Yusuke Okuno

List of Publications by Year in Descending Order

230 papers	7,533 citations	38 h-index	84 g-index
252 ext. papers	9,334 ext. citations	5.9 avg, IF	5 L-index

#	Paper	IF	Citations
230	International Consensus Definition of DNA Methylation Subgroups in Juvenile Myelomonocytic Leukemia. *Clinical Cancer Research*, **2021**, 27, 158-168	12.9	7
229	Report on effective treatment and genetic predisposition in two children with refractory probable catastrophic antiphospholipid syndrome. *Thrombosis Research*, **2021**, 208, 117-120	8.2	0
228	Autoinflammatory Keratinization Disease With Hepatitis and Autism Reveals Roles for JAK1 Kinase Hyperactivity in Autoinflammation.. *Frontiers in Immunology*, **2021**, 12, 737747	8.4	0
227	Mutations in SAM syndrome and palmoplantar keratoderma patients suggest genotype/phenotype correlations in DSG1 mutations. *Journal of the European Academy of Dermatology and Venereology*, **2021**,	4.6	1
226	Clinical diagnostic value of telomere length measurement in inherited bone marrow failure syndromes. *Haematologica*, **2021**, 106, 2511-2515	6.6	0
225	RNAseq analysis identifies involvement of EBNA2 in PD-L1 induction during Epstein-Barr virus infection of primary B cells. *Virology*, **2021**, 557, 44-54	3.6	4
224	A patient with very early onset FH-deficient renal cell carcinoma diagnosed at age seven. *Familial Cancer*, **2021**, 1	3	1
223	Relationship between plasma rabbit anti-thymocyte globulin concentration and immunosuppressive therapy response in patients with severe aplastic anemia. *European Journal of Haematology*, **2021**, 107, 255-264	3.8	0
222	Integrated diagnosis based on transcriptome analysis in suspected pediatric sarcomas. *Npj Genomic Medicine*, **2021**, 6, 49	6.2	2
221	Deletion of Viral microRNAs in the Oncogenesis of Epstein-Barr Virus-Associated Lymphoma. *Frontiers in Microbiology*, **2021**, 12, 667968	5.7	5
220	Detection of subclonal SETBP1 and JAK3 mutations in juvenile myelomonocytic leukemia using droplet digital PCR. *Leukemia*, **2021**, 35, 259-263	10.7	3
219	Phosphorylated proteome analysis of a novel germline ABL1 mutation causing an autosomal dominant syndrome with ventricular septal defect. *International Journal of Cardiology*, **2021**, 326, 81-87	3.2	1
218	Whole-exome sequencing and host cell reactivation assay lead to a diagnosis of xeroderma pigmentosum group D with mild ultraviolet radiation sensitivity. *Journal of Dermatology*, **2021**, 48, 96-100	1.6	
217	Role of Epstein-Barr Virus C Promoter Deletion in Diffuse Large B Cell Lymphoma. *Cancers*, **2021**, 13,	6.6	6
216	Mathematical Modeling and Mutational Analysis Reveal Optimal Therapy to Prevent Malignant Transformation in Grade II IDH-Mutant Gliomas. *Cancer Research*, **2021**, 81, 4861-4873	10.1	0
215	Successful treatment of a novel type I interferonopathy due to a de novo PSMB9 gene mutation with a Janus kinase inhibitor. *Journal of Allergy and Clinical Immunology*, **2021**, 148, 639-644	11.5	8
214	Echocardiography Monitoring of Pulmonary Hypertension after Pediatric Hematopoietic Stem Cell Transplantation: Pediatric Pulmonary Arterial Hypertension and Pulmonary Veno-Occlusive Disease after Hematopoietic Stem Cell Transplantation. *Transplantation and Cellular Therapy*, **2021**, 27, 786.e1-786.e8		1

