

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Ma CS, Avery DT, Chan A, Batten M, Bustamante J, Boisson-Dupuis S, Arkwright PD, Minegishi Y, Nonoyama S, French MA, Choo S, Peake J, Wong M, Cook MC, Fulcher DA, Casanova JL, Deenick EK, Tangye SG.	Functional STAT3 deficiency compromises the generation of human T follicular helper cells.	Blood.	26;119	3997-4008	2012
Honda F, Kano H, Kanegane H, Nonoyama S, Kim ES, Lee SK, Takagi M, Mizutani S, Morio T.	The kinase Btk negatively regulates the production of reactive oxygen species and stimulation-induced apoptosis in human neutrophils.	Nat Immunol.	26;13	369-378	2012
Ishida H, Imai K, Homma K, Tamura S, Imamura T, Itoh M, Nonoyama S.	GATA-2 anomaly and clinical phenotype of a sporadic case of lymphedema, dendritic cell, monocyte, B- and NK-cell (DCML) deficiency, and myelodysplasia.	Eur J Pediatr.	171	1273-1276	2012
Suri D, Singh S, Rawat A, Gupta A, Kamae C, Honma K, Nakagawa N, Imai K, Nonoyama S, Oshima K, Mitsuiki N, Ohara O, Bilhou-Nabera C, Proust A, Ahluwalia J, Dogra S, Saikia B, Walker Minz R, Sehgal S.	Clinical profile and genetic basis of Wiskott-Aldrich syndrome at Chandigarh, north India.	Asian Pac J Allergy Immunol	30	71-78	2012
Nozaki T, Takada H, Ishimura M, Ihara K, Imai K, Morio T, Kobayashi M, Nonoyama S, Hara T.	Endocrine complications in primary immunodeficiency diseases in Japan.	Clin Endocrinol.	77	628-634	2012
Kawai T, Saito M, Nishikomori R, Yasumi T, Izawa K, Murakami T, Okamoto S, Mori Y, Nakagawa N, Imai K, Nonoyama S, Wada T, Yachie A, Ohmori K, Nakahata T, Heike T.	Multiple reversions of an IL2RG mutation restore T cell function in an X-linked severe combined immunodeficiency patient.	J Clin Immunol.	32	690-697	2012
Nakaoka H, Kanegane H, Taneichi H, Miya K, Yang X, Nomura K, Takezaki S, Yamada M, Ohara O, Kamae C, Imai K, Nonoyama S, Wada T, Yachie A, Hershfield MS, Ariga T, Miyawaki T.	Delayed onset adenosine deaminase deficiency associated with acute disseminated encephalomyelitis.	Int J Hematol.	95	692-696	2012
Oshima K, Nagase T, Imai K, Nonoyama S, Obara M, Mizukami T, Nunoi H, Kanegane H, Kuribayashi F, Amemiya S, Ohara O.	A dual reporter splicing assay using HaloTag-containing proteins.	Curr Chem Genomics.	in press		2012
Kakiuchi S, Nonoyama S, Wakamatsu H, Kogawa K, Wang L, Kinoshita-Yamaguchi H, Takayama-Ito M, Lim CK, Inoue N, Mizuguchi M, Igarashi T, Saijo M.	Neonatal herpes encephalitis caused by a virologically confirmed acyclovir resistant herpes simplex virus type 1.	J. Clin. Microbiol.	in press		2012
Yang X, Kanegane H, Nishida N, Imamura T, Hamamoto K, Miyashita R, Imai K, Nonoyama S, Sanayama K, Yamaide A, Kato F, Nagai K, Ishii E, Zelm M, Latour S, Zhao X, Miyawaki T.	Clinical and genetic characteristics of XIAP deficiency in Japan.	J Clin Immunol.	32	411-420	2012
Kobayashi D, Kogawa K, Imai K, Tanaka T, Sada A, Nonoyama S.	Hyper-eosinophilia in granular acute B-cell lymphoblastic leukemia with myeloid antigen expression.	Pediatr Int.	54	543-546	2012
Kakiuchi S, Nonoyama S, Wakamatsu H, Kogawa K, Wang L, Kinoshita-Yamaguchi H, Takayama-Ito M, Lim CK, Inoue N, Mizuguchi M, Igarashi T, Saijo M.	Neonatal herpes encephalitis caused by a virologically confirmed acyclovir resistant herpes simplex virus type 1.	J Clin Microbiol.	in press		2012
Wada T, Muraoka M, Toma T, Imai T, Shigemura T, Agematsu K, Haraguchi K, Moriuchi H, Oh-Ishi T, Kitoh T, Ohara O, Morio T, Yachie A.	Rapid Detection of Intracellular p47phox and p67phox by Flow Cytometry; Useful Screening Tests for Chronic Granulomatous Disease.	J Clin Immunol.	in press		2012

