

研究成果の刊行に関する一覧表

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
小倉浩美, 菅野仁.	【特集 新生児黄疸を再び考える】 各論 先天性溶血性疾患.		周産期医学	東京医学社	東京	2019	211-6
小倉浩美, 菅野仁.	赤血球酵素とその異常. 特集 赤血球		血液フロンティア	医薬ジャーナル社	東京	2018	1333-41
Yabe H, Serafini M, Boelens JJ.	Hematopoietic Stem Cell Transplantation in Mucopolysaccharidoses: The Effects and Limitations.	Tomatsu S, Orii T	Mucopolysaccharidoses update	Nova Science	New York	2018	513-534
伊藤悦朗, 大賀正一, 小原明, 金兼弘和, 唐川修平, 菅野仁, 國島伸治, 小島勢二, 小林正夫, 笹原洋二, 多賀崇, 高田穰, 照井君典, 長谷川大輔, 張替秀郎, 藤原亨, 古山和道, 真部淳, 溝口洋子, 村松秀城, 矢部普正, 山口博樹, 渡邊健一郎.	Diamond-Blackfan貧血 / Fanconi貧血 / 遺伝性鉄芽球性貧血 / Congenital dyserythropoietic anemia / 先天性角化不全症 / Shwachman-Diamond症候群 / 先天性好中球減少症 / 先天性血小板減少症	日本小児血液・がん学会	先天性骨髄不全症診療ガイドライン2017	診断と治療社	東京	2017	4-77
藤原亨, 張替秀郎.	鉄芽球性貧血 疾患概念・病因・病態	谷脇雅史	貧血学 最新の動向・治療動向	日本臨床社	東京	2017	448-452
石合正道, 高田穰.	2.18 放射線応答遺伝子の生物種間の保存・相関	宮川清	放射線医学の事典		東京	2017	
Ito E, Terui K, Toki T.	Inherited bone marrow failure syndrome, TAM.	Eiichi Ishii	In Hematological Disorders in Children.	Springer Natures Singapore Pte Ltd	Shingapore	2017	145-170
Hasegawa D, Manabe A.	Myelodysplastic syndrome and JMML.	Eiichi Ishii	In Hematological Disorders in Children.	Springer Natures Singapore Pte Ltd	Shingapore	2017	87-108
小倉浩美, 菅野仁.	先天性貧血	神田善伸	血液科研修ノート	診断と治療社	東京	2016	462-465

雑誌

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
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			2019 [Epub ahead of print]
Saito K, Fujiwara T, Hatta S, Morita M, Ono K, Suzuki C, Fukuhara N, Onishi Y, Nakamura Y, Kawamata S, Shimizu R, Yamamoto M, Harigae H.	Generation and molecular characterization of human ring sideroblasts: A key role of ferrous iron in erythroid differentiation and ring sideroblast formation.	Mol Cell Biol.			2019 (in press)
Takaoka K, Kawazu M, Koya J, Yoshimi A, Masamoto Y, Maki H, Toya T, Kobayashi T, Nannya Y, Arai S, Ueno H, Suzuki K, Harada H, Manabe A, Hayashi Y, Mano H, Kurokawa M.	A germline HLTF mutation in familial MDS induces DNA damage accumulation through impaired PCNA polyubiquitination.	Leukemia			2019 (in press)
Okamoto Y, James Hejna, Takata M.	Regulation of R-loops and genome instability in Fanconi anemia.	J Biochemistry Tokyo.			2019 (in press)
Ninomiya K, Hata T, Yoshioka H, Ohashi K, Bessho A, Hosokawa S, Ishikawa N, Yamasaki M, Shibayama T, Aoe K, Kozuki T, Harita S, Ueda Y, Murakami T, Fujimoto N, Yanai H, Toyooka S, Takata M, Hotta K, Kiura K.	A prospective cohort study to define the clinical features and outcome of lung cancers harboring HER2 aberration (HER2-CS STUDY) in Japan.	Chest			2019 (in press)
Tachiwada T, Oda K, Tahara M, Sennari K, Nemoto K, Noguchi S, Kawanami T, Kido T, Yamaguchi H, Yatera K.	Fatal Acute Exacerbation of Familial Interstitial Pneumonia Complicated with Dyskeratosis Congenita after Influenza Virus B Infection.	Inter Med.			2019 (in press)
森美奈子, 矢部普正, 矢部みはる, 高田穰.	日本人ファンconi貧血患者のゲノム解析から得られた知見.	臨床血液			2019 (in press)

西村聡, 平林真介, 山本俊亮, 相賀咲央莉, 西谷美佐, 細谷要介, 森慎一郎, 長谷川大輔, 真部淳.	Diamond-Blackfan貧血に対する強度減弱前処置を用いた骨髄移植.	小児血液・がん学会雑誌			2019 (in press)
平林真介, 鈴木美慧, 真部淳.	TP53変異と造血器腫瘍.	臨床血液			2019 (in press)
Usami I, Imamura T, Takahashi Y, Suenobu SI, Hasegawa D, Hashii Y, Deguchi T, Hori T, Shimada A, Kato K, Ito E, Moriya-Saito A, Kawasaki H, Hori H, Yumura-Yagi K, Hara J, Sato A, Horibe K; Japan Association of Childhood Leukemia Study Group (JACLS).	Discontinuation of L-asparaginase and poor response to prednisolone are associated with poor outcome of ETV6-RUNX1-positive pediatric B-cell precursor acute lymphoblastic leukemia.	Int J Hematol.	109(4)	477-482	2019
Kudo K, Tanaka T, Kobayashi A, Terui K, Ito E.	Zoledronic acid for relapsed Langerhans cell histiocytosis with isolated skull bone lesion.	Pediatr Int.	61(3)	315-317	2019
Yabe M, Koike T, Ohtsubo K, Imai E, Morimoto T, Takakura H, Koh K, Yoshida K, Ogawa S, Ito E, Okuno Y, Muramatsu H, Kojima S, Matsuo K, Mori M, Hira A, Takata M, Yabe H.	Associations of complementation group, ALDH2 genotype, and clonal abnormalities with hematological outcome in Japanese patients with Fanconi anemia.	Ann Hematol.	98(2)	271-280	2019
Yagi H, Kageyama K, Kinoshita N, Niioka K, Yamagata S, Ito E, Daimon M.	Relaxin-3 regulates corticotropin-releasing factor gene expression in cultured rat hypothalamic 4B cells.	Neurosci Lett.	692	137-142	2019
Ochi T, Onishi Y, Nasu K, Onodera K, Kobayashi M, Ichikawa S, Fujiwara T, Fukuhara N, Yamada-Fujiwara M, Harigae H.	Umbilical Cord Blood Transplantation Using Reduced-intensity Conditioning without Antithymocyte Globulin in Adult Patients with Severe Aplastic Anemia.	Biol Blood Marrow Transplant.	25	e55-59	2019
Fujiwara T, Harigae H.	Molecular pathophysiology and genetic mutations in congenital sideroblastic anemia.	Free Radic Biol Med.	133	179-185	2019
Ono R, Watanabe T, Kawakami E, Iwasaki M, Tomizawa-Murasawa M, Matsuda M, Najima Y, Takagi S, Fujiki S, Sato R, Mochizuki Y, Yoshida H, Sato K, Yabe H, Kato S, Saito Y, Taniguchi S, Shultz LD, Ohara O, Amagai M, Koseki H, Ishikawa F.	Co-activation of macrophages and T cells contribute to chronic GVHD in human IL-6 transgenic humanised mouse model.	EBioMedicine	41	584-596	2019

