Multicolor Flow Cytometry for the Diagnosis of Primary Immunodeficiency Diseases

Takehiro Takashima1,2, Miko Okamura1, Tzu-wen Yeh1, Tsubasa Okano1, Motoi Yamashita1, Keisuke Tanaka1, Akihiro Hoshino1,3, Noriko Mitsui1, Masatoshi Takagi1, Eiichi Ishii2, Kohtsuke Imai4, Hirokazu Kanegane1, Tomohiro Morio1

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Abstract
Purpose Primary immunodeficiency diseases (PIDDs) are rare inherited diseases that impair the human immune system. We established a multicolor flow cytometric assay to comprehensively evaluate the immune status and immunological characteristics of patients with PIDDs.
Methods Fifty-nine normal controls and 75 patients with PIDDs, including X-linked severe combined immunodeficiency (X-SCID), X-linked agammaglobulinemia (XLA), X-linked hyper IgM syndrome (X-HIGM), ataxia telangiectasia (AT), Wiskott-Aldrich syndrome (WAS), hyper IgE syndrome (HIES), and chronic mucocutaneous candidiasis disease (CMCD), were enrolled in this study. Immunophenotypes were evaluated by multicolor flow cytometry using seven different panels that allowed the detection of major leukocyte populations in peripheral blood.
Results Multicolor flow cytometry revealed distinct leukocyte populations and immunological features of patients with X-SCID, XLA, X-HIGM, AT, WAS, HIES, and CMCD.
Conclusions Immunophenotyping by multicolor flow cytometry is useful to evaluate immune status and contributes to the diagnosis and management of patients with PIDDs.

Keywords Ataxia telangiectasia · Chronic mucocutaneous candidiasis disease · Flow cytometry · Hyper IgE syndrome · Primary immunodeficiency disease

Abbreviations
AT ataxia telangiectasia
BCR B cell receptor
CMCD chronic mucocutaneous candidiasis disease
DC dendritic cell
DNT double negative T
FSC forward scatter
GOF gain of function
HIES hyper IgE syndrome
mNK T invariant NKT
KREC kappa-deleting recombination excision circles
LOF loss of function
mDCs myeloid dendritic cells
PBMCs peripheral blood mononuclear cells
PBS phosphate-buffered saline
pDCs plasmacytoid dendritic cells
PIDDs primary immunodeficiency diseases
RTEs recent thymic emigrants
SCID severe combined immunodeficiency

Takehiro Takashima and Miko Okamura equally contributed to this study.

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1 Department of Pediatrics and Developmental Biology, Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University (TMDU), 1-5-45 Yushima, Bunkyo-ku, Tokyo 113-8519, Japan
2 Department of Pediatrics, Ehime University Graduate School of Medicine, Toon, Ehime, Japan
3 Department of Lifetime Clinical Immunology, Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University (TMDU), Tokyo, Japan
4 Department of Community Pediatrics, Perinatal and Maternal Medicine, Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University (TMDU), 1-5-45 Yushima, Bunkyo-ku, Tokyo 113-8519, Japan

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