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Hori T, Ohnishi H, Teramoto T, Tsubouchi K, Naiki T, Hirose Y, Ohara O, Seishima M, Kaneko H, Fukao T, Kondo N.	Autosomal-Dominant Chronic Mucocutaneous Candidiasis with STAT1-Mutation can be Complicated with Chronic Active Hepatitis and Hypothyroidism.	J Clin Immunol.	32(6)	1213-1220	2012
Takezaki S, Yamada M, Kato M, Park MJ, Maruyama K, Yamazaki Y, Chida N, Ohara O, Kobayashi I, Ariga T.	Chronic mucocutaneous candidiasis caused by a gain-of-function mutation in the STAT1 DNA-binding domain.	J Immunol.	189(3)	1521-1526	2012
Suri D, Singh S, Rawat A, Gupta A, Kamae C, Honma K, Nakagawa N, Imai K, Nonoyama S, Oshima K, Mitsui N, Ohara O, Bilhou-Nabera C, Proust A, Ahluwalia J, Dogra S, Saikia B, Minz RW, Sehgal S.	Clinical profile and genetic basis of Wiskott-Aldrich syndrome at Chandigarh, North India.	Asian Pac J Allergy Immunol.	30(1)	71-78	2012
Mohammadzadeh I, Yeganeh M, Aghamohammadi A, Parvaneh N, Behniafard N, Abolhassani H, Tabassomi F, Hemmat M, Kanegane H, Miyawaki T, Ohara O, Rezaei N.	Severe primary antibody deficiency due to a novel mutation of mu heavy chain.	J Investig Allergol Clin Immunol.	22(1)	78-79	2012
Hiramoto T, Ebihara Y, Mizoguchi Y, Nakamura K, Yamaguchi K, Ueno K, Mochizuki S, Yamamoto S, Nagasaki M, Furukawa Y, Tani K, Nakauchi H, Kobayashi M, Tsuji K	Wnt3a stimulates maturation of impaired neutrophils developed from severe congenital neutropenia-derived pluripotent stem cells with heterozygous ELANE mutation.	Proc Natl Acad Sci USA	in press		2013
Kawai T, Nishikomori R, Izawa K, Murata Y, Tanaka N, Sakai H, Saito M, Yasumi T, Takaoka Y, Nakahata T, Mizukami T, Nunoi H, Kiyohara Y, Yoden A, Murata T, Sasaki S, Ito E, Akutagawa H, Kawai T, Imai C, Okada S, Kobayashi M, Heike T.	Frequent somatic mosaicism of NEMO in T cells of patients with X-linked anhidrotic ectodermal dysplasia with immunodeficiency.	Blood	119(23)	5458-66	2012
Kobayashi Y, Matsui H, Kanai A, Tsumura M, Okada S, Miki M, Nakamura K, Kunishima S, Inaba T, Kobayashi M.	Identification of the integrin $\beta 3$ L718P mutation in a pedigree with autosomal dominant thrombocytopenia with anisocytosis.	Br J Haematol	in press		2012
Tsumura M, Okada S, Sakai H, Yasunaga S, Ohtsubo M, Murata T, Obata H, Yasumi T, Kong XF, Abhyankar A, Heike T, Nakahata T, Nishikomori R, Al-Muhsen S, Boisson-Dupuis S, Casanova JL, Alzahrani M, Shehri MA, Elghazali G, Takihara Y, Kobayashi M.	Dominant-negative STAT1 SH2 domain mutations in unrelated patients with Mendelian susceptibility to mycobacterial disease.	Human Mutation	33(9)	1377-87	2012
Zhang X, Inukai T, Hirose K, Akahane K, Kuroda I, Honna-Oshiro H, Kagami K, Goi K, Nakamura K, Kobayashi M, Endo M, Yagita H, Kurosawa H, Thomas Look A, Honda H, Inaba T, Nakazawa S, Sugita K.	Oncogenic fusion E2A-HLF sensitizes t(17;19)-positive acute lymphoblastic leukemia to TRAIL-mediated apoptosis by upregulating the expression of death receptors.	Leukemia.	26(12)	2483-93	2012
Ohno N, Kobayashi M, Hayakawa S, Utsunomiya A, Karakawa S.	Transient pseudothrombocytopenia in a neonate: Transmission of a maternal EDTA-dependent anticoagulant.	Platelets	23(5)	399-400	2012
Kajiume T, Sera Y, Kawahara Y, Matsumoto M, Fukazawa T, Imura T, Yuge L, Kobayashi M.	Regulation of hematopoietic stem cells using protein transduction domain-fused Polycomb.	Exp Hematol.	40(9)	751-760	2012
Ichikawa M, Suzuki D, Inamoto J, Ohshima J, Cho Y, Saitoh S, Kaneda M, Iguchi A, Ariga T.	Successful alternative treatment containing vindesine for acute lymphoblastic leukemia with Charcot-Marie-Tooth disease.	J Pediatr Hematol Oncol.	34	239-41	2012
Sarashina T, Yoshida M, Iguchi A, Okubo H, Toriumi N, Suzuki D, Sano H, Kobayashi R.	Risk factor analysis of bloodstream infection in pediatric patients after hematopoietic stem cell transplantation.	J Pediatr Hematol Oncol.	35	76-80	2013

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Sato T, Okumura F, Iguchi A, Ariga T, Hatakeyama S.	TRIM32 promotes retinoic acid receptor $\alpha$ -mediated differentiation in human promyelogenous leukemic cell line HL60.	Biochem Biophys Res Commun.	417	594-600	2012
Takezaki S, Okura Y, Ichikawa M, Suzuki D, Ohshima J, Kaneda M, Cho Y, Yamada M, Kawamura N, Iguchi A, Kobayashi I, Ariga T.	Development of germinoma during the treatment of systemic-onset juvenile idiopathic arthritis with infliximab.	Mod Rheumatol.	22	621-4	2012
Okura Y, Nawate M, Takahashi Y, Kobayashi I, Yamada M, Ariga T.	Rheumatoid factor-positive synovitis in a patient with C3 deficiency.	Scand J Rheumatol.	41	405-6	2012
Cho Y, Iizuka S, Hatae Y, Kobayashi K, Hattori Y, Yamashiro Y, Ariga T.	A 25-year observation of a Japanese female patient with Hb Nottingham who has two children with the same disorder.	Hemoglobin.	36	446-55	2012
Shibata M, Sato T, Nukiwa R, Ariga T, Hatakeyama S.	TRIM45 negatively regulates NF- $\kappa$ B-mediated transcription and suppresses cell proliferation.	Biochem Biophys Res Commun.	423	104-9	2012
Ohshima J, Haruta M, Fujiwara Y, Watanabe N, Arai Y, Ariga T, Okita H, Koshinaga T, Oue T, Hinotsu S, Nakadate H, Horie H, Fukuzawa M, Kaneko Y.	Methylation of the RASSF1A promoter is predictive of poor outcome among patients with Wilms tumor.	Pediatr Blood Cancer.	59	499-505	2012
Koseki N, Teramoto S, Kaiho M, Gomi-Endo R, Yoshioka M, Takahashi Y, Nakayama T, Sawada H, Konno M, Ushijima H, Kikuta H, Ariga T, Ishiguro N.	Detection of human bocaviruses 1 to 4 from nasopharyngeal swab samples collected from patients with respiratory tract infections.	J Clin Microbiol.	50	2118-21	2012
Ariga T.	Wiskott-Aldrich syndrome: an x-linked primary immunodeficiency disease with unique and characteristic features.	Allergol Int.	61	183-9	2012
Sato T, Okumura F, Ariga T, Hatakeyama S.	TRIM6 interacts with Myc and maintains the pluripotency of mouse embryonic stem cells.	J Cell Sci.	125	1544-55	2012
Yamada M, Okura Y, Suzuki Y, Fukumura S, Miyazaki T, Ikeda H, Takezaki S, Kawamura N, Kobayashi I, Ariga T.	Somatic mosaicism in two unrelated patients with X-linked chronic granulomatous disease characterized by the presence of a small population of normal cells.	Gene.	497	110-5	2012
Morishima T, Nomura A, Saida S, Watanabe K, Yagi H, Matsumoto M, Fujimura Y, Heike T, Nakahata T, Adachi S.	Pediatric idiopathic TTP diagnosed with decreased ADAMTS13 activity.	Pediatr. Int.	54(3)	422-3	2012
Tsuchiya A, Imai M, Kamimura H, Takamura M, Yamagiwa S, Sugiyama T, Nomoto M, Heike T, Nagasawa T, Nakahata T, Aoyagi Y.	Increased susceptibility to severe chronic liver damage in CXCR4 conditional knock-out mice.	Dig. Dis. Sci.	57(11)	2892-2900	2012
Kikuchi A, Hasegawa D, Ohtsuka Y, Hamamoto K, Kojima S, Okamura J, Nakahata T, Manabe A.	Outcome of children with Refractory Anaemia with Excess of Blast (RAEB) and RAEB in Transformation (RAEB-T) in the Japanese MDS99 study.	Brit. J. Haematol.	158(5)	657-661	2012
Nakazawa Y, Saito S, Yanagisawa R, Suzuki T, Toshiro Ito T, Ishida F, Muramatsu H, Matsumoto K, Kato K, Ishida H, Umeda K, Souichi Adachi S, Nakahata T, Koike K.	Recipient seropositivity for adenovirus type 11 is highly predictive of the development of hemorrhagic cystitis after allogeneic hematopoietic stem cell transplantation.	Bone Marrow Transplant.	in press		2012
Saida S, Watanabe K, Sato-Otsubo A, Terui K, Yoshida K, Okuno Y, Toki T, Wang RN, Shiraiishi Y, Miyano S, Kato I, Morishima T, Fujino H, Umeda K, Hiramatsu H, Adachi S, Ito E, Ogawa S, Ito M, Nakahata T, Heike T.	Clonal selection in xenografted TAM recapitulates the evolutionary process of myeloid leukemia in Down syndrome.	Blood	in press		2012