Miyamura K, Yamashita T, Atsuta Y, Ichinohe T, Kato K, Uchida N, Fukuda T, Ohashi K, Ogawa H, Eto T, Inoue M, Takahashi S, Mori T, Kanamori H, Yabe H, Hama A, Okamoto S, Inamoto Y.	High probability of follow-up termination among AYA survivors after allogeneic hematopoietic cell transplantation.	Blood Adv.	3(3)	397-405	2019
Bianchi P, Elisa Fermo E, Glader B, Kanno H, Agarwal A, Barcellini W, Eber S, Hoyer JD, Kuter DJ, Maia TM, Del Mar Mañu-Pereira M, Kalfa TA, Pissard S, Segovia JC, van Beers E, Gallagher PG, Rees DC, van Wijk R.	Addressing The Diagnostic Gaps in Pyruvate Kinase (PK) Deficiency: Consensus Recommendations on the Diagnosis of PK Deficiency.	Am J Hematol.	94	149-161	2019
Kamio H, Uchiyama T, Kanno H, Onoe Y, Saito K, Kameoka S, Kamio T, Okamoto T. Association between SLCO1B1 rs4149056 and tegafur-uracil-induced hepatic dysfunction in breast cancer.	Association between SLCO1B1 rs4149056 and tegafur-uracil-induced hepatic dysfunction in breast cancer.	Pharmacogenomics	20(5)	353-365	2019
Kohara H, Utsugisawa T, Sakamoto C, Hirose L, Ogawa Y, Ogura H, Sugawara A, Liao J, Aoki T, Iwasaki T, Asai T, Doisaki S, Okuno Y, Muramatsu H, Abe T, Kurita R, Miyamoto S, Sakuma T, Shiba M, Yamamoto T, Ohga S, Yoshida K, Ogawa S, Ito E, Kojima S, Kanno H, Tani K.	KLF1 mutation E325K induces cell cycle arrest in erythroid cells differentiated from congenital dyserythropoietic anemia patient-specific induced pluripotent stem cells.	Exp Hematol.	73	25-37.e8	2019
Hayano S, Okuno Y, Tsutsumi M, Inagaki H, Fukasawa Y, Kurahashi H, Kojima S, Takahashi Y, Kato T.	Frequent intragenic microdeletions of elastin in familial supravalvular aortic stenosis.	Int J Cardiol.	274	290-295	2018
Uemura S, Mori T, Nagano C, Takafuji S, Nishimura N, Toki T, Terui K, Ito E, Iijima K.	Effective response to azacitidine in a child with a second relapse of myeloid leukemia associated with Down syndrome after bone marrow transplantation.	Pediatr Blood Cancer.		e27414	2018
Kudo K, Ueno H, Sato T, Kubo K, Kanazaki R, Kobayashi A, Kamio T, Sasaki S, Terui K, Kurose A, Yoshida K, Shiozawa Y, Toki T, Ogawa S, Ito E.	Two siblings with familial neuroblastoma with distinct clinical phenotypes harboring an ALK germline mutation.	Genes Chromosomes Cancer	57(12)	665-669	2018

Toki T, Yoshida K, Wang R, Nakamura S, Maekawa T, Goi K, Katoh MC, Mizuno S, Sugiyama F, Kanezaki R, Uechi T, Nakajima Y, Sato Y, Okuno Y, Sato-Otsubo A, Shiozawa Y, Kataoka K, Shiraishi Y, Sanada M, Chiba K, Tanaka H, Terui K, Sato T, Kamio T, Sakaguchi H, Ohga S, Kuramitsu M, Hamaguchi I, Ohara A, Kanno H, Miyano S, Kojima S, Ishiguro A, Sugita K, Kenmochi N, Takahashi S, Eto K, Ogawa S, Ito E.	De Novo Mutations Activating Germline TP53 in an Inherited Bone-Marrow-Failure Syndrome.	Am J Hum Genet.	103(3)	440-447	2018
Tsujimoto S, Osumi T, Uchiyama M, Shirai R, Moriyama T, Nishii R, Yamada Y, Kudo K, Sekiguchi M, Arakawa Y, Yoshida M, Uchiyama T, Terui K, Ito S, Koh K, Takita J, Ito E, Tomizawa D, Manabe A, Kiyokawa N, Yang JJ, Kato M.	Diploidy analysis of NUDT15 variants and 6-mercaptopurine sensitivity in pediatric lymphoid neoplasms.	Leukemia	32(12)	2710-2714	2018
Sonoda M, Ishimura M, Ichimiya Y, Terashi E, Eguchi K, Sakai Y, Takada H, Hama A, Kanno H, Toki T, Ito E, Ohga S.	Atypical erythroblastosis in a patient with Diamond-Blackfan anemia who developed del (20q) myelodysplasia.	Int J Hematol.	108(2)	228-231	2018
Kuwahara K, Kudo K, Yashima-Abo A, Katayama K, Kojima K, Tone K, Ito E, Nakazawa A, Iwafuchi H, Kurose A.	Classic Hodgkin lymphoma with osseous involvement mimicking Langerhans cell histiocytosis in a child.	Hum Pathol.	77	147-151	2018
Kato H, Itoh-Nakadai A, Matsumoto M, Ishii Y, Watanabe-Matsui M, Ikeda M, Ebina-Shibuya R, Sato Y, Kobayashi M, Nishizawa H, Suzuki K, Muto A, Fujiwara T, Nannya Y, Cazzola M, Ogawa S, Harigae H, Igarashi K.	Infection perturbs Bach2- and Bach1-dependent erythroid lineage choice to cause anemia.	Nat Immunol.	19	1059-1070	2018
Ishida H, Imamura T, Morimoto A, Fujiwara T, Harigae H.	Five-aminolevulinic acid: New Approach for Congenital Sideroblastic Anemia.	Pediatr Int.	60	496-497	2018

Kawakami T, Nakazawa H, Kawakami F, Matsuzawa S, Sudo Y, Sakai H, Nishina S, Sendo N, Sendo Y, Komatsu M, Umemura T, Yamaguti T, Kosho T, Fujiwara T, Harigae H, Ishida F.	Successful treatment with vitamin B6 for X-linked sideroblastic anemia with ALAS2 R452H mutation.	Rinsho Ketsueki	29	401-406	2018
Hatta S, Fujiwara T, Yamamoto T, Saito K, Kamata M, Tamai Y, Kawamata S, Harigae H.	A defined culture method enabling the establishment of ring sideroblasts from induced pluripotent cells of X-linked sideroblastic anemia.	Haematologica	244	e188-e191	2018
Ohashi K, Fujiwara T, Onodera K, Saito Y, Ichikawa S, Kobayashi M, Okitsu Y, Fukuhara N, Onishi Y, Harigae H.	Establishment of a screening system to identify novel GATA-2 transcriptional regulators.	Tohoku J Exp Med.	44	41-52	2018
Umeda K, Yabe H, Kato K, Imai K, Kobayashi M, Takahashi Y, Yoshida N, Sato M, Sasahara Y, Kato K, Adachi S, Koga Y, Okada K, Inoue M, Hashii Y, Atsuta Y, Morio T; Inherited Disease Working Group of the Japan Society for Hematopoietic Cell Transplantation.	Impact of low-dose irradiation and in vivo T-cell depletion on hematopoietic stem cell transplantation for non-malignant diseases using fludarabine-based reduced-intensity conditioning.	Bone Marrow Transplant.			2018 [Epub ahead of print]
Yakushijin K, Ikezoe T, Ohwada C, Kudo K, Okamura H, Goto H, Yabe H, Yasumoto A, Kuwabara H, Fujii S, Kagawa K, Ogata M, Onishi Y, Kohno A, Watamoto K, Uoshima N, Nakamura D, Ota S, Ueda Y, Oyake T, Koike K, Mizuno I, Iida H, Katayama Y, Ago H, Kato K, Okamura A, Kikuta A, Fukuda T.	Clinical effects of recombinant thrombomodulin and defibrotide on sinusoidal obstruction syndrome after allogeneic hematopoietic stem cell transplantation.	Bone Marrow Transplant.			2018 [Epub ahead of print]
Inamoto Y, Matsuda T, Tabuchi K, Kurosawa S, Nakasone H, Nishimori H, Yamasaki S, Doki N, Iwato K, Mori T, Takahashi S, Yabe H, Kohno A, Nakamae H, Sakura T, Hashimoto H, Sugita J, Ago H, Fukuda T, Ichinohe T, Atsuta Y, Yamashita T; Japan Society for Hematopoietic Cell Transplantation Late Effects and Quality of Life Working Group.	Outcomes of patients who developed subsequent solid cancer after hematopoietic cell transplantation.	Blood Adv.	2(15)	1901-1913	2018

