Ⅲ．　研究成果の刊行に関する一覧表

雑誌（英文）

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Yabe M, Morio T, Tabuchi K, Tomizawa D, Hasegawa D, Ishida H, Yoshida N, Koike T, Takahashi Y, Koh K, Okamoto Y, Sano H, Kato K, Kanda Y, Goto H, Takita J, Miyamura T, Noguchi M, Kato K, Hashii Y, Astuta Y, Yabe H.	Long-term outcome in patients with Fanconi anemia who received hematopoietic stem cell transplantation: a retrospective nationwide analysis.	Int J Hematol.	113	134-144	2021
Rawat A, Jindal AK, Suri D, Vignesh P, Gupta A, Saikia B, Minz RW, Banday AZ, Tyagi R, Arora K, Joshi V, Mondal S, Shandilya JK, Sharma M, Desai M, Taur P, Pandrowala A, Gowri V, Sawant-Desai S, Gupta M, Dalvi AD, Madkaikar M, Aggarwal A, Raj R, Uppuluri R, Bhattad S, Jayaram A, Lashkari HP, Rajasekhar L, Munirathnam D, Kalra M, Shukla A, Saka R, Sharma R, Garg R, Imai K, Nonoyama S, Ohara O, Lee PP, Chan KW, Lau YL, Singh S.	Clinical and Genetic Profile of X-Linked Agammaglobulinemia: A Multicenter Experience From India.	Front Immunol.	11	612323	2021
Mitsui-Sekinaka K, Narumi S, Sekinaka Y, Uematsu K, Yoshida Y, Amano N, Shima H, Hasegawa T and Nonoyama S.	Clinical and immunological analyses of ten patients with MIRAGE syndrome.	J Clin Immunol.	41	709-711	2021
Hagiwara H, Matsumoto H, Uematsu K, Zaha K, Sekinaka Y, Miyake N, Matsumoto N, Nonoyama S.	Immunodeficiency in a patient with microcephalic osteodysplastic primordial dwarfism type I as compared to Roifman syndrome.	Brain Dev	43	337-342	2021
Vignesh P, Loganathan SK, Sudhakar M, Chaudhary H, Rawat A, Sharma M, Shekar A, Vaiphei K, Kumar N, Sachdeva MUS, Jindal AK, Suri D, Gupta A, Ray P, Imai K, Ohara O, Nonoyama S, Lau YL and Singh S.	Hemophagocytic Lymphohistiocytosis in Children with Chronic Granulomatous Disease-Single-Center Experience from North India.	J Allergy Clin Immunol Pract	9	:771-782.e3	2021
Rawat A, Vignesh P, Sudhakar M, Sharma M, Suri D, Jindal A, Gupta A, Shandilya JK, Loganathan SK, Kaur G, Chawla S, Patra1 PK, Khadwal A, Saikia B, Minz RW, Aggarwal V, Taur P, Pandrowala A, Gowri V, Desai M, Kulkarni M, Hule G, Bargir U, Kambli P, Madkaikar M, Bhattad S, Ginigeri C, Kumar H, Jayaram A, Munirathnam D, Sivasankaran M, Raj R, Uppuluri R, Na F, George B, Lashkari HP, Kalra M, Sachdeva A, Seth S, Sabui T, Gupta A, Leeuwen KV, Boer M, Chan KW, Imai K, Ohara O, Nonoyama S, Lau YL and Singh S.	Clinical, Immunological, and Molecular Profile of Chronic Granulomatous Disease: A Multi-Centric Study of Patients From India.	Front Immunol.	12	625320	2021
Nishimura S, Kobayashi Y, Ohnishi H, Moriya K, Tsumura M, Sakata S, Mizoguchi Y, Takada H, Kato Z, Sancho-Shimizu V, Picard C, Irani SR, Ohara O, Casanova JL, Puel A, Ishikawa N, Okada S, Kobayashi M.	IRAK4 Deficiency Presenting with Anti-NMDAR Encephalitis and HHV6 Reactivation.	J Clin Immunol.	41	125-135	2021
Qin T, Jia Y, Liu Y, Dai R, Zhou L, Okada S, Tsumura M, Ohnishi H, Kato Z, Kanegane H, Sun X, Zhao X.	A Novel Homozygous Mutation Destabilizes IKKβ and Leads to Human Combined Immunodeficiency.	Front Immunol.	15;11	517544	2021
Kadowaki S, Hashimoto K, Nishimura T, Kashimada K, Kadowaki T, Kawamoto N, Imai K, Okada S, Kanegane H, Ohnishi H.	Functional analysis of novel A20 variants in patients with atypical inflammatory diseases.	Arthritis Res Ther.	23	52	2021
Kataoka S, Kawashima N, Okuno Y, Muramatsu H, Miwata S, Narita K, Hamada M, Murakami N, Taniguchi R, Ichikawa D, Kitazawa H, Suzuki K, Nishikawa E, Narita A, Nishio N, Yamamoto H, Fukasawa Y, Kato T, Yamamoto H, Natsume J, Kojima S, Nishino I, Taketani T, Ohnishi H, Takahashi Y.	Successful treatment of a novel type I interferonopathy due to a de novo PSMB9 gene mutation with a Janus kinase inhibitor.	J Allergy Cin Immunol.	doi: 10.1016/j.jaci.2021.03.010.		2021
Shiraki M, Kadowaki S, Kadowaki T, Kawamoto N, Ohnishi H.	Primary Immunodeficiency Disease Mimicking Pediatric Bechet's Disease.	Children (Basel)	22;8	75	2021
Kadowaki S, Kimura T, Shiraki M, Mizutani Y, Nakama M, Kobayashi K, Suzui N, Kawamoto N, Ohnishi H, Seishima M.	Case of Muckle-Wells syndrome with erythema dominantly infiltrated by lymphocytes.	J Dermatol	48(2)	e100-e101	2021