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Kanegane H, Yang X, Zhao M, et al.	Clinical features and outcome of X-linked lymphoproliferative syndrome type 1 (SAP deficiency) in Japan identified by the combination of flow cytometric assay and genetic analysis. Pediatric allergy and immunology.	Pediatr Allergy Immunol.	23(5)	488-93	2012
Marsh R. a. Rao, K, Satwani P, et al.	Allogeneic hematopoietic cell transplantation for XIAP deficiency: an international survey reveals poor outcomes.	Blood	121(6)	877-83	2013
Yoshimi A, Kamachi Y, Imai K, Watanabe N, Nakadate H, Kanazawa T, Ozono S, Kobayashi R, Yoshida M, Kobayashi C, Hama A, Muramatsu H, Sasahara Y, Jakob M, Morio T, Ehl S, Manabe A, Niemeyer C, Kojima S.	Wiskott-Aldrich syndrome presenting with a clinical picture mimicking juvenile myelomonocytic leukaemia.	Pediatric blood & cancer	in press		2012
Shimada A, Takahashi Y, Muramatsu H, Hama A, Ismael O, Narita A, Sakaguchi H, Doisaki S, Nishio N, Tanaka M, Yoshida N, Matsumoto K, Kato K, Watanabe N, Kojima S.	Excellent outcome of allogeneic bone marrow transplantation for Fanconi anemia using fludarabine-based reduced-intensity conditioning regimen.	International journal of hematology	95	675-679	2012
Sakaguchi H, Takahashi Y, Watanabe N, Doisaki S, Muramatsu H, Hama A, Shimada A, Yagasaki H, Kudo K, Kojima S.	Mutational analysis of RNA splicing machinery components in 206 children with myeloid malignancies.	Leukemia research	in press		2012
Doisaki S, Muramatsu H, Shimada A, Takahashi Y, Mori-Ezaki M, Sato M, Kawaguchi H, Kinoshita A, Sotomatsu M, Hayashi Y, Furukawa-Hibi Y, Yamada K, Hoshino H, Kiyoi H, Yoshida N, Sakaguchi H, Narita A, Wang X, Ismael O, Xu Y, Nishio N, Tanaka M, Hama A, Koike K, Kojima S.	Somatic mosaicism for oncogenic NRAS mutations in juvenile myelomonocytic leukemia.	Blood	120	1485-1488	2012
Wang X, Muramatsu H, Sakaguchi H, Xu Y, Narita A, Tsumura Y, Doisaki S, Tanaka M, Ismael O, Shimada A, Hama A, Takahashi Y, Kojima S.	Mutation in the THPO gene is not associated with aplastic anaemia in Japanese children.	British journal of haematology	158	553-555	2012
Sakaguchi H, Takahashi Y, Watanabe N, Doisaki S, Muramatsu H, Hama A, Shimada A, Yagasaki H, Kudo K, Kojima S.	Incidence, clinical features, and risk factors of idiopathic pneumonia syndrome following hematopoietic stem cell transplantation in children.	Pediatric blood & cancer	58	780-784	2012
Saito Y, Aoki Y, Muramatsu H, Makishima H, Maciejewski JP, Imaizumi M, Rikiishi T, Sasahara Y, Kure S, Niihori T, Tsuchiya S, Kojima S, Matsubara Y.	Casitas B-cell lymphoma mutation in childhood T-cell acute lymphoblastic leukemia.	Leukemia research	36	1009-1015	2012
Narita A, Muramatsu H, Takahashi Y, Sakaguchi H, Doisaki S, Nishio N, Hama A, Shimada A, Ito M, Kojima S.	Autoimmune-like hepatitis following unrelated BMT successfully treated with rituximab.	Bone marrow transplantation	47	600-602	2012
Muramatsu H, Makishima H, Maciejewski JP.	Chronic myelomonocytic leukemia and atypical chronic myeloid leukemia: novel pathogenetic lesions.	Seminars in oncology	39	67	2012
Makishima H, Sugimoto Y, Szpurka H, Clemente MJ, Ng KP, Muramatsu H, O'Keefe C, Sauntharajah Y, Maciejewski JP.	CBL mutation-related patterns of phosphorylation and sensitivity to tyrosine kinase inhibitors.	Leukemia	26	1547-1554	2012
Kato K, Otake H, Tagaya M, Takahashi Y, Ito Y, Hama A, Muramatsu H, Kojima S, Naganawa S, Nakashima T.	Progressive hearing loss following acquired cytomegalovirus infection in an immunocompromised child.	American journal of otolaryngology	in press		2012