Nakasone H, Tabuchi K, Uchida N, Ohno Y, Matsuhashi Y, Takahashi S, Onishi Y, Onizuka M, Kobayashi H, Fukuda T, Ichinohe T, Takanashi M, Kato K, Atsuta Y, Yabe H, Kanda Y.	Which is more important for the selection of cord blood units for haematopoietic cell transplantation: the number of CD34-positive cells or total nucleated cells?	Br J Haematol.				2018 [Epub ahead of print]
Hamada M, Doisaki S, Okuno Y, Muramatsu H, Hama A, Kawashima N, Narita A, Nishio N, Yoshida K, Kanno H, Manabe A, Taga T, Takahashi Y, Miyano S, Ogawa S, Kojima S.	Whole-exome analysis to detect congenital hemolytic anemia mimicking congenital dyserythropoietic anemia.	Int J Hematol.	108(3)	306-311		2018
Kato K, Miya F, Hamada N, Negishi Y, Narumi-Kishimoto Y, Ozawa H, Ito H, Hori I, Hattori A, Okamoto N, Kato M, Tsunoda T, Kanemura Y, Kosaki K, Takahashi Y, Nagata KI, Saitoh S.	MYCN de novo gain-of-function mutation in a patient with a novel megalencephaly syndrome.	J Med Genet.				2018 [Epub ahead of print]
Takeichi T, Okuno Y, Kawamoto A, Inoue T, Nagamoto E, Murase C, Shimizu E, Tanaka K, Kageshita Y, Fukushima S, Kono M, Ishikawa J, Ihn H, Takahashi Y, Akiyama M.	Reduction of stratum corneum ceramides in Neu-Laxova syndrome caused by phosphoglycerate dehydrogenase deficiency.	J Lipid Res.	59(12)	2413-2420		2018
Kamatani N, Furihata K, Taniguchi A, Fukuuchi T, Yamaoka N, Kaneko K, Kanno H.	In vitro enhancement of ATP in human erythrocytes from a healthy subject and two patients with thalassemia and hemoglobinopathy.	Gout and Nucleic Acid Metabolism	42(1)	59-64		2018
井島廣子, 古賀正史, 杉山正悟, 小倉浩美, 菅野仁, 陣内秀昭.	HbA1cが偽低値をしめしたエノラーゼ、異常症合併2型糖尿病の1例.	Diabetes Journal	46(1)	30-34		2018
van Straaten S, Bierings M, Bianchi P, Akiyoshi K, Kanno H, Serra IB, Chen J, Huang X, van Beers E, Ekwattanakit S, Gungör T, Kors WA, Smiers F, Raymakers R, Yanez L, Sevilla J, van Solinge W, Segovia JC, van Wijk R.	Worldwide study of hematopoietic allogeneic stem cell transplantation in pyruvate kinase deficiency.	Haematologica	103(2)	e82-e86		2018
槍澤大樹, 小林良輔, 磯合綾子, 小野寺博和, 松野義弘, 加藤道夫, 菅野仁.	自己アルブミン製剤としての濾過濃縮腹水の有効性.	日本輸血細胞治療学会誌	645	631-640		2018

Abe T, Ooka M, Kawasumi R, Miyata K, Takata M, Hirota K, Branzei D.	Warsaw Breakage Syndrome DDX11 helicase acts jointly with RAD17 in the repair of bulky lesions and replication through abasic sites.	Proc Natl Acad Sci U S A.	115(33)	8412-8417	2018
Yabe M, Koike T, Ohtsubo K, Imai E, Morimoto T, Takakura H, Koh K, Yoshida K, Ogawa S, Ito E, Okuno Y, Muramatsu H, Kojima S, Matsuo K, Mori M, Hira A, Takata M and Yabe H. Associations of complementation group.	ALDH2 genotype, and clonal abnormalities with hematological outcome in Japanese patients with Fanconi anemia.	Ann Hematol.	98(2)	271-280	2018
Higgs MR, Sato K, Reynolds JJ, Begum S, Bayley R, Goula A, Vernet A, Paquin KL, Skalnik DG, Kobayashi W, Takata M, Howlett NG, Kurumizaka H, Kimura H, Stewart GS.	Histone Methylation by SETD1A Protects Nascent DNA through the Nucleosome Chaperone Activity of FANCD2.	Mol Cell.	71(1)	25-41.e6	2018
Okamoto Y, Iwasaki WM, Kugou K, Takahashi KK, Oda A, Sato K, Kobayashi W, Kawai H, Sakasai R, Takaori-Kondo A, Yamamoto T, Kanemaki MT, Taoka M, Isobe T, Kurumizaka H, Innan H, Ohta K, Ishiai M, Takata	Replication stress induces accumulation of FANCD2 at central region of large fragile genes.	Nucleic Acids Res.	46(6)	2932-2944	2018
高田穰.	ファンコニ貧血の新規原因遺伝子RFWD3の同定とその機能解析.	医学の歩み	TOPICS 266(6/7)	545-546	2018
稲野将二郎, 高田穰.	ファンコニ貧血の新規原因遺伝子RFWD3/FANCD2の機能解析から明らかになった相同組換え反応制御機構.	生化学	90(3)	371-380	2018
Eguchi K, Ishimura M, Sonoda M, Ono H, Shiraishi A, Kanno S, Koga Y, Takada H, Ohga S.	Nontuberculous mycobacteria-associated hemophagocytic lymphohistiocytosis in MonoMAC syndrome.	Pediatr Blood Cancer.	65(7)	e27017	2018
大賀正一, 石村匡崇, 槍澤大樹, 菅野仁.	新生児の遺伝性溶血性貧血～疾患概念の拡張～.	日本産婦人科・新生児血液学会誌	27(2)	41-47	2018
Minakawa S, Matsuzaki Y, Terui K, Kayaba H, Sawamura D.	Tuberculous granuloma developed 9 years after bacillus Calmette-Guérin vaccination in a patient with immunodeficiency.	J Dermatol.	45	e293-5	2018
照井君典.	貧血. 小児疾患の診断治療基準.	小児内科増刊号	50	70-1	2018

Kaneko K, Kubota Y, Nomura K, Hayashimoto H, Chida T, Yoshino N, Wayama M, Ogasawara K, Nakamura Y, Tooyama I, and Furuyama K.	Establishment of a cell model of X-linked sideroblastic anemia using genome editing.	Exp Hematol.	65	57-68.e2	2018
Shimada A, Iijima-Yamashita Y, Tawa A, Tomizawa D, Yamada M, Norio S, Watanabe T, Taga T, Iwamoto S, Terui K, Moritake H, Kinoshita A, Takahashi H, Nakayama H, Koh K, Goto H, Kosaka Y, Saito AM, Kiyokawa N, Horibe K, Hara Y, Oki K, Hayashi Y, Tanaka S, Adachi S.	Risk-stratified therapy for children with FLT3-ITD-positive acute myeloid leukemia: results from the JPLSG AML-05 study.	Int J Hematol.	107	586-595	2018
Yamato G, Shiba N, Yoshida K, Hara Y, Shiraishi Y, Ohki K, Okubo J, Park MJ, Sotomatsu M, Arakawa H, Kiyokawa N, Tomizawa D, Adachi S, Taga T, Horibe K, Miyano S, Ogawa S, Hayashi Y.	RUNX1 mutations in pediatric acute myeloid leukemia are associated with distinct genetic features and an inferior prognosis.	Blood	131	2266-2270	2018
Imamura T, Taga T, Takagi M, Kawasaki H, Koh K, Taki T, Adachi S, Manabe A, Ishida Y; Leukemia/Lymphoma Committee; Japanese Society of Pediatric Hematology Oncology (JSPHO).	Nationwide survey of therapy-related leukemia in childhood in Japan.	Int J Hematol.	108	91-97	2018
Matsuo H, Yoshida K, Fukumura K, Nakatani K, Noguchi Y, Takasaki S, Noura M, Shiozawa Y, Shiraishi Y, Chiba K, Tanaka H, Okada A, Nannya Y, Takeda J, Ueno H, Shiba N, Yamato G, Handa H, Ono Y, Hiramoto N, Ishikawa T, Usuki K, Ishiyama K, Miyawaki S, Itonaga H, Miyazaki Y, Kawamura M, Yamaguchi H, Kiyokawa N, Tomizawa D, Taga T, Tawa A, Hayashi Y, Mano H, Miyano S, Kamikubo Y, Ogawa S, Adachi S.	Recurrent CCND3 mutations in MLL-rearranged acute myeloid leukemia.	Blood Adv.	2	2879-2889	2018
Yabushita T, Hiramoto N, Ono Y, Yoshioka S, Karakawa S, Kobayashi M, Ishikawa T	Adult-onset primary cyclic autoimmune neutropenia: a case report.	Transfusion	58	884-890	2018