Shimizu M, Matsubayashi T, Ohnishi H, Nakama M, Izawa K, Honda Y, Nishikomori R.	Haploinsufficiency of A20 with a novel mutation of deletion of exons 2-3 of TNFAIP3.	Mod Rheumatol	31(2)	493-497	2021
Mizoguchi Y, Okada S	Inborn Errors of STAT1 Immunity	Curr Opin Immunol	doi: 10.1016/j.coi.2021.02.009.		2021
Miyashita K, Matsuda Y, Okajima M, Toma T, Yachie A, Wada T.	Role of E148Q in familial Mediterranean fever with an exon 10 mutation in MEFV.	Pediatr Int.	doi: 10.1111/ped.14696.		2021
Mizuta M, Shimizu M, Irabu H, Usami M, Inoue N, Nakagishi Y, Wada T, Yachie A.	Comparison of serum cytokine profiles in macrophage activation syndrome complicating different background rheumatic diseases in children.	Rheumatology (Oxford)	60(1)	231-238	2021
Hatano M, Fukushima H, Ohto T, Ueno Y, Saeki S, Enokizono T, Tanaka R, Tanaka M, Imagawa K, Kanai Y, Kato M, Shiraku H, Suzuki H, Uehara T, Takenouchi T, Kosaki K, Takada H.	Variants in KIF2A cause broad clinical presentation; the computational structural analysis of a novel variant in a patient with a cortical dysplasia, complex, with other brain malformations 3.	Am J Med Genet A.	doi: 10.1002/ajmg.a.62084.		2021
Uehara T, Morino S, Oishi K, Nakamura Y, Togashi N, Imaizumi M, Nishimura S, Okada S, Yara A, Fukushima H, Imagawa K, Takada H	Pneumococcal serotype-specific opsonophagocytic activity in interleukin-1 receptor-associated kinase 4-deficient patients.	Pediatr Infect Dis J.	doi: 10.1097/INF.0000000000003060.		2021
Yamaki Y, Fukushima T, Yoshida N, Nishimura K, Fukuda A, Hisatake K, Aso M, Sakasai T, Kijima-Tanaka J, Miwa Y, Nakanishi M, Sumazaki R, Takada H.	Utilization of a novel Sendai virus vector in ex vivo gene therapy for hemophilia A. Int J Hematol.	Int J Hematol.	doi: 10.1007/s12185-020-03059-6.		2021
Morita A, Enokizono T, Ohto T, Tanaka M, Watanabe S, Takada Y, Iwama K, Mizuguchi T, Matsumoto N, Morita M, Takashima S, Shimozawa N, Takada H.	Novel ACOX1 mutations in two siblings with peroxisomal acyl-CoA oxidase deficiency.	Brain Dev.	43	475-481	2021
Nakashima Y, Sakai Y, Mizuno Y, Furuno K, Hirono K, Takatsuki S, Suzuki H, Onouchi Y, Kobayashi T, Tanabe K, Hamase K, Miyamoto T, Aoyagi R, Arita M, Yamamura K, Tanaka T, Nishio H, Takada H, Ohga S, Hara T.	Lipidomics links oxidized phosphatidylcholines and coronary arteritis in Kawasaki disease.	Cardiovasc Res.	1;117	96-108	2021
Yamashita M, Inoue K, Okano T, Morio T.	Inborn errors of immunity-recent advances in research on the pathogenesis.	Inflamm Regen.	25;41	9	2021
Morio T, Gotoh K, Imagawa T, Morita K, Ohnishi H, Yasui K, Hofmann J, Lawo JP, Shebl A, Rojavin MA.	Safety and tolerability of IgPro10 in Japanese primary immunodeficiency patients: a registrational study.	Int J Hematol	113(6)	921-929	2021
Tangye SG, Al-Herz W, Bousfiha A, Cunningham-Rundles C, Franco JL, Holland SM, Klein C, Morio T, Oksenhendler E, Picard C, Puel A, Puck J, Seppänen MRJ, Somech R, Su HC, Sullivan KE, Torgerson TR, Meyts I.	The Ever-Increasing array of novel inborn errors of immunity: an interim update by the IUIS committee.	J Clin Immunol.	41	666-679	2021
Imanaka Y, Taniguchi M, Doi T, Tsumura M, Nagaoka R, Shimomura M, Asano T, Kagawa R, Mizoguchi Y, Karakawa S, Arihiro K, Imai K, Morio T, Casanova JL, Puel A, Ohara O, Kamei K, Kobayashi M, Okada S.	Inherited CARD9 deficiency in a child with invasive disease due to exophiala dermatitidis and two older but asymptomatic siblings.	J Clin Immunol	doi: 10.1007/s10875-021-00988-7.		2021

Nihira H, Izawa K, Ito M, Umebayashi H, Okano T, Kajikawa S, Nanishi E, Keino D, Murakami K, Isa-Nishitani M, Shiba T, Honda Y, Hijikata A, Yasu T, Kubota T, Hasegawa Y, Kawashima Y, Nakano N, Takada H, Ohga S, Heike T, Takita J, Ohara O, Takei S, Takahashi M, Kanegane H, Morio T, Iwaki-Egawa S, Sasahara Y, Nishikomori R, Yasumi T.	Detailed analysis of Japanese patients with adenosine deaminase 2 deficiency reveals characteristic elevation of type II interferon signature and STAT1 hyperactivation.	J Allergy Clin Immunol	30;S0091-6749(21)00157-3		2021
