lymphocyte subpopulations(T, B, NK). Also evaluates memory status of CD4 lymphocytes, activation status of CD4 and CD8 cells.	PID2	88184-TC 88185 x10-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 CD45RA CD45RO	CD19	CD16 CD56	CD14 CD45 HLA-DR
T CELL INTERLEUKIN PROLIFERATION (PBMC ISOLATION)	Evaluates the proliferative capacity of T cells in response to CD3 and CD3/28 activation, IL2, IL7 and IL15.	TINTL	86353-TC 88187	CD4 CD8			
T CELL MITOGEN PROLIFERATION (PBMC ISOLATION)	Evaluates the proliferative capacity of lymphocytes to PHA, ConA, and CD3 with or without IL2.	TMITO	86353-TC 88187	CD4 CD8			
T HELPER IL17 (PBMC ISOLATION)	Evaluates IL-17 production in CD4+ T helper cells. Deficient expression is associated with hyper IgE syndrome (HIES)/Job's Syndrome NOTE: <u>NOT</u> to be collected on patients < 1 year of age	THIL17	86353-TC 88184-TC 88185 x9-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8	CD19	CD16 CD56	CD14 CD45 IFN-γ IL-17
TOLL-LIKE RECEPTOR/XIAP	Evaluates ability of monocytes to respond to LPS and MDP. Defective responsive can be seen in IRAK4/MyD88 and XIAP deficiency, respectively.	TLREC/XIAP	83520-TC 86352-TC 88184-TC 88187				CD14 TNFα
STAT GAIN-OF-FUNCTION	Screening assay to determine if mutations in the STAT1 gene are potentially functional.	SGOF	88184-TC 88185 x13-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 CD45RA CD45RO	CD19	CD16 CD56	CD14 CD45 HLA-DR pSTAT1

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T REGULATORY –FOXP3 (PBMC ISOLATION)	Evaluates lymphocyte subpopulations and the presence of Foxp3+ T regulatory cells.	TREG	88184-TC 88185 x12-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 CD45RA CD45RO CD25 FoxP3	CD19	CD16 CD56	CD14 CD45 HLA-DR
X-LINKED LYPHOPROLIFERATIVE SYNDROME (PBMC ISOLATION)	Determines the presence of SAP and XIAP expression in lymphocytes to evaluate for XLP type 1 and type 2, respectively.	XLP	88184-TC 88185 x12-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 CD45RA CD45RO	CD19	CD16 CD56	CD14 CD45 HLA-DR SAP XIAP
LIPOPOLYSACCHARIDE-RESPONSIVE BEIGE-LIKE ANCHOR PROTEIN	Determines LRBA expression by lymphocytes. In addition, we evaluate FOXP3 expression as well as CTLA4.	LRBA	88184-TC 88185 x12-TC 86355-TC 86359-TC 88188	CD3 CD4 CD8 CD45RA CD45RO CD25 FoxP3	CD19	CD16 CD56	CD14 CD45 HLA-DR CTLA4 LRBA
NADPH OXIDASE COMPLEX	Evaluates NADPH Oxidase protein subunit expression in neutrophils and monocytes	PHOX	88184-TC 88185x2-TC 88187				CD45 CD14 p22 ^{PHOX} p40 ^{PHOX} p47 ^{PHOX} p67 ^{PHOX} p91 ^{PHOX}