Tomizawa D, Tanaka S, Hasegawa D, Iwamoto S, Hiramatsu H, Kiyokawa N, Miyachi H, Horibe K, Saito AM, Taga T, Adachi S.	Evaluation of high-dose cytarabine in induction therapy for children with de novo acute myeloid leukemia: a study protocol of the Japan Children's Cancer Group Multi-Center Seamless Phase II-III Randomized Trial (JPLSG AML-12).	Jpn J Clin Oncol.	48	587-593	2018
Asano T, Okada S, Tsumura M, Yeh TW, Mitsui-Sekinaka K, Tsujita Y, Ichinose Y, Shimada A, Hashimoto K, Wada T, Imai K, Ohara O, Morio T, Nonoyama S, Kobayashi M.	Enhanced AKT Phosphorylation of Circulating B Cells in Patients With Activated PI3K δ Syndrome.	Frontier in Immunology	9	568	2018
Zhang SY, Clark NE, Freije CA, Pauwels E, Taggart AJ, Okada S, Mandel H, Garcia P, Ciancanelli MJ, Biran A, Lafaille FG, Tsumura M, Cobat A, Luo J, Volpi S, Zimmer B, Sakata S, Dinis A, Ohara O, Garcia Reino EJ, Dobbs K, Hasek M, Holloway SP, McCammon K, Hussong SA, DeRosa N, Van Skike CE, Katolik A, Lorenzo L, Hyodo M, Faria E, Halwani R, Fukuhara R, Smith GA, Galvan V, Damha MJ, Al-Muhsen S, Itan Y, Boeke JD, Notarangelo LD, Studer L, Kobayashi M, Diogo L, Fairbrother WG, Abel L, Rosenberg BR, Hart PJ, Etzioni A, Casanova JL.	Errors of RNA Lariat Metabolism in Humans with Brainstem Viral Infection.	Cell	172	952-965	2018
Leiding JW, Okada S, Hagin D, Abinun M, Shcherbina A, Balashov DN, Kim VHD, Ovadia A, Guthery SL, Pulsipher M, Lilic D, Devlin LA, Christie S, Depner M, Fuchs S, van Royen-Kerkhof A, Lindemans C, Petrovic A, Sullivan KE, Bunin N, Kilic SS, Arpaci F, Calle-Martin O, Martinez-Martinez L, Aldave JC, Kobayashi M, Ohkawa T, Imai K, Iguchi A, Roifman CM, Gennery AR, Slatter M, Ochs HD, Morio T, Torgerson TR.	Inborn Errors Working Party of the European Society for Blood and Marrow Transplantation and the Primary Immune Deficiency Treatment Consortium: Hematopoietic stem cell transplantation in patients with gain-of-function signal transducer and activator of transcription 1 mutations.	Journal of Allergy & Clinical Immunology	141	704-717	2018

<p>Schwab C, Gabrysch A, Olbrich P, Patiño V, Warnatz K, Wolff D, Hoshino A, Kobayashi M, Imai K, Takagi M, Dybedal I, Haddock JA, Sansom DM, Lucena JM, Seidl M, Schmitt-Graeff A, Reiser V, Emmerich F, Frede N, Bulashevskaya A, Salzer U, Schubert D, Hayakawa S, Okada S, Kanariou M, Kucuk ZY, Chapdelaine H, Petruzelkova L, Sumnik Z, Sediva A, Slatter M, Arkwright PD, Cant A, Lorenz HM, Giese T, Lougaris V, Plebani A, Price C, Sullivan KE, Moutschen M, Litzman J, Freiburger T, van de Veerdonk FL, Recher M, Albert MH, Hauck F, Seneviratne S, Pachlopnik Schmid J, Kolios A, Unglik G, Klemann C, Speckmann C, Ehl S, Leichtner A, Blumberg R, Franke A, Snapper S, Zeissig S, Cunningham-Rundles C, Giulino-Roth L, Elemento O, Dückers G, Niehues T, Fronkova E, Kanderová V, Platt CD, Chou J, Chatila TA, Geha R, McDermott E, Bunn S, Kurzai M, Schulz A, Alsina L, Casals F, Deyà-Martinez A, Hambleton S, Kanegane H, Taskén K, Neth O, Grimbacher B.</p>	<p>Phenotype, penetrance, and treatment of 133 cytotoxic T-lymphocyte antigen 4-insufficient subjects.</p>	<p>Journal of Allergy & Clinical Immunology</p>	<p>142</p>	<p>1932-1946</p>	<p>2018</p>
<p>Mallick R, Jolles S, Kanegane H, Agbor-Tarh D, Rojavin M.</p>	<p>Treatment Satisfaction with Subcutaneous Immunoglobulin Replacement Therapy in Patients with Primary Immunodeficiency: a Pooled Analysis of Six Hizentra® Studies.</p>	<p>J Clin Immunol.</p>	<p>38(8)</p>	<p>886-897</p>	<p>2018</p>
<p>Tanaka-Kubota M, Shinozaki K, Miyamoto S, Yanagimachi M, Okano T, Mitsuiki N, Ueki M, Yamada M, Imai K, Takagi M, Agematsu K, Kanegane H, Morio T.</p>	<p>Hematopoietic stem cell transplantation for pulmonary alveolar proteinosis associated with primary immunodeficiency disease.</p>	<p>Int J Hematol.</p>	<p>107(5)</p>	<p>610-614</p>	<p>2018</p>
<p>Aoki T, Kunishima S, Yamashita Y, Minamitani K, Ota S.</p>	<p>Macrothrombocytopenia with congenital bilateral cataracts: a phenotype of <i>MYH9</i> disorder with exon 24 indel mutations.</p>	<p>J Pediatr Hematol/Oncol.</p>	<p>40(1)</p>	<p>76-78</p>	<p>2018</p>

Okano T, Tsujita Y, Kanegane H, Mitsui-Sekinaka K, Tanita K, Miyamoto S, Yeh TW, Yamashita M, Terada N, Ogura Y, Takagi M, Imai K, Nonoyama S, Morio T.	Droplet Digital PCR-Based Chimerism Analysis for Primary Immunodeficiency Diseases.	J. Clin. Immunol.	38(3)	300-306	2018
Sonoda M, Ishimura M, Ichimiya Y, Terashi E, Eguchi K, Sakai Y, Takada H, Hama A, Kanno H, Toki T, Ito E, Ohga S.	Atypical erythroblastosis in a patient with Diamond-Blackfan anemia who developed del(20q) myelodysplasia.	Int J Hematol.			2018
Hatta S, Fujiwara T, Yamamoto T, Saito K, Kamata M, Tamai Y, Kawamata S, Harigae H.	A defined culture method enabling the establishment of ring sideroblasts from induced pluripotent cells of X-linked sideroblastic anemia.	Haematologica	103	e188-e191	2018
Ohashi K, Fujiwara T, Onodera K, Saito Y, Ichikawa S, Kobayashi M, Okitsu Y, Fukuhara N, Onishi Y, Harigae H.	Establishment of a screening system to identify novel GATA-2 transcriptional regulators.	Tohoku J Exp Med.	244	41-52	2018
Morishima Y, Azuma F, Kashiwase K, Matsumoto K, Orihara T, Yabe H, Kato S, Kato K, Kai S, Mori T, Nakajima K, Morishima S, Satake M, Takanashi M, Yabe T; Japanese Cord Blood Transplantation Histocompatibility Research Group.	Risk of HLA Homozygous Cord Blood Transplantation: Implications for Induced Pluripotent Stem Cell Banking and Transplantation.	Stem Cells Transl Med.	7(2)	173-179	2018
Horikoshi Y, Umeda K, Imai K, Yabe H, Sasahara Y, Watanabe K, Ozawa Y, Hashii Y, Kurosawa H, Nonoyama S, Morio T.	Allogeneic Hematopoietic Stem Cell Transplantation for Leukocyte Adhesion Deficiency.	J Pediatr Hematol Oncol.	40(2)	137-140	2018
Oshima K, Saiki N, Tanaka M, Imamura H, Niwa A, Tanimura A, Nagahashi A, Hirayama A, Okita K, Hotta A, Kitayama S, Osawa M, Kaneko S, Watanabe A, Asaka I, Fujibuchi W, Imai K, Yabe H, Kamachi Y, Hara J, Kojima S, Tomita M, Soga T, Noma T, Nonoyama S, Nakahata T, Saito M.	Human AK2 links intracellular bioenergetic redistribution to the fate of hematopoietic progenitors.	Biochem Biophys Res Commun.	497(2)	719-725	2018
Ichimiya Y, Wada Y, Kunishima S, Tsukamoto K, Kosaki R, Sago H, Ishiguro A, Ito Y.	11q23 deletion syndrome (Jacobsen syndrome) with severe bleeding: a case report.	J Med Case Rep.	12	3	2018

