

**CLINICAL IMMUNODIAGNOSTIC AND RESEARCH LAB**

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Children's Hospital  
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Children's Specialty Group™



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>   |
| <b>ABSOLUTE AT4</b>                                       | To monitor patient's individual CD4 and CD8 T cell populations. Panel is routinely used in determining therapeutic considerations for HIV+ patients.   | <b>AT4</b>    | 88184-TC<br>88185 x4-TC<br>86359-TC<br>88187                          | CD3<br>CD4<br>CD8                           |                             |                        | CD14<br>CD45   |
| <b>AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME</b>            | Evaluates the presence of TCR alpha/beta positive –CD4 and CD8 double negative T cells, referred to as TCRab-DNTCs.  | <b>AILYMP</b> | 88184-TC<br>88185 x13-TC<br>86355-TC<br>86359-TC<br>88188             | CD3<br>CD4<br>CD8<br>B220<br>TCRab<br>TCRgd | CD19<br>CD21<br>CD27<br>IgD | CD16<br>CD56           | CD14<br>CD45   |
| <b>BRUTON'S TYROSINE KINASE</b>                           | Evaluates the presence of Bruton's Tyrosine Kinase (BTK) expression in monocytes and B cells.  | <b>BTK</b>    | 88184-TC<br>88185 x8-TC<br>86355-TC<br>86359-TC<br>88187              | CD3<br>CD4<br>CD8                           | CD19<br>BTK                 | CD16<br>CD56           | CD14<br>CD45   |
| <b>COMMON VARIABLE IMMUNODEFICIENCY</b>                   | Evaluates lymphocyte subpopulations, T cell memory and activation status, and B cells naïve/memory/switched memory status based on IgD and CD27 expression.  | <b>CVID</b>   | 88184-TC<br>88185 x13-TC<br>86355-TC<br>86359-TC<br>88188             | CD3<br>CD4<br>CD8<br>CD45RA<br>CD45RO       | CD19<br>CD21<br>CD27<br>IgD | CD16<br>CD56           | CD14<br>CD45<br>HLA-DR                                       |
| <b>CYTOKINE-IBD (PBMC ISOLATION)</b>                      | 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)  | <b>CYTIBD</b> | 86353 x4-TC<br>83520-TC   |   |                             |                        | IL-6   |
| <b>FUNCTIONAL ASPLENIA/HOWELL-JOLLY BOBY DETECTION</b>    | 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   | <b>FAHJB</b>  | 88184-TC<br>88185x2-TC<br>88187                                       |   |                             |                        | CD45<br>CD61<br>CD71   |
| <b>CYTOTOXICITY/APOPTOSIS (PBMC ISOLATION)</b>            | Evaluates NK cell cytotoxicity by measuring apoptosis of tumor cells. Abnormal in hemophagocytic lymphohistiocytosis.  | <b>CYTAPO</b> | 86352-TC<br>88184-TC<br>88185x3-TC<br>88187                           | CD3   |                             | CD56<br>CD16<br>CD107a |  |
| <b>HYPER IGM</b>  | Evaluates the expression of CD40L on activated CD4 cells, and CD40 expression. Can be used to evaluate carrier status.   | <b>HIGM</b>   | 88184-TC<br>88185 x10-TC<br>86355-TC<br>86359-TC<br>88188             | CD3<br>CD4<br>CD8<br>CD154<br>CD40-Fc       | CD19<br>CD40                | CD16<br>CD56           | CD14<br>CD15   |
| <b>MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASES</b> | 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. | <b>MSMD</b>   | 86353-TC<br>88184-TC<br>88185 x15-TC<br>86355-TC<br>86359-TC<br>88188 | CD3<br>CD4<br>CD8<br>CD45RA<br>CD45RO       | CD19                        | CD16<br>CD56<br>CD212  | CD14<br>CD45<br>HLA-DR<br>CD119<br>IL-12<br>pSTAT1<br>pSTAT4 |

|   |   |            |                                 |  |  |  |                      |
|---|---|------------|---------------------------------|--|--|--|----------------------|
| <b>NEUTROPHIL PHENNOTYPE and FUNCTION (formerly NEUTROPHIL OXIDATIVE BURST, NEUOXB)</b> | 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. | <b>NPF</b> | 88184-TC<br>88185x2-TC<br>88187 |  |  |  | CD15<br>CD18<br>CD45 |
|---|---|------------|---------------------------------|--|--|--|----------------------|