Nishimura A, Aoki Y, Ishiwata Y, Ichimura T, Ueyama J, Kawahara Y, Tomoda T, Inoue M, Matsumoto K, Inoue K, Hiroki H, Ono S, Yamashita M, Okano T, Tanaka-Kubota M, Ashiarai M, Miyamoto S, Miyawaki R, Yamagishi C, Tezuka M, Okawa T, Hoshino A, Endo A, Yasuhara M, Kamiya T, Mitsuiki N, Ono T, Isoda T, Yanagimachi M, Tomizawa D, Nagasawa M, Mizutani S, Kajiwara M, Takagi M, Kanegane H, Imai K, Morio T.	Hematopoietic cell transplantation with reduced intensity conditioning using Fludarabine/Busulfan or Fludarabine/Melphalan for primary immunodeficiency diseases.	J Clin Immunol.	doi: 10.1007/s10875-021-00966-z.		2021
Tanita K, Sakura F, Nambu R, Tsumura M, Imanaka Y, Ohnishi H, Kato Z, Pan J, Hoshino A, Suzuki K, Yasutomi M, Umetsu S, Okada C, Takagi M, Imai K, Ohara O, Muise AM, Okada S, Morio T, Kanegane H.	Clinical and immunological heterogeneity in Japanese patients with Gain-of-Function variants in STAT3.	J Clin Immunol.	41	780-790	2021
Inoue M, Isoda T, Yamashita M, Tomoda T, Inoue K, Okano T, Ohkawa T, Endo A, Mitsuiki N, Kamiya T, Yanagimachi M, Yamamoto K, Inaba Y, Sasaki T, Takagi M, Kanegane H, Imai K, Morio T.	Cytomegalovirus Laryngitis in Primary Combined Immunodeficiency Diseases.	J Clin Immunol.	41	243-247	2021
Dezfouli M, Bergström S, Skattum L, Abolhassani H, Neiman M, Torabi-Rahvar M, Franco-Jarava C, Nalda MA, Ferrer JM, Slade C, Roos A, Fernandez Pereira LM, López-Trascasa M, Gonzalez-Granado LI, Luis M. Allende, Mizuno Y, Yoshida Y, Friman V, Lundgren A, Aghamohammadi A, Rezaei N, Hernández-Gonzalez M, Ulrika Von Döbeln, Truedsson L, Hara T, Nonoyama S, Schwenk JM, Nilsson P, Hammarström L.	Newborn Screening for Presymptomatic Diagnosis of Complement and Phagocyte Deficiencies.	Front Immunol.	11	455	2020
Matsuda T, Kambe N, Ueki Y, Kanazawa N, Izawa K, Honda Y, Kawakami A, Takei S, Tonomura K, Inoue M, Kobayashi H, Okafuji I, Sakurai Y, Kato N, Maruyama Y, Inoue Y, Otsubo Y, Makino T, Okada S, Kobayashi I, Yashiro M, Ito S, Fujii H, Kondo Y, Okamoto N, Ito S, Iwata N, Kaneko U, Doi M, Hosokawa J, Ohara O, Saito MK, Nishikomori R	PIDJ members in the JSIAD; PIDJ (Primary Immunodeficiency and Autoinflammatory Diseases Database Project) members in the JSIAD (Japanese Society for Immunodeficiency and Autoinflammatory Diseases). Clinical characteristics and treatment of 50 cases of Blau syndrome in Japan confirmed by genetic analysis of the NOD2 mutation.	Ann Rheum Dis.	79	1492-1499	2020
Vignesh P, Rawat A, Kumrah R, Singh A, Anjani G, Sharma M, Kaur A, Nameirakpam J, Jindal A.K, Suri D, Gupta A, Khadwal A, Saikia B, Minz R.W., Desai M, Taur P, Gowri V, Pandrowala A.A, Dalvi A.D, Jodhawat N, KAMBLI P.M, Madkaikar M.R, Bhattad S, Ramprakash S, Raghuram CP, Vikas A, Sivasankaran M, Munirathnam D, Balaji S, Rajendran A, Aggarwal A, Singh K, Fouzia Na, GEORGE B, Mehta A, Lashkari H.P, Uppuluri R, REVATHI RAJ, Bartakke S, Gupta K, Sreedharanunni S, Ogura Y, Kato T, Imai K, Chan K-W, Leung D, Ohara O, Nonoyama S, Hershfield M.S, Lau Y.L, Singh S.	Clinical, Immunological, and Molecular Features of Severe Combined Immune Deficiency: A multi-institutional experience from India.	Front Immunol	11	619146	2020
Takeuchi I, Kawai T, Nambu M, Migita O, Yoshimura S, Nishimura K, Yoshioka T, Ogura M, Kyodo R, Shimizu H, Ito S, Kato M, Onodera M, Hata K, Matsubara Y, Arai K.	X-linked inhibitor of apoptosis protein deficiency complicated with Crohn's disease-like enterocolitis and Takayasu arteritis: A case report.	Clin Immunol	217	108495	2020

Osumi T, Yoshimura S, Sako M, Uchiyama T, Ishikawa T, Kawai T, Inoue E, Takimoto T, Takeuchi I, Yamada M, Sakamoto K, Yoshida K, Kimura Y, Matsukawa Y, Matsumoto K, Imadome KI, Arai K, Deguchi T, Imai K, Yuza Y, Matsumoto K, Onodera M, Kanegane H, Tomizawa D, Kato M.	A prospective study of allogeneic hematopoietic stem cell transplantation with post-transplantation cyclophosphamide and anti-thymoglobulin from HLA-mismatched related donors for non-malignant diseases. Biol Blood Marrow Transplant, 26: e286-e291, 2020.	Biol Blood Marrow Transplant	26	e286-e291	2020