Okamoto Y, Iwasaki WM, Kugou K, Takahashi KK, Oda A, Sato K, Kobayashi W, Kawai H, Sakasai R, Takaori-Kondo A, Yamamoto T, Kanemaki MT, Taoka M, Isobe T, Kurumizaka H, Innan H, Ohta K, Ishiai M, Takata M.	Replication stress induces accumulation of FANCD2 at central region of large fragile genes.	Nucleic Acid Res.	46(6)	2932-2944	2018
Hiejima E, Shibata H, Yasumi T, Shimodera S, Hori M, Izawa K, Kawai T, Matsuoka M, Kojima Y, Ohara A, Nishikomori R, Ohara O, Heike T.	Characterization of a large UNC13D gene duplication in a patient with familial hemophagocytic lymphohistiocytosis type 3.	Clin Immunol.	191	63-66	2018
Hao J, Kada A, Kunishima S.	Further classification of neutrophil non-muscle myosin heavy chain IIA localization for efficient genetic diagnosis of MYH9 disorders.	Ann Hematol.	97(4)	709-711	2018
Miyashita N, Onozawa M, Hayasaka K, Yamada T, Migita O, Hata K, Okada K, Goto H, Nakagawa M, Hashimoto D, Kahata K, Kondo T, Kunishima S, Teshima T.	A novel heterozygous ITGB3 p.T720del inducing spontaneous activation of integrin α IIb β 3 in autosomal dominant macrothrombocytopenia with aggregation dysfunction.	Ann Hematol.	97(4)	629-640	2018
Leiding JW, Okada S, Hagin D, Abinun M, Shcherbina A, Balashov DN, Kim VHD, Ovadia A, Guthery SL, Pulsipher M, Lilic D, Dvlin LA, Chritie S, Depner M, Fuchs S, van Royden-Kerkhof A, Lindemans C, Petrovic A, Sullivan KE, Bunin N, Kilic SS, Arpacı F, Calle-Martin O, Martinez-Martinez L, Alddave JC, Kobayashi M, Ohkawa T, Imai K, Iguchi A, Roifman CM, Genney AR, Slatter M, Ochs HD, Morio T, Torgerson TR, Inborn Errors Working Party of the European Society for Blood and Marrow Transplantation and the Primary Immune Deficiency Treatment Consortium.	Hematopoietic stem cell transplantation in patients with gain-of-function signal transducer and activator of transcription 1 mutations.	Journal of Allergy & Clinical Immunology	141.e5	704-717	2018

Noguchi J, Kanno H, Chiba Y, Ito E, Ishiguro A.	Discrimination of Diamond-Blackfan anemia from parvovirus B19 infection by RBC glutathione.	Pediatr Int.	59(7)	838-840	2017
Ogasawara T, Kawauchi K, Mori N, Sakura H, Katoh F, Kanno H, Ito E.	Successful long-term management with low-dose prednisolone in an adult patient with Diamond-Blackfan anemia.	Rinsho Ketsueki	58(8)	917-921	2017
Miot C, Imai K, Imai C, Mancini AJ, Kucuk XY, Kawai T, Nishikomori R, Ito E, Pellier I, Girod SD, Rosain J, Sasaki S, Chandrakasan S, Schmid JP, Okano T, Colin E, Olaya-Vargas A, Yamazaki-Nakashimada M, Qasim W, Padilla SE, Jones A, Krol A, Cole N, Jolles S, Bleesing J, Vraetz T, Gennery AR, Abinun M, Gungör T, Carvalho BC, Condino-Neto A, Veys P, Holland SM, Uzel G, Moshous D, Neven B, Ehl S, Döffinger R, Patel SY, Puel A, Bustamante J, Gelfand EW, Casanova JL, Orange JS, and Picard C.	Hematopoietic stem cell transplantation in 29 patients hemizygous for hypomorphic IKBKG / NEMO mutations.	Blood	130(12)	1456-1467	2017
Ichimura T, Yoshida K, Okuno Y, Yujiri T, Nagai K, Nishi M, Shiraishi Y, Ueno H, Toki T, Chiba K, Tanaka H, Muramatsu H, Hara T, Kanno H, Kojima S, Miyano S, Ito E, Ogawa S, Ohga S.	Diagnostic challenge of Diamond-Blackfan anemia in mothers and children by whole-exome sequencing.	Int J Hematol.	105(4)	515-520	2017
Ikeda F, Yoshida K, Toki T, Uechi T, Ishida S, Nakajima Y, Sasahara Y, Okuno Y, Kanezaki R, Terui K, Kamio T, Kobayashi A, Fujita T, Sato-Otsubo A, Shiraishi Y, Tanaka H, Chiba K, Muramatsu H, Kanno H, Ohga S, Ohara A, Kojima S, Kenmochi N, Miyano S, Ogawa S, Ito E.	Exome sequencing identified RPS15A as a novel causative gene for Diamond-Blackfan anemia.	Haematologica	102(3)	e93-e96	2017

Muramatsu H, Okuno Y, Yoshida K, Shiraishi Y, Doisaki S, Narita A, Sakaguchi H, Kawashima N, Wang X, Xu Y, Chiba K, Tanaka H, Hama A, Sanada M, Takahashi Y, Kanno H, Yamaguchi H, Ohga S, Manabe A, Harigae H, Kunishima S, Ishii E, Kobayashi M, Koike K, Watanabe K, Ito E, Takata M, Yabe M, Ogawa S, Miyano S, Kojima S.	Clinical utility of next-generation sequencing for inherited bone marrow failure syndromes.	Genet Med.	19(7)	796-802	2017
Hasegawa S, Fujiwara T, Okitsu Y, Kato H, Sato Y, Fukuhara N, Onishi Y, Shimizu R, Yamamoto M, Harigae H.	Effects of in vivo deletion of GATA2 in bone marrow stromal cells.	Exp Hematol.	56	31-45	2017
Fujiwara T, Fukuhara N, Ichikawa S, Kobayashi M, Okitsu Y, Onishi Y, Furuyama K, Harigae H.	A novel heterozygous ALAS2 mutation in a female with macrocytic sideroblastic anemia resembling myelodysplastic syndrome with ring sideroblasts: A case report and literature review.	Ann Hematol.	96	1955-1957	2017
Saito K, Fujiwara T, Ota U, Hatta S, Ichikawa S, Kobayashi M, Okitsu Y, Fukuhara N, Onishi Y, Ishizuka M, Tanaka T, Harigae H.	Dynamics of absorption, metabolism, and excretion of 5-aminolevulinic acid in human intestinal Caco-2 cells.	Biochem Biophys Rep.	11	105-111	2017
Fujiwara T, Sasaki K, Saito K, Hatta S, Ichikawa S, Kobayashi M, Okitsu Y, Fukuhara N, Onishi Y, Harigae H.	Forced FOG1 expression in erythroleukemia cells: induction of erythroid genes and repression of myelo-lymphoid transcription factor PU.1.	Biochem Biophys Res Commun.	485	380-387	2017
Inokura K, Fujiwara T, Saito K, Iino T, Hatta S, Okitsu Y, Fukuhara N, Onishi Y, Ishizawa K, Shimoda K, Harigae H.	Impact of TET2 deficiency on iron metabolism in erythroblasts.	Exp Hematol.	49	56-67	2017
Nishikawa E, Yagasaki H, Hama A, Yabe H, Ohara A, Kosaka Y, Kudo K, Kobayashi R, Ohga S, Morimoto A, Watanabe Ki, Yoshida N, Muramatsu H, Takahashi Y, Kojima S.	Long-term outcomes of 95 children with moderate aplastic anemia treated with horse antithymocyte globulin and cyclosporine.	Pediatr Blood Cancer.	64(5)	e26305	2017
Stapleton M, Kubaski F, Mason RW, Yabe H, Suzuki Y, Orii K, Orii T, Tomatsu S.	Presentation and Treatments for Mucopolysaccharidosis Type II (MPS II; Hunter Syndrome).	Expert Opin Orphan Drugs.	5(4)	295-307	2017