213	A simple and robust methylation test for risk stratification of patients with juvenile myelomonocytic leukemia. *Blood Advances*, **2021**,	7.8	1
212	Novel COL4A1 mutations identified in infants with congenital hemolytic anemia in association with brain malformations. *Human Genome Variation*, **2020**, 7, 42	1.8	1
211	Two Aldehyde Clearance Systems Are Essential to Prevent Lethal Formaldehyde Accumulation in Mice and Humans. *Molecular Cell*, **2020**, 80, 996-1012.e9	17.6	39
210	Genetic analysis in patients with newly diagnosed glioblastomas treated with interferon-beta plus temozolomide in comparison with temozolomide alone. *Journal of Neuro-Oncology*, **2020**, 148, 17-27	4.8	2
209	A novel sensitive detection method for DNA methylation in circulating free DNA of pancreatic cancer. *PLoS ONE*, **2020**, 15, e0233782	3.7	6
208	H3F3A mutant allele specific imbalance in an aggressive subtype of diffuse midline glioma, H3 K27M-mutant. *Acta Neuropathologica Communications*, **2020**, 8, 8	7.3	6
207	Deep phenotyping of ichthyosis follicularis with atrichia and photophobia syndrome associated with MBTPS2 mutations. *Journal of Dermatology*, **2020**, 47, e87-e88	1.6	1
206	Diagnostic Whole Exome Sequencing for 166 Patients with Inherited Bone Marrow Failure Syndrome. *Blood*, **2020**, 136, 9-9	2.2	
205	Digenic mutations in and impair formaldehyde clearance and cause a multisystem disorder, AMeD syndrome. *Science Advances*, **2020**, 6,	14.3	14
204	SDR9C7 catalyzes critical dehydrogenation of acylceramides for skin barrier formation. *Journal of Clinical Investigation*, **2020**, 130, 890-903	15.9	29
203	Peptides containing the MXXCW motif inhibit oncogenic RET kinase activity with a novel mechanism of action. *American Journal of Cancer Research*, **2020**, 10, 336-349	4.4	
202	Novel compound heterozygous MCOLN1 mutations identified in a Japanese girl with severe developmental delay and thin corpus callosum. *Brain and Development*, **2020**, 42, 298-301	2.2	1
201	Oncogenesis of CAEBV revealed: Intragenic deletions in the viral genome and leaky expression of lytic genes. *Reviews in Medical Virology*, **2020**, 30, e2095	11.7	16
200	Novel biallelic FA2H mutations in a Japanese boy with fatty acid hydroxylase-associated neurodegeneration. *Brain and Development*, **2020**, 42, 217-221	2.2	4
199	Reduced stratum corneum acylceramides in autosomal recessive congenital ichthyosis with a NIPAL4 mutation. *Journal of Dermatological Science*, **2020**, 97, 50-56	4.3	8
198	Comprehensive pathogen detection in sera of Kawasaki disease patients by high-throughput sequencing: a retrospective exploratory study. *BMC Pediatrics*, **2020**, 20, 482	2.6	3
197	Multi-Lineage Expression in Philadelphia Chromosome-Positive Acute Lymphoblastic Leukemia Is Associated With Improved Prognosis but No Specific Molecular Features. *Frontiers in Oncology*, **2020**, 10, 586567	5.3	2
196	Targetable driver mutations in multicentric reticulohistiocytosis. *Haematologica*, **2020**, 105, e61-e64	6.6	7

195	A founder variant in the South Asian population leads to a high prevalence of FANCL Fanconi anemia cases in India. *Human Mutation*, **2020**, 41, 122-128	4.7	5
194	Frequent -Activating Mutations in Extramammary Paget's Disease. *Cancers*, **2020**, 12,	6.6	10
193	Direct Evidence of Abortive Lytic Infection-Mediated Establishment of Epstein-Barr Virus Latency During B-Cell Infection. *Frontiers in Microbiology*, **2020**, 11, 575255	5.7	5
192	Next Generation Sequencing-Based Transcriptome Predicts Bevacizumab Efficacy in Combination with Temozolomide in Glioblastoma. *Molecules*, **2019**, 24,	4.8	5
191	A novel splice site variant in a young male exhibiting less pronounced features. *Human Genome Variation*, **2019**, 6, 43	1.8	1
190	Metagenomic analysis using next-generation sequencing of pathogens in bronchoalveolar lavage fluid from pediatric patients with respiratory failure. *Scientific Reports*, **2019**, 9, 12909	4.9	19
189	Pathogenic Epigenetic Consequences of Genetic Alterations in IDH-Wild-Type Diffuse Astrocytic Gliomas. *Cancer Research*, **2019**, 79, 4814-4827	10.1	2
188	Defective Epstein-Barr virus in chronic active infection and haematological malignancy. *Nature Microbiology*, **2019**, 4, 404-413	26.6	80
187	Essential role of PTPN11 mutation in enhanced haematopoietic differentiation potential of induced pluripotent stem cells of juvenile myelomonocytic leukaemia. *British Journal of Haematology*, **2019**, 187, 163-173	4.5	4
186	Elucidation of the pathogenic mechanism and potential treatment strategy for a female patient with spastic paraplegia derived from a single-nucleotide deletion in PLP1. *Journal of Human Genetics*, **2019**, 64, 665-671	4.3	6
185	KLF1 mutation E325K induces cell cycle arrest in erythroid cells differentiated from congenital dyserythropoietic anemia patient-specific induced pluripotent stem cells. *Experimental Hematology*, **2019**, 73, 25-37.e8	3.1	11
184	Pathogenic mutations identified by a multimodality approach in 117 Japanese Fanconi anemia patients. *Haematologica*, **2019**, 104, 1962-1973	6.6	9
183	A Syrian Refugee in Iraq Diagnosed as a Case of IL12RB1 Deficiency in Japan Using Dried Blood Spots. *Frontiers in Immunology*, **2019**, 10, 58	8.4	4
182	Aberrant Active cis-Regulatory Elements Associated with Downregulation of RET Finger Protein Overcome Chemoresistance in Glioblastoma. *Cell Reports*, **2019**, 26, 2274-2281.e5	10.6	5
181	Study of pathophysiology and molecular characterization of congenital anemia in India using targeted next-generation sequencing approach. *International Journal of Hematology*, **2019**, 110, 618-626	2.3	12
180	DOCK8 mutation diagnosed using whole-exome sequencing of the dried blood spot-derived DNA: a case report of an Iraqi girl diagnosed in Japan. *BMC Medical Genetics*, **2019**, 20, 114	2.1	2
179	Utility of Newborn Screening for Severe Combined Immunodeficiency and X-Linked Agammaglobulinemia Using TREC and KREC Assays. *Blood*, **2019**, 134, 3604-3604	2.2	1
178	Comprehensive Mutational Analysis of Juvenile Myelomonocytic Leukemia Using Whole-Genome Sequencing. *Blood*, **2019**, 134, 2974-2974	2.2	

