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ORIGINAL ARTICLE

Multicolor Flow Cytometry for the Diagnosis of Primary Immunodeficiency Diseases

Takehiro Takashima[1,2] · Miko Okamura[1] · Tzu-wen Yeh[1] · Tsubasa Okano[1] · Motoi Yamashita[1] · Keisuke Tanaka[1] · Akihiro Hoshino[1,3] · Noriko Mitsuiki[1] · Masatoshi Takagi[4] · Eiichi Ishii[2] · Kohsuke Imai[4] · Hirokazu Kanegane[1] · Tomohiro Morio[1]

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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. Immunophenotyes 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

Takehiro Takashima and Miko Okamura equally contributed to this study.

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✉ Kohsuke Imai
kimai.ped@tmd.ac.jp

✉ Hirokazu Kanegane
hkanegane.ped@tmd.ac.jp

[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

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
iNKT	invariant NKT
KRECs	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

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