Hoenig M, Lagresle-Peyrou C, Pannicke U, Notarangelo LD, Porta F, Gennery AR, Slatter M, Cowan MJ, Stepensky P, Al-Mousa H, Al-Zahrani D, Pai SY, Al Herz W, Gaspar HB, Veys P, Oshima K, Imai K, Yabe H, Noroski LM, Wulffraat NM, Sykora KW, Soler-Palacin P, Muramatsu H, Al Hilali M, Moshous D, Debatin KM, Schuetz C, Jacobsen EM, Schulz AS, Schwarz K, Fischer A, Friedrich W, Cavazzana M.	Reticular dysgenesis: international survey on clinical presentation, transplantation and outcome.	Blood	129	2928-2938	2017
Sekinaka Y, Mitsui N, Imai K, Yabe M, Yabe H, Mitsui-Sekinaka K, Honma K, Takagi M, Arai A, Yoshida K, Okuno Y, Shiraishi Y, Chiba K, Tanaka H, Miyano S, Muramatsu H, Kojima S, Hira A, Takata M, Ohara O, Ogawa S, Morio T, Nonoyama S.	Common Variable Immunodeficiency Caused by FANCA Mutations.	J Clin Immunol.	37(5)	434-444	2017
Kubaski F, Yabe H, Suzuki Y, Seto T, Hamazaki T, Mason RW, Xie L, Onsten TGH, Leistner-Segal S, Giugliani R, Dūng VC, Ngoc CTB, Yamaguchi S, Montaña AM, Orii K, Fukao T, Shintaku H, Orii T, Tomatsu S.	Hematopoietic Stem Cell Transplantation for Patients with Mucopolysaccharidosis II.	Biol Blood Marrow Transplant.	23(10)	1795-1803	2017
Onishi Y, Mori T, Kako S, Koh H, Uchida N, Kondo T, Kobayashi T, Yabe H, Miyamoto T, Kato K, Suzuki R, Nakao S, Yamazaki H; Adult Aplastic Anemia Working Group of the Japan Society for Hematopoietic Cell Transplantation.	Outcome of Second Transplantation Using Umbilical Cord Blood for Graft Failure after Allogeneic Hematopoietic Stem Cell Transplantation for Aplastic Anemia.	Biol Blood Marrow Transplant.	23(12)	2137-2142	2017
Kanamitsu K, Shimada A, Nishiuchi R, Shigemura T, Nakazawa Y, Koike K, Kodama Y, Shinkoda Y, Kawano Y, Yasui K, Sasaki K, Kajiwara R, Tsukahara H, Manabe A.	Pediatric intestinal Behcet disease complicated by myeloid malignancies.	Int J Hematol.	105	377-382	2017

Hirabayashi S, Seki M, Hasegawa D, Kato M, Hyakuna N, Shuo T, Kimura S, Yoshida K, Kataoka K, Fujii Y, Shiraishi Y, Chiba K, Tanaka H, Kiyokawa N, Miyano S, Ogawa S, Takita J, Manabe A.	Constitutional abnormalities of IDFH1 combined with secondary mutations predispose a patient with Maffucci syndrome to acute lymphoblastic leukemia.	Pediatr Blood Cancer.	64(12)		2017
Narita A, Muramatsu H, Okuno Y, Sekiya Y, Suzuki K, Hamada M, Kataoka S, Ichikawa D, Taniguchi R, Murakami N, Kojima D, Nishikawa E, Kawashima N, Nishio N, Hama A, Takahashi Y, Kojima S.	Development of clinical paroxysmal nocturnal haemoglobinuria in children with aplastic anaemia.	Br J Haematol.	78(6)	954-958	2017
Matsumaru S, Oguni H, Ogura H, Shimojima K, Nagata S, Kanno H, Yamamoto T.	A novel PGK1 mutation associated with neurological dysfunction and the absence of episodes of hemolytic anemia or myoglobinuria.	Intractable Rare Diseases Research	6(2)	132-136	2017
Sakaue S, Kasai T, Mizuta I, Suematsu M, Osone S, Azuma Y, Imamura T, Tokuda T, Kanno H, El-Agnaf OMA, Morimoto M, Nakagawa M, Hosoi H, Mizuno T.	Early-onset parkinsonism in a pedigree with phosphoglycerate kinase deficiency and a heterozygous carrier: do PGK-1 mutations contribute to vulnerability to parkinsonism?.	NPJ Parkinsons Dis.	3(1)	13	2017
Niizuma H, Kanno H, Sato A, Ogura H, Imaizumi M.	Splenectomy resolves hemolytic anemia caused by adenylate kinase deficiency.	Pediatr Int.	59(2)	228-230	2017
Kadoda K, Moriwaki T, Tsuda M, Sasanuma H, Ishiai M, Takata M, Ide H, Masunaga SI, Takeda S, Tano K.	Selective cytotoxicity of the anti-diabetic drug, metformin, in glucose-deprived chicken DT40 cells.	PLoS One	12(9)	e0185141	2017
Mochizuki AL, Katanaya A, Hayashi E, Hosokawa M, Moribe E, Motegi A, Ishiai M, Takata M, Kondoh G, Watanabe H, Nakatsuji N, Chuma S.	PARI regulates stalled replication fork processing to maintain genome stability upon replication stress in mice.	Mol Cell Biol.	37(23)	e00117-17	2017
Knies K, Inano S, Ramírez MJ, Ishiai M, Surallés J, Takata M, and Schindler D.	Biallelic mutations in the ubiquitin ligase <i>RFWD3</i> cause Fanconi anemia.	J Clin Invest.	127(8)	3013-3027	2017

Inano S, Sato K, Katsuki Y, Kobayashi W, Tanaka H, Nakajima K, Nakada S, Hiroyuki Miyoshi, Knies K, Takaori-Kondo A, Schindler D, Ishiai M, Kurumizaka H, Takata M.	RFWD3-mediated ubiquitination promotes timely removal of both RPA and RAD51 from DNA damage sites to facilitate homologous recombination.	Mol Cell.	66(5)	622-634	2017
Ishiai M, Sato K, Tomida J, Kitao H, Kurumizaka H, Takata M.	Mutation Research special section "Protein modifications in DNA repair and cancer" Activation of the FA pathway mediated by phosphorylation and ubiquitination.	Mutat Res.	803-805	89-95	2017
Watanabe M, Nishikomori R, Fujimaki Y, Heike T, Ohara A, Saji T.	Live-attenuated vaccines in a cryopyrin-associated periodic syndrome patient receiving canakinumab treatment during infancy.	Clin Case Rep.	5	1750-1755	2017
Yusa T, Tateda K, Ohara A, Miyazaki S.	New possible biomarkers for diagnosis of infections and diagnostic distinction between bacterial and viral infections in children.	J Infect Chemother.	23	96-100	2017
Onodera R, Kurita E, Taniguchi K, Karakawa S, Okada S, Kihara H, Fujii T, Kobayashi M.	Anti-human neutrophil antigen-1a, -and -2 antibodies in neonates and children with immune neutropenia analyzed by extracted granulocyte antigen immunofluorescence assay.	Transfusion	57	2586-2594	2017
Hayakawa S, Ohno N, Okada S, Kobayashi M.	Significant augmentation of regulatory T cell numbers occurs during the early neonatal period.	Clin Exp Immunol.	190	268-279	2017
Fujiki R, Hijikata A, Shirai T, Okada S, Kobayashi M, Ohara O.	Molecular mechanism and structural basis of gain-offunction of STAT1 caused by pathogenic R274Q mutation.	Journal of Biological Chemistry	292	6240-6254	2017
Yamasaki F, Takayasu T, Nosaka R, Nishibuchi I, Kawaguchi H, Kolakshyapati M M, Onishi S, Saito T, Sugiyama K, Koabayashi M, Kurisu K.	Development of cystic malacia after high-dose cranial irradiation of pediatric CNS tumors in long-term follow-up.	Child's Nervous System	33	957-964	2017
矢部みはる, 矢部普正	Fanconi貧血 臨床診断・検査・治療	日本臨床	75 (増刊1)	414-417	2017
矢部普正, 矢部みはる	成人のFanconi貧血の特徴と管理	日本臨床	75 (増刊1)	418-421	2017

Kobayashi T, Nannya Y, Ichikawa M, Oritani K, Kanakura Y, Tomita A, Kiyoi H, Kobune M, Kato J, Kawabata H, Shindo M, Torimoto Y, Yonemura Y, Hanaoka N, Nakakuma H, Hasegawa D, Manabe A, Fujishima N, Fujii N, Tanimoto M, Morita Y, Matsuda A, Fujieda A, Katayama N, Ohashi H, Nagai H, Terada Y, Hino M, Sato K, Obara N, Chiba S, Usuki K, Ohta M, Imataki O, Uemura M, Takaku T, Komatsu N, Kitanaka A, Shimoda K, Watanabe K, Tohyama K, Takaori-Kondo A, Harigae H, Arai S, Miyazaki Y, Ozawa K, Kurokawa M; for National Research Group on Idiopathic Bone Marrow Failure Syndromes.	A nationwide survey of hypoplastic myelodysplastic syndrome (a multicenter retrospective study).	Am J Hematol.	92	1224-1232	2017
Kagawa R, Fujiki R, Tsumura M, Sakata S, Nishimura S, Itan Y, Kong XF, Kato Z, Ohnishi H, Hirata O, Saito S, Ikeda M, El Baghdadi J, Bousfiha A, Fujiwara K, Oleastro M, Yancoski J, Perez L, Danielian S, Ailal F, Takada H, Hara T, Pue; A, Boisson-Dupuis S, Bustamate J, Casanovva JL, Ohara O, Okada S, Kobayashi M.	Alanine-scanning mutagenesis of human signal transducer and activator of transcription 1 to estimate loss- or gain-of-function variants.	Journal of Allergy & Clinical Immunology	140	232-241	2017
Hoshino A, Okada S, Yoshida K, Nishida N, Okuno Y, Ueno H, Yamashita M, Okano T, Tsumura M, Nishimura S, Sakata S, Kobayashi M, Nakamura H, Kamizono J, Mitsui-Sekinaka K, Ichimura T, Ohga S, Nakazawa Y, Takagi M, Imai K, Shiraishi Y, Chiba K, Tanaka H, Miyano S, Ogawa S, Kojima S, Nonoyama S, Morio T, Kanegane H.	Abnormal hematopoiesis and autoimmunity in human subjects with germline IKZF1 mutations.	Journal of Allergy & Clinical Immunology	140	223-231	2017
Kanda K, Kunishima S, Sato A, Abe D, Nishijima S, Ishigami T.	A Brazilian case of Bernard-Soulier syndrome with two distinct founder mutations.	Hum Genome Var.	4	17030	2017