Yamaguchi T, Uchida E, Okada T, Ozawa K, Onodera M, Kume A, Shimada T, Takahashi S, Tani K, Nasu Y, Mashimo T, Mizuguchi H, Mitani K, Maki K.	Aspects of gene therapy products using current genome-editing technology in Japan.	Hum Gene Ther	31(19-20)	1043-1053	2020
Ishikawa T, Okai M, Mochizuki E, Uchiyama T, Onodera M, Kawai T.	BCG infections at high frequency in both AR-CGD and X-CGD patients following BCG vaccination.	Clin Infect Dis			2020
Kataura T, Tashiro E, Nishikawa S, Shibahara K, Muraoka Y, Miura M, Sakai S, Katoh N, Totsuka M, Onodera M, Shin-Ya K, Miyamoto K, Sasazawa Y, Hattori N, Saiki S, Imoto M.	A chemical genomics-aggrephagy integrated method studying functional analysis of autophagy inducers.	Autophagy	7	1-17	2020
Oka Y, Hamada M, Nakazawa Y, Muramatsu H, Okuno Y, Higasa K, Shimada M, Takeshima H, Hanada K, Hirano T, Kawakita T, Sakaguchi H, Ichimura T, Ozono S, Yuge K, Watanabe Y, Kotani Y, Yamane M, Kasugai Y, Tanaka M, Suganami T, Nakada S, Mitsutake N, Hara Y, Kato K, Mizuno S, Miyake N, Kawai Y, Tokunaga K, Nagasaki M, Kito S, Isoyama K, Onodera M, Kaneko H, Matsumoto N, Matsuda F, Matsuo K, Takahashi Y, Mashimo T, Kojima S, Ogi T.	Digenic mutations in ALDH2 and ADH5 impair formaldehyde clearance and cause a multisystem disorder, AMeD syndrome.	Sci Adv	6(51)	eabd7197	2020
Uchiyama T, Kawakami S, Masuda H, Yoshida K, Niizeki H, Mochizuki E, Edasawa K, Ishiguro A, Onodera M.	A Distinct Feature of T Cell Subpopulations in a Patient with CHARGE Syndrome and Omenn Syndrome.	J Clin Immunol	41(1)	233-237	2020
Yamazaki E, Kikuchi K, Sasahara Y, Kono M, Akiyama M, Aiba S.	Atopic dermatitis without serum IgE elevation or loss-of-function filaggrin gene mutation in a patient with X-linked agammaglobulinemia.	J Dermatol	47(1)	58-60	2020
Ogata M, Uchida N, Fukuda T, Ikegami K, Kamimura T, Onizuka M, Kobayashi H, Sasahara Y, Sawa M, Sawada A, Hasegawa D, Masuko M, Miyamoto T, Okamoto S.	Clinical practice recommendation for the diagnosis and management of HHV-6 infection after allogeneic hematopoietic cell transplantation: the Japan Society of Hematopoietic Cell Transplantation.	Bone Marrow Transplant	55	1004-1013	2020
Nochi T, Suzuki S, Ito S, Morita S, Furukawa M, Fuchimoto D, Sasahara Y, Usami K, Niimi K, Kitago M, Matsuda S, Matsuo A, Suyama Y, Sakai Y, Wu G, Bazer FW, Watanabe K, Onishi A, Aso H.	Elucidation of the effects of a current X-SCID therapy on intestinal lymphoid organogenesis using an in vivo animal model.	Cell Mol Gastroenterol Hepatol	10	83-100	2020
Moriya K, Suzuki T, Uchida N, Nakano T, Katayama S, Irie M, Rikiishi T, Niizuma H, Okada S, Imai K, Sasahara Y, Kure S.	Ruxolitinib treatment of a patient with steroid-dependent severe autoimmunity due to STAT1 gain-of-function mutation.	Int J Hematol	112	258-262	2020
Moriya K, Kadowaki S, Nakano T, Kutukculer N, Aksu G, Sasahara Y, Kure S, Ohnishi H, Jean-Laurent Casanova JL, Puel A, Fukao T.	The IL1RN mutation creating the most-upstream premature stop codon is hypomorphic because of a reinitiation of translation.	J Clin Immunol	40	463-465	2020

Uchida T, Suzuki T, Kikuchi A, Kakuta H, Ishige T, Nakayama Y, Kanegane H, Etani Y, Mizuochi T, Fujiwara S, Nambu R, Suyama K, Tanaka M, Yoden A, Abukawa D, Sasahara Y, Kure S.	Comprehensive targeted sequencing identifies monogenic disorders in patients with early-onset refractory diarrhea.	J Pediatr Gastroenterol Nutr	71	333-339	2020
Shinoda Y, Hori T, Sasai H, Ikebe T, Ohnishi H.	Neonatal bacteremia caused by emm type 80 group A Streptococcus: A case report.	Pediatr Int.	62(11)	1305-1306	2020
Nozawa A, Ozeki M, Yasue S, Endo S, Kadowaki T, Ohnishi H, Muramatsu H, Hama A, Takahashi Y, Kojima S, Fukao T.	Myelodysplastic syndromes in a pediatric patient with Cri du Chat syndrome with a ring chromosome 5.	Int J Hematol.	112(5)	728-733	2020
Fujii K, Takahashi T, Matsuyama K, Fujii A, Mizutani Y, Ohnishi H, Seishima M.	Impetigo herpetiformis with a CARD14 Thr79Ile variant successfully treated with granulocyte and monocyte adsorption apheresis.	J Dermatol.	47(3)	e84-e85.	2020