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|--|---|------------------------|--|--|---|--|---|
| <b>PERFORIN GRANZYME</b>                                 | Determines the presence of intracellular perforin and granzyme B within natural killer cells.   | <b>PERGRA</b>          | 88184-TC<br>88185 x9-TC<br>86355-TC<br>86359-TC<br>88188             | <u><b>T-</b></u><br><u><b>Subsets</b></u><br>CD3<br>CD4<br>CD8 | <u><b>B-</b></u><br><u><b>Subsets</b></u><br>CD19 | <u><b>NK-</b></u><br><u><b>Subsets</b></u><br>CD56<br>CD16 | <u><b>Other</b></u><br>CD14<br>CD45<br>Granzyme B<br>Perforin |
| <b>PRIMARY IMMUNODEFICIENCY 1</b>                        | Evaluates lymphocyte subpopulations(T, B, NK)   | <b>PID1</b>            | 88184-TC<br>88185 x7-TC<br>86355-TC<br>86359-TC<br>88187             | CD3<br>CD4<br>CD8  | CD19  | CD16<br>CD56   | CD14<br>CD45  |
| <b>PRIMARY IMMUNODEFICIENCY 2</b>                        | Evaluates lymphocyte subpopulations(T, B, NK). Also evaluates memory status of CD4 lymphocytes, activation status of CD4 and CD8 cells.   | <b>PID2</b>            | 88184-TC<br>88185 x10-TC<br>86355-TC<br>86359-TC<br>88188            | CD3<br>CD4<br>CD8<br>CD45RA<br>CD45RO                          | CD19  | CD16<br>CD56   | CD14<br>CD45<br>HLA-DR  |
| <b>T CELL INTERLEUKIN PROLIFERATION (PBMC ISOLATION)</b> | Evaluates the proliferative capacity of T cells in response to CD3 and CD3/28 activation, IL2, IL7 and IL15.  | <b>TINTL</b>           | 86353-TC<br>88187  | CD4<br>CD8   |   |  |   |
| <b>T CELL MITOGEN PROLIFERATION (PBMC ISOLATION)</b>     | Evaluates the proliferative capacity of lymphocytes to PHA, ConA, and CD3 with or without IL2.  | <b>TMITO</b>           | 86353-TC<br>88187  | CD4<br>CD8   |   |  |   |
| <b>T HELPER IL17 (PBMC ISOLATION)</b>                    | Evaluates IL-17 production in CD4+ T helper cells. Deficient expression is associated with hyper IgE syndrome (HIES)/Job's Syndrome<br><br><b>NOTE: <u>NOT</u> to be collected on patients &lt; 1 year of age</b> | <b>THIL17</b>          | 86353-TC<br>88184-TC<br>88185 x9-TC<br>86355-TC<br>86359-TC<br>88188 | CD3<br>CD4<br>CD8  | CD19  | CD16<br>CD56   | CD14<br>CD45<br>IFN-γ<br>IL-17                                |
| <b>TOLL-LIKE RECEPTOR/XIAP</b>                           | Evaluates ability of monocytes to respond to LPS and MDP. Defective responsive can be seen in IRAK4/MyD88 and XIAP deficiency, respectively.  | <b>TLREC/<br/>XIAP</b> | 83520-TC<br>86352-TC<br>88184-TC<br>88187                            |  |   |  | CD14<br>TNFα  |
| <b>STAT GAIN-OF-FUNCTION</b>                             | Screening assay to determine if mutations in the STAT1 gene are potentially functional.   | <b>SGOF</b>            | 88184-TC<br>88185 x13-TC<br>86355-TC<br>86359-TC<br>88188            | CD3<br>CD4<br>CD8<br>CD45RA<br>CD45RO                          | CD19  | CD16<br>CD56   | CD14<br>CD45<br>HLA-DR<br>pSTAT1<br>CD126<br>CD130            |

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|--|--|-------------|---|---|---|--|---|
| <b>T REGULATORY –FOXP3 (PBMC ISOLATION)</b>                    | Evaluates lymphocyte subpopulations and the presence of Foxp3+ T regulatory cells.                                     | <b>TREG</b> | 88184-TC<br>88185 x12-TC<br>86355-TC<br>86359-TC<br>88188 | <u><b>T-</b></u><br><u><b>Subsets</b></u><br>CD3<br>CD4<br>CD8<br>CD45RA<br>CD45RO<br>CD25<br>FoxP3 | <u><b>B-</b></u><br><u><b>Subsets</b></u><br>CD19 | <u><b>Nk-</b></u><br><u><b>Subsets</b></u><br>CD16<br>CD56 | <u><b>Other</b></u><br>CD14<br>CD45<br>HLA-DR |
| <b>X-LINKED LYPHOPROLIFERATIVE SYNDROME (PBMC ISOLATION)</b>   | Determines the presence of SAP and XIAP expression in lymphocytes to evaluate for XLP type 1 and type 2, respectively. | <b>XLP</b>  | 88184-TC<br>88185 x12-TC<br>86355-TC<br>86359-TC<br>88188 | CD3<br>CD4<br>CD8<br>CD45RA<br>CD45RO   | CD19  | CD16<br>CD56   | CD14<br>CD45<br>HLA-DR<br>SAP<br>XIAP         |
| <b>LIPOPOLYSACCHARIDE-RESPONSIVE BEIGE-LIKE ANCHOR PROTEIN</b> | Determines LRBA expression by lymphocytes. In addition, we evaluate FOXP3 expression as well as CTLA4.                 | <b>LRBA</b> | 88184-TC<br>88185 x12-TC<br>86355-TC<br>86359-TC<br>88188 | CD3<br>CD4<br>CD8<br>CD45RA<br>CD45RO<br>CD25<br>FoxP3  | CD19  | CD16<br>CD56   | CD14<br>CD45<br>HLA-DR<br>CTLA4<br>LRBA       |