Sivapalaratnam S, Westbury SK, Stephens JC, Greene D, Downes K, Kelly AM, Lentaigne C, Astle WJ, Huizinga EG, Nurden P, Papadia S, Peerlinck K, Penkett CJ, Perry DJ, Roughley C, Simeoni I, Stirrups K, Hart DP, Tait RC, Mumford AD; NIHR BioResource., Laffan MA, Freson K, Ouwehand WH, Kunishima S, Turro E.	Rare variants in GP1BB are responsible for autosomal dominant macrothrombocytopenia.	Blood	129(4)	520-524	2017
Ogawa Y, Kunishima S, Yanagisawa K, Osaki Y, Uchiyama Y, Matsumoto N, Tokiniwa H, Horiguchi J, Nojima Y, Handa H.	Successful management of perioperative hemostasis in a patient with Glanzmann thrombasthenia who underwent a right total mastectomy.	Int J Hematol.	105(2)	221-225	2017
Kanehira M, Fujiwara T, Nakajima S, Okitsu Y, Onishi Y, Fukuhara N, Ichinohasama R, Harigae H.	An LPA1/3 axis governs cellular senescence of mesenchymal stromal cells and promotes growth and vascularization of multiple myeloma.	Stem Cells.	35(3)	739-753	2017
Kobayashi M, Kato H, Hada H, Itoh-Nakadai A, Fujiwara T, Inoguchi Y, Ichianagi K, Muto A, Tomosugi N, Sasaki H, Harigae H, Igarashi K.	Iron-heme-Bach1 axis is involved in erythroblast adaptation to iron deficiency.	Haematologica	102(3)	454-465	2017
Ishiguro A, Ezinne CC, Michihata N, Nakadate H, Manabe A, Taki M, Shima M.	Pediatric Thromboembolism: A National Survey in Japan.	Int J Hematol.	105	52-58	2017
山口博樹	骨髓不全症におけるテロメア制御異常.	血液フロンティア	27(1)	5-9	2017
Yabe M, Yabe H, Morimoto T, Fukumura A, Ohtsubo K, Koike T, Yoshida K, Ogawa S, Ito E, Okuno Y, Muramatsu H, Kojima S, Matsuo K, Hira A, Takata M.	The phenotype and clinical course of Japanese Fanconi Anaemia infants is influenced by patient, but not maternal ALDH2 genotype.	Br J Haematol.	175	457-461	2016
Utsugisawa T, Uchiyama T, Toki T, Ogura H, Aoki T, Hamaguchi I, Ishiguro A, Ohara A, Kojima S, Ohga S, Ito E, Kanno H.	Erythrocyte glutathione is a novel biomarker of Diamond-Blackfan anemia.	Blood Cells, Molecules and Diseases	59	31-36	2016
Wasano K, Matsunaga T, Ogawa K, Kunishima S.	Late onset and high-frequency dominant hearing loss in a family with MYH9 disorder.	Eur Arch Otorhinolaryngol	273(11)	3547-3552	2016

Kubota Y 他	Novel Mechanisms for Heme-dependent Degradation of ALAS1 Protein as a Component of Negative Feedback Regulation of Heme Biosynthesis.	J Biol Chem.	291	20516-29	2016
Ikeda F, Toki T, Kanezaki R, Terui K, Yoshida K, Kanno H, Ohga S, Ohara A, Kojima S, Ogawa S, Ito E.	ALDH2 polymorphism in patients with Diamond-Blackfan anemia in Japan.	Int J Hematol.	103(1)	112-4	2016
Shim YJ, Kim HS, Do YR, Ha JS, Yabe H.	Sequential strategy for umbilical cord blood transplantation in a Korean Fanconi anemia girl with refractory acute myelomonocytic leukemia and complex karyotype.	Pediatr Transplant.	doi: 10.1111/ petr.12871		2016 [Epub ahead of print]
Yabe M, Yabe H, Morimoto T, Fukumura A, Ohtsubo K, Koike T, Yoshida K, Ogawa S, Ito E, Okuno Y, Muramatsu H, Kojima S, Matsuo K, Hira A, Takata M.	The phenotype and clinical course of Japanese Fanconi Anaemia infants is influenced by patient, but not maternal ALDH2 genotype.	Br J Haematol.	175(3)	457-461	2016
Yamashita Y, Matsuura R, Kunishima S, Oikawa Y, Ariizumi H, Hamada S, Shirato N, Matsuoka R, Ogawa K, Sekizawa A.	Perinatal Management for a Pregnant Woman with an MYH9 Disorder.	Case Rep Obstet Gynecol.	2016	6730174	2016
Kitamura K, Okuno Y, Yoshida K, Sanada M, Shiraishi Y, Muramatsu H, Kobayashi R, Furukawa K, Miyano S, Kojima S, Ogawa S, Kunishima S.	Functional characterization of a novel GFI1B mutation causing congenital macrothrombocytopenia.	J Thromb Haemost.	14(7)	1462-9	2016
Yokoi S, Kunishima S, Takahashi Y, Morishita M, Kojima S.	A Japanese pedigree with a p.A95V mutation in the MYH9 gene demonstrates inherited macrothrombocytopenia without Alport manifestations.	Ann Hematol.	95(5)	831-3	2016
Kondo A, Fujiwara T, Okitsu Y, Fukuhara N, Onishi Y, Nakamura Y, Sawada K, Harigae H.	Identification of a novel putative mitochondrial protein FAM210B associated with erythroid differentiation.	Int J Hematol.	103	387-395	2016
Sakurai K, Fujiwara T, Hasegawa S, Okitsu Y, Fukuhara N, Onishi Y, Yamada-Fujiwara M, Ichinohasama R, Harigae H.	Inhibition of human primary megakaryocyte differentiation by anagrelide: A gene expression profiling analysis.	Int J Hematol.	104	190-199	2016

Simeoni I, Stephens JC, Hu F, Deevi SV, Megy K, Bariana TK, Lentaigne C, Schulman S, Sivapalaratnam S, Vries MJ, Westbury SK, Greene D, Papadia S, Alessi MC, Attwood AP, Ballmaier M, Baynam G, Bermejo E, Bertoli M, Bray PF, Bury L, Cattaneo M, Collins P, Daugherty LC, Favier R, French DL, Furie B, Gattens M, Germeshausen M, Ghevaert C, Goodeve AC, Guerrero JA, Hampshire DJ, Hart DP, Heemskerk JW, Henskens YM, Hill M, Hogg N, Jolley JD, Kahr WH, Kelly AM, Kerr R, Kostadima M, Kunishima S, Lambert MP, Liesner R, López JA, Mapeta RP, Mathias M, Millar CM, Nathwani A, Neerman-Arbez M, Nurden AT, Nurden P, Othman M, Peerlinck K, Perry DJ, Poudel P, Reitsma P, Rondina MT, Smethurst PA, Stevenson W, Szkotak A, Tuna S, van Geet C, Whitehorn D, Wilcox DA, Zhang B, Revel-Vilk S, Gresele P, Bellissimo DB, Penkett CJ, Laffan MA, Mumford AD, Rendon A, Gomez K, Freson K, Ouwehand WH, Turro E.	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders.	Blood	127(23)	2791-803	2016
Onodera K, Fujiwara T, Onishi Y, Okitsu Y, Itoh-Nakadai A, Okitsu Y, Fukuhara N, Ishizawa K, Shimizu R, Yamamoto M, Harigae H.	GATA2 regulates dendritic cell differentiation	Blood	128	508-518	2016
Imai J, Suzuki T, Yoshikawa M, Dekiden M, Nakae H, Nakahara F, Tsuda S, Mizukami H, Koike J, Igarashi M, Yabe H, Mine T.	Fatal Hemorrhagic Gastrointestinal Angioectasia after Bone Marrow Transplantation for Dyskeratosis Congenita.	Intern Med	55	3341-3444	2016
Otsubo K, Yabe M, Yabe H, Fukumura A, Morimoto T, Kato M, Mochizuki H.	Successful acute lymphoblastic leukemia-type therapy in two children with mixed-phenotype acute leukemia.	Pediatr Int.	58	1072-1076	2016
Yabe H.	Allogeneic hematopoietic stem cell transplantation for inherited diseases.	Rinsho Ketsueki	57	2199-2207	2016