Kadowaki T, Ohnishi H, Kawamoto N, Kadowaki S, Hori T, Nishimura K, Kobayashi C, Shigemura T, Ogata S, Inoue Y, Hiejima E, Izawa K, Matsubayashi T, Matsumoto K, Imai K, Nishikomori R, Ito S, Kanegane H, Fukao T.	Immunophenotyping of A20 haploinsufficiency by multicolor flow cytometry.	Clin Immunol.	216	108441	2020
Mizutani Y, Mizutani YH, Matsuyama K, Kawamura M, Fujii A, Shu E, Ohnishi H, Seishima M.	Generalized pustular psoriasis in pregnancy, successfully treated with certolizumab pegol.	J Dermatol.	47	e262-e263.	2020
Dingler FA, Wang M, Mu A, Millington CL, Oberbeck N, Watcham S, Pontel LB, Kamimae-Lanning AN, Langevin F, Nadler C, Cordell RL, Monks PS, Yu R, Wilson NK, Hira A, Yoshida K, Mori M, Okamoto Y, Okuno Y, Muramatsu H, Shiraishi Y, Kobayashi M, Moriguchi T, Osumi T, Kato M, Miyano S, Ito E, Kojima S, Yabe H, Yabe M, Matsuo K, Ogawa S, Göttgens B, Hodskinson MRG, Takata M, Patel KJ.	Two Aldehyde Clearance Systems Are Essential to Prevent Lethal Formaldehyde Accumulation in Mice and Humans.	Mol Cell	80	996-1012	2020 Dec
Okada S, Asano T, Moriya K, Boisson-Dupuis S, Kobayashi M, Casanova JL, Puel A.	Human STAT1 gain-of-function heterozygous mutations: chronic mucocutaneous candidiasis and type I interferonopathy.	J Clin Immunol.	40	1065-1081	2020
Tamaura M, Satoh-Takayama N, Tsumura M, Sasaki T, Goda S, Kageyama T, Hayakawa S, Kimura S, Asano T, Nakayama M, Koseki H, Ohara O, Okada S, Ohno H, Kobayashi M.	Human Gain-of-Function STAT1 Mutation disturbs IL-17 Immunity in Mice	Int Immunol	32	259-272	2020
Goda S, Hayakawa S, Karakawa S, Okada S, Kawaguchi H, Kobayashi M.	Possible involvement of regulatory T cell abnormalities and variational usage of TCR repertoire in children with autoimmune neutropenia.	Clin Exp Immunol.	204	1-13	2020
Yasumura J, Shimizu M, Toma T, Yashiro M, Yachie A, Okada S.	Clinical significance of serum soluble TNF receptor I/II ratio for the differential diagnosis of tumor necrosis factor receptor-associated periodic syndrome from other autoinflammatory diseases	Front Immunol.	14	576152	2020
Nakamura-Utsunomiya A, Tsumura M, Okada S, Kawaguchi H, Kobayashi M.	Downregulation of endothelial nitric oxide synthase (eNOS) and endothelin-1 (ET-1) in a co-culture system with human stimulated X-linked CGD neutrophils.	PLoS One.	15	e0230665	2020

Shimomura M, Doi T, Nishimura S, Imanaka Y, Karakawa S, Okada S, Kawaguchi H, Kobayashi M.	Successful allogeneic bone marrow transplantation using immunosuppressive conditioning regimen for a patient with red blood cell transfusion-dependent pyruvate kinase deficiency anemia.	Hematol Rep.	12	8305	2020
Acker KP, Borlack R, Iuga A, Remotti HE, Soderquist CR, Okada S, Tsumura M, Casanova JL, Picoraro J, Puel A, Kinberg S, Demirdag Y.	Ruxolitinib Response in an Infant With Very-early-onset Inflammatory Bowel Disease and Gain-of-function STAT1 Mutation.	J Pediatr Gastroenterol Nutr.	71	e132-e133.	2020
Nemoto K, Kawanami T, Hoshina T, Ishimura M, Yamasaki K, Okada S, Kanegane H, Yatera K, Kusuhara K	Impaired B-cell differentiation in a patient with STAT1 gain-of-function mutation.	Front Immunol.	11	557521	2020
Miyazono Y, Arai J, Kanai Y, Hitaka D, Kajikawa D, Takeuchi S, Nagafuji M, Fujiyama S, Saito M, Takada H.	Nationwide survey of late-onset hemolysis in very low birthweight infants.	Pediatr Int	doi: 10.1111/ped.14493		2020
Ishikawa M, Tada Y, Tanaka H, Morii W, Inaba M, Takada H, Mori T, Noguchi E.	A family with Gitelman syndrome with asymptomatic phenotypes while carrying reported SLC12A3 mutations.	Case Rep Nephrol Dial	13;10	71-78	2020
Takeshita Y, Ohto T, Enokizono T, Tanaka M, Suzuki H, Fukushima H, Uehara T, Takenouchi T, Kosaki K, Takada H.	Novel ARX mutation identified in infantile spasm syndrome patient.	Hum Genome Var.	31;7:9		2020
Kido T, Iwagami M, Yasunaga H, Abe T, Enomoto Y, Matsui H, Fushimi K, Takada H, Tamiya N.	Outcomes of paediatric out-of-hospital cardiac arrest according to hospital characteristic defined by the annual number of paediatric patients with invasive mechanical ventilation: A nationwide study in Japan.	Resuscitation.	1;148	49-56	2020