177	Genome-Wide Methylation Analysis Using the Digital Restriction Enzyme Analysis of Methylation for Stratification of Patients with Juvenile Myelomonocytic Leukemia. *Blood*, **2019**, 134, 2973-2973	2.2	
176	Detection of Subclonal SETBP1 and JAK3 Mutations in Patients with Juvenile Myelomonocytic Leukemia Using Droplet Digital PCR. *Blood*, **2019**, 134, 4213-4213	2.2	
175	Transcriptome analysis offers a comprehensive illustration of the genetic background of pediatric acute myeloid leukemia. *Blood Advances*, **2019**, 3, 3157-3169	7.8	21
174	Frequent intragenic microdeletions of elastin in familial supravalvular aortic stenosis. *International Journal of Cardiology*, **2019**, 274, 290-295	3.2	8
173	Modification of cellular and humoral immunity by somatically reverted T cells in X-linked lymphoproliferative syndrome type 1. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 421-424.e11	11.5	5
172	Associations of complementation group, ALDH2 genotype, and clonal abnormalities with hematological outcome in Japanese patients with Fanconi anemia. *Annals of Hematology*, **2019**, 98, 271-280	3	6
171	An infant with generalized pustular psoriasis and geographic tongue had a heterozygous IL36RN mutation and IgG2 deficiency. *Journal of Dermatological Science*, **2018**, 90, 216-218	4.3	2
170	Comprehensive detection of pathogens in immunocompromised children with bloodstream infections by next-generation sequencing. *Scientific Reports*, **2018**, 8, 3784	4.9	31
169	Dysregulation of Epstein-Barr Virus Infection in Hypomorphic ZAP70 Mutation. *Journal of Infectious Diseases*, **2018**, 218, 825-834	7	10
168	Integrated molecular profiling of juvenile myelomonocytic leukemia. *Blood*, **2018**, 131, 1576-1586	2.2	51
167	Mild case of Hailey-Hailey disease caused by a novel ATP2C1 mutation. *Journal of Dermatology*, **2018**, 45, e207-e208	1.6	3
166	Enhanced Expression of Anti-CD19 Chimeric Antigen Receptor in Transposon-Engineered T Cells. *Molecular Therapy - Methods and Clinical Development*, **2018**, 8, 131-140	6.4	33
165	A novel IFIH1 mutation in the pincer domain underlies the clinical features of both Aicardi-Goutières and Singleton-Merten syndromes in a single patient. *British Journal of Dermatology*, **2018**, 178, e111-e113	4	9
164	Biallelic mutations in SZT2 cause a discernible clinical entity with epilepsy, developmental delay, macrocephaly and a dysmorphic corpus callosum. *Brain and Development*, **2018**, 40, 134-139	2.2	13
163	Genetic heterogeneity of uncharacterized childhood autoimmune diseases with lymphoproliferation. *Pediatric Blood and Cancer*, **2018**, 65, e26831	3	12
162	Trichothiodystrophy, complementation group A complicated with squamous cell carcinoma. *Journal of the European Academy of Dermatology and Venereology*, **2018**, 32, e75-e77	4.6	1
161	Comprehensive genetic analysis of donor cell derived leukemia with KMT2A rearrangement. *Pediatric Blood and Cancer*, **2018**, 65, e26823	3	4
160	Integration Mapping of piggyBac-Mediated CD19 Chimeric Antigen Receptor T Cells Analyzed by Novel Tagmentation-Assisted PCR. *EBioMedicine*, **2018**, 34, 18-26	8.8	16