Umeda K, Adachi S, Tanaka S, Miki M, Okada K, Hashii Y, Inoue M, Cho Y, Koh K, Goto H, Kajiwara R, Hyakuna N, Kato K, Morio T, Yabe H; Inherited Disease Working Group of the Japan Society for Hematopoietic Cell Transplantation.	Comparison of second transplantation and donor lymphocyte infusion for donor mixed chimerism after allogeneic stem cell transplantation for nonmalignant diseases.	Pediatr Blood Cancer.	63	2221-2229	2016
Sakaguchi H, Watanabe N, Matsumoto K, Yabe H, Kato S, Ogawa A, Inagaki J, Goto H, Koh K, Yoshida N, Kato K, Cho Y, Kosaka Y, Takahashi Y, Inoue M, Kato K, Atsuta Y, Miyamura K; Donor/Source Working Group of Japan Society of Hematopoietic Cell Transplantation.	Comparison of Donor Sources in Hematopoietic Stem Cell Transplantation for Childhood Acute Leukemia: A Nationwide Retrospective Study.	Biol Blood Marrow Transplant.	22	2226-2234	2016
Atsuta Y, Hirakawa A, Nakasone H, Kurosawa S, Oshima K, Sakai R, Ohashi K, Takahashi S, Mori T, Ozawa Y, Fukuda T, Kanamori H, Morishima Y, Kato K, Yabe H, Sakamaki H, Taniguchi S, Yamashita T; Late Effect and Quality of Life Working Group of the Japan Society for Hematopoietic Cell Transplantation.	Late Mortality and Causes of Death among Long-Term Survivors after Allogeneic Stem Cell Transplantation.	Biol Blood Marrow Transplant.	22	1702-1709	2016
Yasuda E, Suzuki Y, Shimada T, Sawamoto K, Mackenzie WG, Theroux MC, Pizarro C, Xie L, Miller F, Rahman T, Kecskemethy HH, Nagao K, Morlet T, Shaffer TH, Chinen Y, Yabe H, Tanaka A, Shintaku H, Orii KE, Orii KO, Mason RW, Montaña AM, Fukao T, Orii T, Tomatsu S.	Activity of daily living for Morquio A syndrome.	Mol Genet Metab.	118	111-22	2016

Kato S, Yabe H, Takakura H, Mugishima H, Ishige M, Tanaka A, Kato K, Yoshida N, Adachi S, Sakai N, Hashii Y, Ohashi T, Sasahara Y, Suzuki Y, Tabuchi K.	Hematopoietic stem cell transplantation for inborn errors of metabolism: A report from the Research Committee on Transplantation for Inborn Errors of Metabolism of the Japanese Ministry of Health, Labour and Welfare and the Working Group of the Japan Society for Hematopoietic Cell Transplantation.	Pediatr Transplant.	20	203-214	2016
Elmahdi,S, Hama A, Manabe A, Hasegawa D, Muramatsu H, Narita A, Nishio N, Ismael O, Kawashima N, Okuno Y, Xu Y, Wang X, Takahashi Y, Ito M, Kojima S.	A cytokine-based diagnostic program in pediatric aplastic anemia and hypocellular refractory cytopenia of childhood.	Pediatr Blood Cancer.	63	652-658	2016
Elmahadi S, Muramatsu H, Kojima S.	Allogeneic hematopoietic stem cell transplantation for dyskeratosis congenita.	Curr Opin Hematol.	23(6)	501-507	2016
Kojima D, Wang X, Muramatsu H, Okuno Y, Nishio N, Hama A, Tsuge I, Takahashi Y, Kojima S.	Application of extensively targeted next-generation sequencing for the diagnosis of primary immunodeficiencies.	J Allergy Clin Immunol.	138(1)	303-305.e3	2016
Imashuku S, Muramatsu H, Sugihara T, Okuno Y, Wang X, Yoshida K, Kato A, Kato K, Tatsumi Y, Hattori A, Kita S, Oe K, Sueyoshi A, Usui T, Shiraishi Y, Chiba K, Tanaka H, Miyano S, Ogawa S, Kojima S, Kanno H.	PIEZO1 gene mutation in a Japanese family with hereditary high phosphatidylcholine hemolytic anemia and hemochromatosis-induced diabetes mellitus.	Int J Hematol.	104(1)	125-129	2016
Arashiki N, Takakuwa Y, Mohandas N, Hale J, Yoshida K, Ogura H, Utsugisawa T, Ohga S, Miyano S, Ogawa S, Kojima S, Kanno H.	ATP11C is a major flippase in human erythrocytes and its defect causes congenital hemolytic anemia.	Haematologica.	101(5)	59-65	2016
小倉浩美, 菅野仁	【貧血 実地医家に役立つ知識と診療の進めかた】セミナー臨床に役立つ知識と情報 溶血性貧血の鑑別診断の進め方	MedicalPractic	33(9)	1387-1391	2016
大賀正一, 山城安啓, 菅野仁	【貧血性疾患診療の進歩】先天性溶血性貧血の遺伝子診断	血液内科	73(2)	149-154	2016
新敷信人, 菅野仁, 高桑雄一	ヒト赤血球膜におけるフリッパーゼ分子の同定とリン脂質非対称性維持のメカニズム	脂質生化学研究	58	72-73	2016
小倉浩美, 菅野仁	【血球の増加と減少】赤血球 貧血 遺伝性貧血	小児内科	48(7)	1000-3	2016

小林博人, 菅野仁	型T細胞を用いた癌免疫療法	日本輸血細胞治療学会誌	62(1)	3-12	2016
Ling C, Huang J, Yan Z, Li Y, Ohzeki M, Ishiai M, Xu D, Takata M, Seidman M, and Wang W.	Bloom syndrome complex promotes FANCM recruitment to stalled replication forks and facilitates both repair and traverse of DNA interstrand crosslinks.	Cell Discov.	2	16047	2016
Tian X, Patel K, Ridpath JR, Chen Y, Zhou YH, Neo D, Clement J, Takata M, Takeda S, Sale J, Wright FA, Swenberg JA, Nakamura J.	Homologous recombination and translesion DNA synthesis play critical roles on tolerating DNA damage caused by trace levels of hexavalent chromium.	pLOS One	11(12)	e0167503	2016
Sato K, Shimomuki M, Katsuki Y, Takahashi D, Kobayashi W, Ishiai M, Miyoshi H, Takata M, Kurumizaka H.	FANCI-FANCD2 stabilizes the RAD51-DNA complex by binding RAD51 and protects the 5'-DNA end.	Nucleic Acids Res.	44	10758-10771	2016
Katsuki Y, Takata M.	Defects in HR repair behind the human diseases: FA and HBOC.	Endocrine Related Cancer.	23	T19-37	2016
Hashimoto K, Sharma V, Sasanuma H, Tian X, Takata M, Takeda S, Swenberg J and Nakamura J.	Poor recognition of O6-isopropyl dG by MGMT triggers double strand break-mediated cell death and micronucleus induction in FANC-deficient cells.	Oncotarget	7	59795-59808	2016
Mu A, Li M, Tanaka M, Adachi Y, Tai TT, Liem PH, Izawa S, Furuyama K, Taketani S	Enhancements of the production of bilirubin and the expression of β -globin by carbon monoxide during erythroid differentiation.	FEBS Lett.	590	1447-54	2016
Moriya K, Suzuki T, Watanabe Y, Saito- Nanjo Y, Niizuma H, Onuma M, Rikiishi T, Kakuta F, Abukawa D, Yamaguchi H, Sasahara Y, Kure S.	Hoyeraal-Hreidarsson syndrome in a patient with novel compound heterozygous RTEL1 gene mutations.	Pediatric Blood & Cancer	63(9)	1683-4	2016