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Jerez A, Sugimoto Y, Makishima H, Verma A, Jankowska AM, Przychodzen B, Visconte V, Tiu RV, O'Keefe CL, Mohamedali AM, Kulasekararaj AG, Pellagatti A, McGraw K, Muramatsu H, Moliterno AR, Sekeres MA, McDevitt MA, Kojima S, List A, Boulwood J, Mufti GJ, Maciejewski JP.	Loss of heterozygosity in 7q myeloid disorders: clinical associations and genomic pathogenesis.	Blood	119	6109-6117	2012
Ismael O, Shimada A, Hama A, Sakaguchi H, Doisaki S, Muramatsu H, Yoshida N, Ito M, Takahashi Y, Akita N, Sunami S, Ohtsuka Y, Asada Y, Fujisaki H, Kojima S.	Mutations profile of polycythemia vera and essential thrombocythemia among Japanese children.	Pediatric blood & cancer	59	530-535	2012
Ismael O, Shimada A, Hama A, Elshazley M, Muramatsu H, Goto A, Sakaguchi H, Tanaka M, Takahashi Y, Yinyan X, Fukuda M, Miyajima Y, Yamashita Y, Horibe K, Hanada R, Ito M, Kojima S.	De novo childhood myelodysplastic/myeloproliferative disease with unique molecular characteristics.	British journal of haematology	158	129-137	2012
Hama A, Muramatsu H, Makishima H, Sugimoto Y, Szpurka H, Jasek M, O'Keefe C, Takahashi Y, Sakaguchi H, Doisaki S, Shimada A, Watanabe N, Kato K, Kiyoi H, Naoe T, Kojima S, Maciejewski JP.	Molecular lesions in childhood and adult acute megakaryoblastic leukaemia.	British journal of haematology	15	316-325	2012
Fukushima H, Fukushima T, Hiraki A, Suzuki R, Mahmoud SS, Yoshimi A, Nakao T, Kato K, Kobayashi C, Koike K, Fukasawa M, Morishita Y, Doisaki S, Muramatsu H, Sumazaki R.	Central nervous system lesions due to juvenile myelomonocytic leukemia progressed in a boy undergoing first line chemotherapy.	International journal of hematology	95	581-584	2012
Abu Kar S, Jankowska AM, Makishima H, Visconte V, Jerez A, Sugimoto Y, Muramatsu H, Traina F, Afafe M, Guinta K, Tiu RV, Przychodzen B, Sakaguchi H, Kojima S, Sekeres MA, List AF, McDevitt MA, Maciejewski JP.	Spliceosomal gene mutations are frequent events in the diverse mutational spectrum of chronic myelomonocytic leukemia but largely absent in juvenile myelomonocytic leukemia.	Haematologica	in press		2012
Obinata K, Lee T, Niizuma T, Kinoshita K, Shimizu T, Hoshina T, Sasaki Y, Hara T.	Two cases of partial dominant interferon- $\gamma$ receptor 1 deficiency that presented with different clinical courses of bacille Calmette-Guérin multiple osteomyelitis.	J Infect Chemother.	in press		2012
Kusuhara K, Hoshina T, Saito M, Ishimura M, Inoue H, Horiuchi T, Sato T, Hara T.	Successful treatment of a patient with tumor necrosis factor receptor-associated periodic syndrome using a half-dose of etanercept.	Pediatr Int.	54	552-555	2012
Hoshina T, Kusuhara K, Saito M, Mizuno Y, Hara T.	NKRP1A+ $\gamma\delta$ and $\alpha\beta$ T cells are preferentially induced in patients with Salmonella infection.	Hum Immunol.	73	623-628	2012
Yamamura K, Ihara K, Ikeda K, Nagata H, Mizuno Y, Hara T.	Histo-blood group gene polymorphisms as potential genetic modifiers of the development of coronary artery lesions in patients with Kawasaki disease.	Int J Immunogenet.	39(2)	119-125	2012
Onoyama S, Ihara K, Yamaguchi Y, Ikeda K, Yamaguchi K, Yamamura K, Hoshina T, Mizuno Y, Hara T.	Genetic susceptibility to Kawasaki disease: analysis of pattern recognition receptor genes.	Hum Immunol.	73	654-660	2012
Yamamura K, Joo K, Ohga S, Nagata H, Ikeda K, Muneuchi J, Watanabe M, Hara T.	Thrombocytosis in asplenia syndrome with congenital heart disease: a previously unrecognized risk factor for thromboembolism.	Int J Cardiol.	in press		2012
Shiraishi A, Ohga S, Doi T, Ishimura M, Takimoto T, Takada H, Miyamoto T, Abe Y, Hara T.	Treatment choice of immunotherapy or further chemotherapy for Epstein-Barr virus-associated hemophagocytic lymphohistiocytosis.	Pediatr Blood Cancer.	59	265-270	2012