Yokoyama K, Horiuchi T, Hashimura C, Yoshida A	A novel C1 inhibitor gene mutation in a family with hereditary angioedema: Use of genetic analysis to facilitate early diagnosis.	Allergol. Int.	69	148-9	2020
Nakajima D, Kawashima Y, Shibata H, Yasumi T, Isa M, Izawa K, Nishikomori R, Heike T, Ohara O.	Simple and Sensitive Analysis for Dried Blood Spot Proteins by Sodium Carbonate Precipitation for Clinical Proteomics.	J Proteome Res.	19	2821-2827	2020

Bastard P, Rosen LB, Zhang Q, Michailidis E, Hoffmann HH, Zhang Y, Dorgham K, Philippot Q, Rosain J, Béziat V, Manry J, Shaw E, Haljasmägi L, Peterson P, Lorenzo L, Bizien L, Trouillet-Assant S, Dobbs K, de Jesus AA, Belot A, Kallaste A, Catherinot E, Tandjaoui-Lambiotte Y, Le Pen J, Kerner G, Bigio B, Seeleuthner Y, Yang R, Bolze A, Spaan AN, Delmonte OM, Abers MS, Aiuti A, Casari G, Lampasona V, Piemonti L, Ciceri F, Bilguvar K, Lifton RP, Vasse M, Smadja DM, Migaud M, Hadjadj J, Terrier B, Duffy D, Quintana-Murci L, van de Beek D, Roussel L, Vinh DC, Tangye SG, Haerynck F, Dalmau D, Martinez-Picado J, Brodin P, Nussenzweig MC, Boisson-Dupuis S, Rodríguez-Gallego C, Vogt G, Mogensen TH, Oler AJ, Gu J, Burbelo PD, Cohen JI, Biondi A, Bettini LR, D'Angio M, Bonfanti P, Rossignol P, Mayaux J, Rieux-Laucat F, Husebye ES, Fusco F, Ursini MV, Imberti L, Sottini A, Paghera S, Quiros-Roldan E, Rossi C, Castagnoli R, Montagna D, Licari A, Marseglia GL, Duval X, Ghosn J; HGID Lab; NIAID-USUHS Immune Response to COVID Group; COVID Clinicians; COVID-STORM Clinicians; Imagine COVID Group; French COVID Cohort Study Group; Milieu Intérieur Consortium; CoV-Contact Cohort; Amsterdam UMC Covid-19 Biobank; COVID Human Genetic Effort, Tsang JS, Goldbach-Mansky R, Kisand K, Lionakis MS, Puel A, Zhang SY, Holland SM, Gorochov G, Jouanguy E, Rice CM, Cobat A, Notarangelo LD, Abel L, Su HC, Casanova JL.	Autoantibodies against type I IFNs in patients with life-threatening COVID-19.	Science.	370(6515)	eabd4585	2020
Zhang Q, Bastard P, Liu Z, Le Pen J, Moncada-Velez M, Chen J, Ogishi M, Sabli IKD, Hodeib S, Korol C, Rosain J, Bilguvar K, Ye J, Bolze A, Bigio B, Yang R, Arias AA, Zhou Q, Zhang Y, Onodi F, Korniotis S, Karpf L, Philippot Q, Chbihi M, Bonnet-Madin L, Dorgham K, Smith N, Schneider WM, Razooky BS, Hoffmann HH, Michailidis E, Moens L, Han JE, Lorenzo L, Bizien L, Meade P, Neehus AL, Ugurbil AC, Corneau A, Kerner G, Zhang P, Rapaport F, Seeleuthner Y, Manry J, Masson C, Schmitt Y, Schlüter A, Le Voyer T, Khan T, Li J, Fellay J, Roussel L, Shahrooei M, Alosaimi MF, Mansouri D, Al-Saud H, Al-Mulla F, Almourfi F, Al-Muhsen SZ, Alsohime F, Al Turki S, Hasanato R, van de Beek D, Biondi A, Bettini LR, D'Angio' M, Bonfanti P, Imberti L, Sottini A, Paghera S, Quiros-Roldan E, Rossi C, Oler AJ, Tompkins MF, Alba C, Vandernoot I, Goffard JC, Smits G, Migeotte I, Haerynck F, Soler-Palacin P, Martin-Nalda A, Colobran R, Morange PE, Keles S, Çölkesen F, Ozcelik T, Yasar KK, Senoglu S, Karabela ŞN, Rodríguez-Gallego C, Novelli G, Hraiech S, Tandjaoui-Lambiotte Y, Duval X, Laouénan C; COVID-STORM Clinicians; COVID Clinicians; Imagine COVID Group; French COVID Cohort Study Group; CoV-Contact Cohort; Amsterdam UMC Covid-19 Biobank; COVID Human Genetic Effort; NIAID-USUHS/TAGC COVID Immunity Group, Snow AL, Dalgard CL, Milner JD, Vinh DC, Mogensen TH, Marr N, Spaan AN, Boisson B, Boisson-Dupuis S, Bustamante J, Puel A, Ciancanelli MJ, Meyts I, Maniatis T, Soumelis V, Amara A, Nussenzweig M, García-Sastre A, Krammer F, Pujol A, Duffy D, Lifton RP, Zhang SY, Gorochov G, Béziat V, Jouanguy E, Sancho-Shimizu V, Rice CM, Abel L, Notarangelo LD, Cobat A, Su HC, Casanova JL.	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19.	Science.	370(6515)	eabd4570	2020
Inoue K, Sasaki S, Yasumi T, Imai K, Kusunoki T, Morio T, Kanegane H.	Helicobacter cinaedi-Associated refractory cellulitis in patients with X-linked agammaglobulinemia. Helicobacter cinaedi-Associated refractory cellulitis in patients with X-linked agammaglobulinemia.	J Clin Immunol	40	1132-1137	2020
Matsumoto K, Hoshino A, Nishimura A, Kato T, Mori Y, Shimomura M, Naito C, Watanabe K, Hamazaki M, Mitsuiki N, Takagi M, Imai K, Nonoyama S, Kanegane H, Morio T.	DNA ligase IV deficiency identified by chance following vaccine-derived rubella virus infection.	J Clin Immunol.	40	1187-1190	2020
