ARUP LABORATORIES

800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	5/9/1958
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Lymphocyte Subset Panel 7 - Congenital Immunodeficiencies
ARUP test code 0095899

Test	Result	Flag	Reference
% CD2	84 %		(Ref Interval: 78-92)
Absolute CD2	218 cells/uL	L	(Ref Interval: 680-2400)
% CD3	16 %	L	(Ref Interval: 62-89)
Absolute CD3	43 cells/uL	L	(Ref Interval: 660-2200)
% CD4	9 %	L	(Ref Interval: 35-68)
Absolute CD4	23 cells/uL	L	(Ref Interval: 490-1600)
% CD8	7 %	L	(Ref Interval: 10-46)
Absolute CD8	20 cells/uL	L	(Ref Interval: 150-1050)
CD4:CD8 Ratio	1.29 ratio		(Ref Interval: 0.80-6.17)
% Natural Killer Cells	81 %	H	(Ref Interval: 5-28)
Absolute Natural Killer Cells	213 cells/uL		(Ref Interval: 74-620)
% CD19	0 %	L	(Ref Interval: 5-21)

H=High, L=Low, *=Abnormal, C=Critical

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Patient: Patient, Example
ARUP Accession: 24-064-145092
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Absolute CD19	**1 cells/uL**	**L**	**(Ref Interval: 74-510)**
% CD45RA	21 %		(Ref Interval: 19-62)
Absolute CD45RA	**6 cells/uL**	**L**	**(Ref Interval: 260-1000)**
% CD45RO	79 %		(Ref Interval: 38-81)
Absolute CD45RO	**23 cells/uL**	**L**	**(Ref Interval: 490-1200)**
% HLA-DR	**4 %**	**L**	**(Ref Interval: 7-20)**
Absolute HLA-DR	**10 cells/uL**	**L**	**(Ref Interval: 98-430)**

Lymphocyte Subset Panel 7 Information	See Note

INTERPRETIVE INFORMATION: Lymphocyte Subset 7,
Congenital Immunodeficiencies
This profile screens for inherited immunodeficiencies. The CD4 cells are Helper T-cells expressing both CD3 and CD4. The CD8 cells are Cytotoxic T-cells expressing both CD3 and CD8. The B-cells express CD19 but not CD3. The NK-cells express either CD16 or CD56 (or both) but not CD3. CD3, CD4, CD8, CD19 and NK-cell percentages are reported as a percent of total lymphocytes. The CD45RA cells express both CD4 and "naive" CD45RA antigens while CD45RO cells express both CD4 and CD45RO "memory" antigens. CD45RA and CD45RO percentages are reported as a percent of total CD4 cells. Primary immune deficiencies that show phenotypic abnormalities include X-linked hypogammaglobulinemia, DiGeorge syndrome, bare lymphocyte syndrome, and severe combined immunodeficiency disease (SCID).

X-linked hypogammaglobulinemia (X-linked agammaglobulinemia, Bruton's agammaglobulinemia) is caused by defective B-cell maturation secondary to mutations in the BTK (Bruton/B-cell tyrosine kinase) gene. T-cells (CD2, CD3) are normal or increased in number, and the CD4:CD8 ratio is normal or decreased. Most of the CD4 cells express the CD45RA antigen characteristic of naive rather than memory cells. B-cells (CD19, HLA-DR) are severely decreased or absent in the peripheral blood.

X-linked hypogammaglobulinemia can be distinguished from transient hypogammaglobulinemia of infancy by the absence of B-cells. Transient hypogammaglobulinemia of infancy results from delayed capacity for immunoglobulin synthesis and spontaneously resolves with age.

Thymic aplasia (congenital thymic aplasia, DiGeorge syndrome) results in impaired T-cell maturation and function. B-cells (CD19, HLA-DR) and NK-cells (CD16/CD56) are normal but T-cells (CD2, CD3) are usually decreased with an elevated CD4:CD8 ratio.

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Patient Report | **FINAL**

The clinical course is variable, ranging from "partial DiGeorge syndrome" to cases that resemble SCID.

SCID has multiple genetic causes, including mutations in the gamma chain of the interleukin 2 receptor and the purine degradation enzymes, adenosine deaminase, and nucleoside phosphorylase. In adenosine deaminase deficiency, both B-cells (CD19, HLA-DR) and T-cells (CD2, CD3) are decreased in the peripheral blood. In other forms of SCID, the lymphopenia is not as severe, but the lymphocyte count is usually less than 1,000/uL even though B-cells (CD19, HLA-DR) may be normal or increased. In contrast to thymic aplasia, any T-cells present may have an immature phenotype.

Major histocompatibility complex class II deficiency, bare lymphocyte syndrome, is caused by defective transcription of HLA class II genes; B-cells (CD19) and T-cells (CD2, CD3) are present in normal numbers, but HLA-DR is absent. The CD4+ cells are usually CD45RA+.

Common variable immunodeficiency (CVID) describes a heterogeneous group of disorders with defective antibody formation. B-cells (CD19, HLA-DR) and T-cells (CD2, CD3) are usually normal in number, although B-cells may be decreased when CVID occurs concurrently with systemic lupus erythematosus. The CD4:CD8 ratio may be normal or decreased.

Wiskott-Aldrich syndrome includes immunodeficiency with thrombocytopenia and eczema. Lymphopenia is usually present with a progressive decline in T-cells numbers. The CD4:CD8 ratio is normal. The gene is X-linked and encodes the Wiskott-Aldrich syndrome protein.

Immunophenotyping is generally not useful in characterizing selective IgA deficiency, IgG subclass deficiencies, the hyper IgM syndrome, or hyperimmunoglobulin E syndrome (Job syndrome).

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
% CD2	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute CD2	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
% CD3	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute CD3	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
% CD4	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute CD4	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
% CD8	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute CD8	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
CD4:CD8 Ratio	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
% Natural Killer Cells	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute Natural Killer Cells	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
% CD19	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute CD19	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
% CD45RA	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute CD45RA	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
% CD45RO	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute CD45RO	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
% HLA-DR	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Absolute HLA-DR	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Lymphocyte Subset Panel 7 Information	24-064-145092	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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