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Imagawa T, Takei S, Umebayashi H, Yamaguchi K, Itoh Y, Kawai T, Iwata N, Murata T, Okafuji I, Miyoshi M, Onoe Y, Kawano Y, Kinjo N, Mori M, Mozaffarian N, Kupper H, Santra S, Patel G, Kawai S, Yokota S.	Efficacy, pharmacokinetics, and safety of adalimumab in pediatric patients with juvenile idiopathic arthritis in Japan.	Clin Rheumatol.	31	1713-1721	2012
Yokota S, Nishikomori R, Takada H, Kikuchi M, Nozawa T, Kanetaka T, Kizawa T, Miyamae T, Mori M, Heike T, Hara T, Imagawa T.	Guidance on the use of canakinumab in patients with cryopyrin-associated periodic syndrome in Japan.	Mod Rheumatol.	in press		2012
Kitajima J, Inoue H, Ohga S, Kinjo T, Ochiai M, Yoshida T, Kusuhara K, Hara T.	Differential transmission and postnatal outcome in triplets with congenital cytomegalovirus infection.	Pediatr Development Patho.	15	151-155	2012
Kamae C, Nakagawa N, Sato H, Honma K, Mitsui N, Ohara O, Kanegane H, Pasic S, Pan-Hammarström Q, van Zelm MC, Morio T, Imai K, Nonoyama S.	Common variable immunodeficiency classification by quantifying T-cell receptor and immunoglobulin κ-deleting recombination excision circles.	J Allergy Clin Immunol.	in press		2012
Kawasaki Y, Toyoda H, Otsuki S, Iwasa T, Iwamoto S, Azuma E, Itoh-Habe N, Wada H, Fujimura Y, Morio T, Imai K, Mitsui N, Ohara O, Komada Y.	A novel Wiskott-Aldrich syndrome protein mutation in an infant with thrombotic thrombocytopenic purpura.	Eur J Haematol.	in press		2012
Kanegane, H, H. Taneichi, K. Nomura, T. Wada, A. Yachie, K. Imai, T. Ariga, I. Santisteban, M.S. Hershfield, and T. Miyawaki.	Successful bone marrow transplantation with reduced intensity conditioning in a patient with delayed-onset adenosine deaminase deficiency.	Pediatr Transplant	in press		2012
Kamae C, Nakagawa N, Sato H, Honma K, Mitsui N, Ohara O, Kanegane H, Pasic S, Pan-Hammarström Q, van Zelm MC, Morio T, Imai K, Nonoyama S.	Common variable immunodeficiency classification by quantifying T-cell receptor and immunoglobulin κ-deleting recombination excision circles.	J Allergy Clin Immunol.	131	1437-1440	2013
Kakiuchi S, Nonoyama S, Wakamatsu H, Kogawa K, Wang L, Kinoshita-Yamaguchi H, Takayama-Ito M, Lim CK, Inoue N, Mizuguchi M, Igarashi T, Saijo M.	Neonatal herpes encephalitis caused by a virologically confirmed acyclovir resistant herpes simplex virus type 1.	J. Clin. Microbiol.	51	356-359	2013
Wakabayashi M, Mori T, Isobe K, Sohara E, Susa K, Araki Y, Chiga M, Kikuchi E, Nomura N, Mori Y, Matsuo H, Murata T, Nomura S, Asano T, Kawaguchi H, Nonoyama S, Rai T, Sasaki S, Uchida S.	Impaired KLHL3-Mediated Ubiquitination of WNK4 Causes Human Hypertension.	Cell Reports.	3	858-868	2013
Bousfiha A, Jeddane L, Ailal F, Al-Herz, Conley M.E, Cunningham-Rundles C, Etzioni A, Fischer A, Franco J.L, Geha R.S, Hammarström L, Nonoyama S, Ochs H.D, Roifman C, Seger R, Tang M.L.K, Puck J.M, Chapel H, Notarangelo L.D, Casanova J.L.	A Phenotypic Approach for IUIS PID Classification and Diagnosis: Guidelines for Clinicians at the Bedside.	J Clin Immunol.	33	1078-1087	2013
Osawa M, Ogura Y, Isobe K, Uchida S, Nonoyama S and Kawaguchi H.	CUL3 gene analysis enables early intervention for pediatric pseudohypaldosteronism type II in infancy.	Pediatr Nephrol.	28	1881-1884	2013
Kumaki S, Sasahara Y, Kamachi Y, Muramatsu H, Morio T, Goi K, Sugita K, Urabe T, Takada H, Kojima S, Tsuchiya S, Hara T.	B cell function after unrelated umbilical cord blood transplantation using minimal-intensity conditioning regimen in patients with X-SCID.	Int J Hematol	98	355-60	2013
Ishimura M, Yamamoto H, Mizuno Y, Takada H, Goto M, Doi T, Hoshina T, Ohga S, Ohshima K, Hara T.	A non-invasive diagnosis of histiocytic necrotizing lymphadenitis by means of gene expression profile analysis of peripheral blood mononuclear cells.	J Clin Immunol.	33(5)	1018-26	2013

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Imagawa T, Nishikomori R, Takada H, Takeshita S, Patel N, Kim D, Lheritier K, Heike T, Hara T, Yokota S.	Safety and efficacy of canakinumab in Japanese patients with phenotypes of cryopyrin-associated periodic syndrome as established in the first open-label, phase-3 pivotal study (24-week results).	Clin Exp Rheumatol.	31	302-9	2013
Yokota S, Nishikomori R, Takada H, Kikuchi M, Nozawa T, Kanetaka T, Kizawa T, Miyamae T, Mori M, Heike T, Hara T, Imagawa T.	Guidance on the use of canakinumab in patients with cryopyrin-associated periodic syndrome in Japan.	Mod Rheumatol	23	425-9	2013
Ninomiya T, Takada H, Nagatomo Y, Nanishi E, Nagata H, Yamamura K, Doi T, Ikeda I, Hara T.	Development of Kawasaki disease in a patient with PFAPA.	Pediatrics International	55(6)	801-2	2013
Fukazawa M, Hoshina T, Nanishi E, Nishio H, Doi T, Ohga S, Hara T.	Neonatal hemophagocytic lymphohistiocytosis associated with a vertical transmission of coxsackievirus B1.	J Infect Chemother	19(6)	1210-3	2013
Higuchi Y, Shimizu J, Hatanaka M, Kitano E, Kitamura H, Takada H, Ishimura M, Hara T, Ohara O, Asagoe K, Kubo T.	The identification of a novel splicing mutation in C1qB in a Japanese family with C1q deficiency: a case report.	Pediatr Rheumatol Online J.	11(1)	41	2013
Obinata K, Lee T, Niizuma T, Kinoshita K, Shimizu T, Hoshina T, Sasaki Y, Hara T.	Two cases of partial dominant interferon- $\gamma$ receptor 1 deficiency that presented with different clinical courses of bacille Calmette-Guérin multiple osteomyelitis.	J Infect Chemother.	19	757-60	2013
Kanegane H, Taneichi H, Nomura K, Wada T, Yachie A, Imai K, Ariga T, Santisteban I, Hershfield MS, Miyawaki T.	Successful bone marrow transplantation with reduced intensity conditioning in a patient with delayed-onset adenosine deaminase deficiency.	Pediatr Transplant.	17	E29-E32	2013
Rawat A, Singh S, Suri D, Gupta A, Saikia B, Minz RW, Sehgal S, Vaiphei K, Kamae C, Honma K, Nakagawa N, Imai K, Nonoyama S, Oshima K, Mitsuiki N, Ohara O, Chan KW, Lau YL.	Chronic Granulomatous Disease: Two Decades of Experience From a Tertiary Care Centre in North West India.	J Clin Immunol.	33(4)	857-864	2013
Wada T, Sakakibara Y, Nishimura R, Toma T, Ueno Y, Horita S, Tanaka T, Nishi M, Kato K, Yasumi T, Ohara O, Yachie A.	Down-regulation of CD5 expression on activated CD8(+) T cells in familial hemophagocytic lymphohistiocytosis with perforin gene mutations.	Hum Immunol.	74:12	1579-1585	2013
Lee YW, Yang EA, Kang HJ, Yang X, Mitsuiki N, Ohara O, Miyawaki T, Kanegane H, Lee JH.	Novel mutation of IL2RG gene in a Korean boy with X-linked severe combined immunodeficiency.	J Investig Allergol Clin Immunol.	23:1	65-67	2013
Suzuki J, Kuwahara M, Tofukuji S, Imamura M, Kato F, Nakayama T, Ohara O, Yamashita M.	A novel small compound SH-2251 suppresses Th2 cell-dependent airway inflammation through selective modulation of chromatin status at the Il5 gene locus.	PLoS One.	16:8	e61785	2013
Wada T, Muraoka M, Toma T, Imai T, Shigemura T, Agematsu K, Haraguchi K, Moriuchi H, Oh-Ishi T, Kitoh T, Ohara O, Morio T, Yachie A.	Rapid detection of intracellular p47phox and p67phox by flow cytometry; useful screening tests for chronic granulomatous disease.	J Clin Immunol.	33:4	857-864	2013
Sakaguchi H, Okuno Y, Muramatsu H, Yoshida K, Shiraishi Y, Takahashi M, Kon A, Sanada M, Chiba K, Tanaka H, Makishima H, Wang X, Xu Y, Doisaki S, Hama A, Nakanishi K, Takahashi Y, Yoshida N, Maciejewski JP, Miyano S, Ogawa S, Kojima S.	Exome sequencing identifies secondary mutations of SETBP1 and JAK3 in juvenile myelomonocytic leukemia.	Nat Genet	45(8)	937-41	2013
Takahashi Y, Muramatsu H, Sakata N, Hyakuna N, Hamamoto K, Kobayashi R, Ito E, Yagasaki H, Ohara A, Kikuchi A, Morimoto A, Yabe H, Kudo K, Watanabe K, Ohga S, Kojima S, Japan Childhood Aplastic Anemia Study G.	Rabbit antithymocyte globulin and cyclosporine as first-line therapy for children with acquired aplastic anemia.	Blood	121(5)	862-3	2013