Yanagimachi M, Kato K, Iguchi A, Sasaki K, Kiyotani C, Koh K, Koike T, Sano H, Shigemura T, Muramatsu H, Okada K, Inoue M, Tabuchi K, Nishimura T, Mizukami T, Nunoi H, Imai K, Kobayashi M, Morio T.	Hematopoietic cell transplantation for chronic granulomatous disease in Japan.	Front Immunol.	11	1617	2020

Sakata S, Tsumura M, Matsubayashi T, Karakawa S, Kimura S, Tamaura M, Okano T, Naruto T, Mizoguchi Y, Kagawa R, Nishimura S, Imai K, Le Voyer T, Casanova JL, Bustamante J, Morio T, Ohara O, Kobayashi M, Okada S.	Autosomal recessive complete STAT1 deficiency caused by compound heterozygous intronic mutations.	Int Immunol	32	663-671	2020
Okano T, Imai K, Naruto T, Okada S, Yamashita M, Yeh TW, Ono S, Tanaka K, Okamoto K, Tanita K, Matsumoto K, Toyofuku E, Kumaki-Matsumoto E, Okamura M, Ueno H, Ogawa S, Ohara O, Takagi M, Kanegane H, Morio T.	Whole-Exome Sequencing-Based aroach for germline mutations in patients with inborn errors of immunity.	J Clin Immunol.	40	729-740	2020
Inoue K, Miura H, Hoshino A, Kamiya T, Tanita K, Ohye T, Park MJ, Yanagimachi M, Takagi M, Imai K, Morio T, Yoshikawa T, Kanegane H.	Inherited chromosomally integrated human herpesvirus-6 in a patient with XIAP deficiency.	Transpl Infect Dis.	22	e13331	2020
Yeh TW, Okano T, Naruto T, Yamashita M, Okamura M, Tanita K, Du L, Pan-Hammarström Q, Mitsuiki N, Okada S, Kanegane H, Imai K, Morio T.	APRIL-dependent life-long plasmacyte maintenance and immunoglobulin production in humans.	J Allergy Clin Immunol.	146	1109-1120	2020
Mori M, Hira A, Yoshida K, Muramatsu H, Okuno Y, Shiraishi Y, Anmae M, Yasuda J, Tadaka S, Kinoshita K, Osumi T, Noguchi Y, Adachi S, Kobayashi R, Kawabata H, Imai K, Morio T, Tamura K, Takaori-Kondo A, Yamamoto M, Miyano S, Kojima S, Ito E, Ogawa S, Matsuo K, Yabe H, Yabe M, Takata M.	Pathogenic mutations identified by a multimodality approach in 117 Japanese fanconi anemia patients.	Haematologica.	105	1166-1167	2020
Hoshino A, Nishimura A, Naruto T, Okano T, Matsumoto K, Okamoto K, Shintaku H, Tokoro S, Okamoto H, Wada T, Takagi M, Imai K, Kanegane H, Morio T.	High-throughput analysis revealed the unique immunoglobulin gene rearrangements in Plasmacytoma-like Post-transplant lymphoproliferative disorder.	Br J Haematol.	189	e164-e168	2020
Umeda K, Imai K, Yanagimachi M, Yabe H, Kobayashi M, Takahashi Y, Kajiwara M, Yoshida N, Cho Y, Inoue M, Hashii Y, Atsuta Y, Morio T; Inherited Disease Working Group of the Japan Society for Hematopoietic Cell Transplantation.	Impact of Graft-versus-host disease on the clinical outcome of allogeneic hematopoietic stem cell transplantation for Non-Malignant diseases.	Int J Hematol.	111	869-876	2020
Bousfiha A, Jeddane L, Picard C, Al-Herz W, Ailal F, Chatila T, Cunningham-Rundles C, Etzioni A, Franco JL, Holland SM, Klein C, Morio T, Ochs HD, Oksenhendler E, Puck J, Torgerson TR, Casanova JL, Sullivan KE, Tangye SG.	Human inborn errors of immunity: 2019 update of the IUIS phenotypical classification.	J Clin Immunol.	40	66-81	2020
Ureshino H, Koarada S, Kamachi K, Yoshimura M, Yokoo M, Kubota Y, Ando T, Ichinohe T, Morio T, Kimura S.	Immune dysregulation syndrome with de novo CTLA4 germline mutation responsive to abatacept therapy.	Int J Hematol.	111	897-902	2020
Tangye SG, Al-Herz W, Bousfiha A, Chatila T, Cunningham-Rundles C, Etzioni A, Franco JL, Holland SM, Klein C, Morio T, Ochs HD, Oksenhendler E, Picard C, Puck J, Torgerson TR, Casanova JL,Sullivan KE	Human inborn errors of immunity: 2019 update on the classification from the international union of immunological societies expert committee.	J Clin Immunol.	40	24-64	2020
Suzuki T, Kohyama K, Moriyama K, Ozaki M, Hasegawa S, Ueno T, Saitoe M, Morio T, Hayashi M, Sakuma H.	Extracellular ADP augments microglial inflammasome and NF-κB activation via the P2Y12 receptor.	Eur J Immunol.	50	205-219	2020

雑誌（和文）

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
森尾友宏	高IgM症候群、重症好中球減少症、家族性地中海熱	Year Note			2017, 2018, 2019, 2020, 2021.