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Yoshimi A, Kamachi Y, Imai K, Watanabe N, Nakadate H, Kanazawa T, Ozono S, Kobayashi R, Yoshida M, Kobayashi C, Hama A, Muramatsu H, Sasahara Y, Jakob M, Morio T, Ehl S, Manabe A, Niemeyer C, Kojima S	Wiskott-Aldrich syndrome presenting with a clinical picture mimicking juvenile myelomonocytic leukaemia.	Pediatr Blood Cancer	60(5)	836-41	2013
Morishima T, Watanabe K, Niwa A, Hirai H, Saida S, Tanaka T, Kato I, Umeda K, Hiramatsu H, Saito M, Matsubara K, Adachi S, Kobayashi M, Nakahata T, Heike T.	Genetic correction of HAX1 in induced pluripotent stem cells from a patient with severe congenital neutropenia improves defective granulopoiesis.	Haematologica	99	19-27	2014
Mizoguchi Y, Tsumura M, Okada S, Hirata O, Minegishi S, Imai K, Hyakuna N, Muramatsu H, Kojima S, Ozaki Y, Imai T, Takeda S, Okazaki T, Yasunaga S, Takihara Y, Bryant V, Kong X, Crypwy S, Dupuis S, Casanova JL, Morio T, Kobayashi M	Simple diagnosis of STAT1 gain-of-function alleles in patients with chronic mucocutaneous candidiasis.	J Leukoc Biol.		Epub ahead of print	2013
Deenick EK, Avery DT, Chan A, Berglund LJ, Ives ML, Bustamante J, Boisson-Dupuis S, Tsumura M, Kobayashi M, Arkwright PD, Averbuch D, Engelhard D, Roesler J, Peake J, Wong M, Adelstein S, Choo S, Smart JM, French MA, Fulcher DA, Cook MC, Picard C, Durandy A, Klein C, Holland SM, Uzel G, Casanova JL, Ma CS, Tangye SG.	Naïve and memory human B cells have distinct requirements for STAT3 activation to differentiate into antibody-secreting plasma cells.	J Exp Med.	210	2739-53	2013
Berglund LJ, Ma CS, Avery DT, Moens L, Deenick EK, Bustamante J, Boisson-Dupuis S, Wong M, Adelstein S, Arkwright PD, Fulcher DA, Ziegler JB, Smart JM, Kobayashi M, Casanova JL, Cook MC, Uzel G, Tangye SG	IL-21 signalling via STAT3 primes human naïve B cells to respond to IL-2 to enhance their differentiation into plasmablasts.	Blood	122	3940-50	2013
Shiba N, Funato M, Ohki K, Park MJ, Mizushima Y, Adachi S, Kobayashi M, Kinoshita A, Sotomatsu M, Arakawa H, Tawa A, Horibe K, Tsukimoto I, Hayashi Y.	Mutations of the GATA2 and CEBPA genes in paediatric acute myeloid leukaemia.	Br J Haematol.		Epub ahead of print	2013
Ives ML, Ma CS, Palendira U, Chan A, Bustamante J, Boisson-Dupuis S, Arkwright PD, Engelhard D, Averbuch D, Magdorf K, Roesler J, Peake J, Wong M, Adelstein S, Choo S, Smart JM, Frnch MA, Fulcher DA, Cook MC, Picard C, Durandy A, Tsumura M, Kobayashi M, Uzel G, Casanova JL, Tangye SG, Deenick EK.	Signal transducer and activator of transcription 3 (STAT3) mutations underlying autosomal dominant hyper-IgE syndrome impair human CD8(+) T-cell memory formation and function.	J Allergy Clin Immunol.	132	400-11	2013
Hirata O, Okada S, Tsumura M, Kagawa R, Miki M, Kawaguchi H, Nakamura K, Boisson-Dupuis S, Casanova JL, Takihara Y, Kobayashi M.	Heterozygosity for the Y701C STAT1 mutation in a multiplex kindred with multifocal osteomyelitis.	Haematologica	98	1641-9	2013
Hiramoto T, Ebihara Y, Mizoguchi Y, Nakamura K, Yamaguchi K, Ueno K, Mochizuki S, Yamamoto S, Nagasaki M, Furukawa Y, Tani K, Nakauchi H, Kobayashi M, Tsuji K.	Wnt3a stimulates maturation of impaired neutrophils developed from severe congenital neutropenia-derived pluiipotent stem cells with heterozygous ELANE mutation.	Proc Natl Acad Sci USA	110	3023-8	2013
Kobayashi Y, Matsui H, Kanai A, Tsumura M, Okada S, Miki M, Nakamura K, Kunishima S, Inaba T, Kobayashi M.	Identification of the integrin β3 L718P mutation in a pedigree with autosomal dominant thrombocytopenia with anisocytosis.	Br J Haematol.	160	521-9	2013



## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Kumaki S, Sasahara Y, Kamachi Y, Muramatsu H, Morio T, Goi K, Sugita K, Urabe T, Takada H, Kojima S, Tsuchiya S, Hara T.	B-cell function after unrelated umbilical cord blood transplantation using a minimal-intensity conditioning regimen in patients with X-SCID.	Int J Hematol.	98	355-360	2013
Isoda T, Mitsuiki N, Ohkawa T, Kaneko S, Endo A, Ono T, Aoki Y, Tomizawa D, Kajiwara M, Araki S, Nagasawa M, Morio T, Takagi M, Mizutani S.	Irreversible Leukoencephalopathy After Reduced-intensity Stem Cell Transplantation in a Dyskeratosis Congenita Patient With TINF2 Mutation.	J Pediatr Hematol Oncol.	35	e178-182	2013
Kawasaki Y, Toyoda H, Otsuki S, Iwasa T, Iwamoto S, Azuma E, Itoh-Habe N, Wada H, Fujimura Y, Morio T, Imai K, Mitsuiki N, Ohara O, Komada Y.	A novel Wiskott-Aldrich syndrome protein mutation in an infant with thrombotic thrombocytopenic purpura.	Eur J Haematol.	290	164-168	2013
Rebecca Marsh, Kanchan Rao,Prakash Satwani, Kai Lehmborg, Ingo Müller, Dandan Li, Mi-Ok Kim, Alain Fischer, Sylvain Latour, Petr Sedlacek, Vincent Barlogis, Kazuko Hamamoto, Hirokazu Kanegane, Sam Milanovich, David Margolis, David Dimmock, James Casper, Dorothea N Douglas, Persis J Amrolia, Paul Veys, Ashish R Kumar, Michael B Jordan, Jack J Bleesing, Filipovich, Alexandra H.	Allogeneic hematopoietic cell transplantation for XIAP deficiency: an international survey reveals poor outcomes.	Blood	121:6	877-83	2013
Tomizawa D, Tawa A, Watanabe T, Saito A.M, Kudo K, Taga T, Iwamoto S, Shimada A, Terui K, Moritake H, Kinoshita A, Takahashi H, Nakayama H, Kiyokawa N, Isoyama K, Mizutani S, Hara J, Horibe K, Nakahata T, Souichi Adachi S.	Excess reduction of anthracyclines results in inferior event-free survival in core binding factor acute myeloid leukemia in children: a report from the Japanese Pediatric Leukemia/Lymphoma Study Group (JPLSG).	Leukemia		in press	
Kodera Y, Yamamoto K, Harada M, Morishima Y, Dohy H, Asano S, Ikeda Y, Nakahata T, Imamura M, Kawa K, Kato S, Tanimoto M, Kanda Y, Tanosaki R, Shiobara S, Kim SW, Nagafuji K, Hino M, Miyamura K, Suzuki R, Hamajima N, Fukushima M, Tamakoshi A; for the Japan Society for Hematopoietic Cell Transplantation, Halter J, Schmitz N, Niederwieser D, Gratwohl A.	PBSC collection from family donors in Japan: a prospective survey.	Bone Marrow Transplant.		in press	
Honda Y, Tsuchida M., Zaike Y, Masunaga A, Yoshimi A, Kojima S, Ito M, Kikuchi A, Nakahata T, Manabe A.	Clinical characteristics of 15 children with juvenile myelomonocytic leukemia who developed blast crisis: MDS Committee of Japanese Society of Pediatric Hematology/Oncology (JSPHO).	Brit. J. Haematol.		in press	
Saida S, Watanabe KI, Sato-Otsubo A, Terui K, Yoshida K, Okuno Y, Toki T, Wang R, Shiraiishi Y, Miyano S, Kato I, Morishima T, Fujino H, Umeda K, Hiramatsu H, Adachi S, Ito E, Ogawa S, Ito M, Nakahata T, Heike T	Clonal selection in xenografted TAM recapitulates the evolutionary process of myeloid leukemia in Down syndrome.	Blood.	121(21)	4377-87	2013
Yanagimachi MD, Niwa A, Tanaka T, Ozaki F, Nishimoto S, Murata Y, Yasumi T, Ito J, Tomida S, Oshima K, Asaka I, Goto H, Heike T, Nakahata T, Saito MK	Robust and highly-efficient differentiation of functional monocytic cells from human pluripotent stem cells under serum- and feeder cell- free conditions.	PLoS ONE.	8(4)	e59243	2013
Nakazawa Y, Saito S, Yanagisawa R, Suzuki T, Toshiro Ito T, Ishida F, Muramatsu H, Matsumoto K, Kato K, Ishida H, Umeda K, Souichi Adachi S, Nakahata T, Koike K.	Recipient seropositivity for adenovirus type 11 is a highly predictive factor for the development of AdV11-induced hemorrhagic cystitis after allogeneic hematopoietic SCT.	Bone Marrow Transplant.	48	737-739	2013

## 雑誌(英文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Ichikawa M, Arai Y, Haruta M, Furukawa S, Ariga T, Kajii T, Kaneko Y.	Meiosis error and subsequent genetic and epigenetic alterations invoke the malignant transformation of germ cell tumor.	Gene, Chromosome & Cancer	52	274-286	2013
Horino S, Sasahara Y, Sato M, Niizuma H, Kumaki S, Abukawa D, Sato A, Imaizumi M, Kanegane H, Kamachi Y, Sasaki S, Terui K, Ito E, Kobayashi I, Ariga T, Tsuchiya S, Kure S.	Selective expansion of donor-derived regulatory T cells after allogeneic bone marrow transplantation in a patient with IPEX syndrome.	Pediatr Transplant	18	E25-E30	2013