森尾友宏	第12章　感染症　76麻疹　77風疹	病期・病態・重症度からみた疾患別看護過程+病態関連図 第4版		p1395-p1398,p1407-1409	2020
森尾友宏	第Ⅳ部　原発性免疫不全症候群 1.総論	小児感染免疫学		p566-p578	2020
大西秀典	TIRAP欠損症	日本臨床	78巻 増刊号	360-362	2020
大西秀典	IRAK1欠損症	日本臨床	78巻 増刊号	357-359	2020
門脇朋範、門脇紗織、大西秀典	A20ハプロ不全症	日本臨床	78巻 増刊号	443-446	2020
大西秀典	A20ハプロ不全症	小児リウマチ学		232-235	2020
大西秀典	メンデル遺伝型マイコバクテリア易感染症	小児感染免疫学		717-724	2020
和田泰三	III. 複合免疫不全症 (細胞免疫及び液性免疫の異常) .	日本臨牀	増刊号 78(7)	51-86	2020
今中 雄介, 岡田 賢	ゲノム編集と治療　ゲノム編集技術を活用した重症先天性好中球減少症の治療開発.	医学のあゆみ	273巻9号	835-840	2020
江口 勇太, 土居 岳彦, 野間 康輔, 浅野 孝基, 岡田 賢, 小林 正夫	T+/low B+NK+の表現型を呈した非典型的X連鎖性重症複合免疫不全症.	日本小児科学会雑誌	123巻6号	1009-1014	2020
土居 岳彦, 岡田 賢	リンパ増殖性疾患と原発性免疫不全	臨床血液	61巻9号	1365-1372	2020
野間康輔, 岡田 賢	急性壊死性脳症	日本臨床	8巻増刊7	366-368	2020
藤川 皓基, 岡田 賢	Acute liver failure due to NBAS deficiency.	日本臨床	8巻増刊7	363-365	2020
齋藤 聡志, 岡田 賢	IL-17RC欠損症	日本臨床	8巻増刊7	354-356	2020
坂田 園子, 岡田 賢	DBR1欠損	日本臨床	8巻増刊7	351-353	2020
佐倉 文祥, 岡田 賢	IRF3欠損症	日本臨床	8巻増刊7	348-350	2020
今中 雄介, 岡田 賢	RNA polymerase III欠損症	日本臨床	8巻増刊7	339-341	2020
江藤 昌平, 岡田 賢.	CD16欠損症	日本臨床	8巻増刊7	363-365	2020
郷田 聡, 岡田 賢	IFNAR2欠損症	日本臨床	8巻増刊7	335-338	2020
郷田 聡, 岡田 賢	IFNAR1欠損症	日本臨床	8巻増刊7	331-334	2020
加藤 豊, 岡田 賢	IRF9欠損症	日本臨床	8巻増刊7	328-330	2020
溝口 洋子, 岡田 賢	CIB1欠損症	日本臨床	8巻増刊7	325-327	2020
西村 志帆, 岡田 賢	JAK1欠損症	日本臨床	8巻増刊7	322-324	2020
岡田 賢.	RORγT欠損症	日本臨床	8巻増刊7	319-321	2020
早川 誠一, 岡田 賢	P1104A TYK2 homozygosity	日本臨床	8巻増刊7	316-318	2020
冨岡 啓太, 岡田 賢	SPPL2a欠損症	日本臨床	8巻増刊7	313-315	2020
岡田 賢	内因性あるいは自然免疫の異常概論およびトピックス	日本臨床	8巻増刊7	303-312	2020
岡田 賢	原発性免疫不全症候群　研究の進歩　責任遺伝子探索.	日本臨床	8巻増刊7	21-26	2020
前田豊樹、堀内孝彦	遺伝性血管性浮腫ー多彩な症状を呈する反復性浮腫ー.	Medical Practice	37	148-50	2020
堀内孝彦	内科医が知っておきたい補体関連疾患	日本内科学会雑誌	109	1925-31	2020
玉浦 萌, 岡田 賢	先天性免疫異常	臨床免疫・アレルギー科	75巻1号	63-76	2021

書籍

著者氏名	論文タイトル名	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
塚本浩、堀内孝彦	先天性補体欠損症	一般社団法人日本小児感染症学会編	小児感染免疫学	朝倉書店	東京	2020	pp.744-8
堀内孝彦	遺伝性血管性浮腫	一般社団法人日本小児感染症学会編	小児感染免疫学	朝倉書店	東京	2020	pp.749-57
堀内孝彦	補体異常症．概論およびトピックス		原発性免疫不全症候群　最新の疾患分類と新規疾患を中心に	日本臨牀社	東京	2020	pp.461-7