## 雑誌(和文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
溝口 洋子, 鎌田 綾, 三木 瑞香, 谷 博雄, 世羅 康彦, 中村 和洋, 小林 正夫	Glanzmann thrombasthenia への遺伝子組み 換え活性型第 VII 因子製剤による止血効果	日本小児血液・がん 学会雑誌	49(1-2)9	61-66	2012
梶梅 輝之, 浅野 孝基, 世羅 康彦, 小林 正夫	輸血後蕁麻疹発症前の末梢血一般検査所見	アレルギー	61(8)	1086- 1091	2012
溝口洋子, 津村弥来, 岡田賢, 小林正夫	慢性性皮膚粘膜カンジダ症と機能獲得性 STAT1 変異	臨床免疫・アレルギー 科	57(4)	437-443	2012
中畑龍俊	白血病治療の進歩と今後の展望	日本小児血液・がん 学会雑誌	第 49 巻 1・2 号		2012
中畑龍俊, 丹羽明	幹細胞増幅, 第 10 章 内科疾患と再生医療	カラー版内科学 門脇孝, 永井良三( 総 編集)		447-450	2012
今井 耕輔	原発性免疫不全症の最新国際分類. 臨床免 疫	アレルギー科	58	446-466	2012
今井耕輔	原発性免疫不全症の遺伝子診断・治療	臨床血液	53	1865- 1873	2012
今井耕輔	特集 乳幼児健診 Q&A . 歯科 Q 口の 中にミルクのかすのようなものがいつもあ るのですが、大丈夫ですか	小児科診療	11	2045- 2048	2012
今井耕輔	臨床検査の意義と限界 T 細胞・B 細胞サブ セット	小児内科	44	645-648	2012
今井耕輔	【知っておきたい内科症候群】 膠原病・免 疫・アレルギー《免疫不全症》 ウィスコ ット・アルドリッチ症候群	内科	109	1501- 1503	2012
原 寿郎	2. 幹細胞異常と内科系疾患、現状と展望 1) 造血幹細胞の異常: 先天性免疫不全症	日本内科学会雑誌	102(9)	2255- 2261	2013
原 寿郎	小児感染・免疫疾患の発症におけるヒト - 環境相互作用	小児感染免疫	25(1)	41-53	2013
原 寿郎	シリーズ小児医療第 6 回 原発性免疫不全 症研究: 最新の進歩	あいみっく	34(3)	50-5	2013
原 寿郎	こどもの発熱の原因とその対処法	ふたば	77	18-24	2013
中村和洋, 小林正夫	新生児同種免疫性好中球減少症	臨床免疫・アレルギー 科	60	78-82	2013
波多野 修一, 駒澤 克孝, 西村 真一郎, 藤江 篤志, 大野 令央 義, 川口 浩史, 小林 正夫, 高尾 信一.	マイコプラズマ感染症検査法の検討 マイ コプラズマ抗原迅速診断キットの有用性 について	小児科臨床	66	2105- 2115	2013
下村 麻衣子, 千々松 郁枝, 浅 野 孝基, 古江 綾, 三木 瑞香, 川口 浩史, 中村 和洋, 小林 正夫	慢性肉芽腫症における消化管病変	広島医学	66	473-474	2013
高坂 卓馬, 秀 道広, 小林 正 夫	Omenn 症候群の 1 例	西日本皮膚科	75	269	2013
唐川 修平, 中村 和洋, 小林 正夫	【クローズアップ 新しい子どもの病気】 血液腫瘍疾患 新しい診断技術で診断可能 となった疾患 好中球減少症 遺伝子変異 と抗好中球抗体	小児内科	45	1131- 1133	2013

## 雑誌(和文)

発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
三木 瑞香, 小林 正夫	【クローズアップ 負荷試験の実際 2013】 血液系機能検査 好中球減少症の負荷試験	小児内科	45	989-991	2013
平田 修, 小林 正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】リンパ球の異常 リンパ球機能異常と類縁疾患 原発性免疫不全症候群 単独 IgG サブクラス欠損症	日本臨床別冊血液症候群 第2版 II		250-253	2013
早川 誠一, 小林 正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】リンパ球の異常 リンパ球機能異常と類縁疾患 原発性免疫不全症候群 IgM 単独(選択的)欠損症	日本臨床別冊血液症候群 第2版 II		246-249	2013
平田 修, 小林 正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】リンパ球の異常 リンパ球機能異常と類縁疾患 原発性免疫不全症候群 選択的 IgA 欠損症	日本臨床別冊血液症候群第2版 II		242-245	2013
平田 修, 中村 和洋, 小林 正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】白血球(顆粒球)の異常(悪性腫瘍を除く) 好中球の異常 好中球減少症 周期性好中球減少症	日本臨床別冊血液症候群 第2版 II		57-60	2013
唐川 修平, 中村 和洋, 小林 正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】白血球(顆粒球)の異常(悪性腫瘍を除く) 好中球の異常 好中球減少症 自己免疫性好中球減少症	日本臨床別冊血液症候群 第2版 II		54-56	2013
溝口 洋子, 中村 和洋, 小林 正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】白血球(顆粒球)の異常(悪性腫瘍を除く) 好中球の異常 好中球減少症 同種免疫性好中球減少症.	日本臨床別冊血液症候群 第2版 II		50-53	2013
唐川 修平, 小林 正夫	【知っておきたい最新の免疫不全症分類-診断から治療まで】治療 原発性免疫不全症に対する造血幹細胞移植	小児科診療	76	476-480	2013
宮地 隆史, 丸山 博文, 小林 正夫, 松本 昌泰	【クローズアップ 呼吸管理】 <在宅呼吸ケア> 在宅人工呼吸器装着者の災害時対策	小児内科	45	116-120	2013
斎藤潤, 中畑龍俊	疾患特異的 iPS 細胞	再生医療	12(1)	19-29	2013
川口裕之	Virus-associated hemophagocytic syndrome	日本臨床	別冊 No.23	466-9	2013
戸田尚子, 原 寿郎	2. 疾患と栄養 先天性免疫不全症と低栄養	臨床栄養		印刷中	2014

## 書籍

著者氏名	論文タイトル	書籍全体の編集者名	書籍名	出版社名	出版地	出版年	ページ
原 寿郎	第1章：血液系疾患の医療ニーズ 第3節 原発性免疫不全症候群		希少疾患/難病の診断・治療と製品開発	(株)技術情報協会	東京	2013	593-610
原 寿郎	免疫疾患	原 寿郎/高橋孝雄/ 細井 創	標準小児科学 第8版	医学書院	東京	2014	258-79
原 寿郎	原発性免疫不全症候群 Primary immunodeficiency syndrome	福井次矢/高木 誠/ 小室一成	今日の治療指針 2014年版 - 私はこう治療している	医学書院	東京	2014	1270-1