159	Identification of potential pathogenic viruses in patients with acute myocarditis using next-generation sequencing. *Journal of Medical Virology*, **2018**, 90, 1814-1821	19.7	21
158	De Novo Mutations Activating Germline TP53 in an Inherited Bone-Marrow-Failure Syndrome. *American Journal of Human Genetics*, **2018**, 103, 440-447	11	18
157	DNA Methylation Subgroups in Juvenile Myelomonocytic Leukemia: An International Collaborative Analysis and Development of a Common Diagnostic Platform. *Blood*, **2018**, 132, 3093-3093	2.2	2
156	Combination of TREC Measurement and Next-Generation Sequencing in Newborn Screening for Severe Combined Immunodeficiency: A Pilot Program in Japan. *Blood*, **2018**, 132, 3717-3717	2.2	2
155	Dopamine and Serotonin Receptors Cooperatively Modulate Pacemaker Activity of Intestinal Cells of Cajal. *Chinese Journal of Physiology*, **2018**, 61, 302-312	1.6	3
154	Analysis of Genomic Predispositions to Sporadic Myeloid Neoplasms Mediated By DDX41 in Japan. *Blood*, **2018**, 132, 4371-4371	2.2	
153	Characterization of Pathogenic Variants and Clinical Phenotypes in 117 Japanese Fanconi Anemia Patients. *Blood*, **2018**, 132, 3860-3860	2.2	
152	The Presence of Defective Epstein-Barr Virus (EBV) Infection in Patients with EBV-Associated Hematological Malignancy. *Blood*, **2018**, 132, 1562-1562	2.2	
151	Clinical and Genetic Characteristics of Patients with Shwachman-Diamond Syndrome in Japan. *Blood*, **2018**, 132, 3862-3862	2.2	
150	Regulation of Epstein-Barr Virus Life Cycle and Cell Proliferation by Histone H3K27 Methyltransferase EZH2 in Akata Cells. *MSphere*, **2018**, 3,	5	13
149	Reduction of stratum corneum ceramides in Neu-Laxova syndrome caused by phosphoglycerate dehydrogenase deficiency. *Journal of Lipid Research*, **2018**, 59, 2413-2420	6.3	7
148	Gain-of-function mutation causes human combined immune deficiency. *Journal of Experimental Medicine*, **2018**, 215, 2715-2724	16.6	40
147	Sterol profiles are valuable biomarkers for phenotype expression of Conradi-HÜermann-Happle syndrome with EBP mutations. *British Journal of Dermatology*, **2018**, 179, 1186-1188	4	0
146	A patient with a GNAO1 mutation with decreased spontaneous movements, hypotonia, and dystonic features. *Brain and Development*, **2018**, 40, 926-930	2.2	6
145	Whole-exome analysis to detect congenital hemolytic anemia mimicking congenital dyserythropoietic anemia. *International Journal of Hematology*, **2018**, 108, 306-311	2.3	4
144	Germline IKAROS mutation associated with primary immunodeficiency that progressed to T-cell acute lymphoblastic leukemia. *Leukemia*, **2017**, 31, 1221-1223	10.7	31
143	Clinical utility of next-generation sequencing for inherited bone marrow failure syndromes. *Genetics in Medicine*, **2017**, 19, 796-802	8.1	45
142	A case of GATA2-related myelodysplastic syndrome with unbalanced translocation der(1;7)(q10;p10). *Pediatric Blood and Cancer*, **2017**, 64, e26419	3	3

141	Exome sequencing identified as a novel causative gene for Diamond-Blackfan anemia. *Haematologica*, **2017**, 102, e93-e96	6.6	25
140	Common Variable Immunodeficiency Caused by FANC Mutations. *Journal of Clinical Immunology*, **2017**, 37, 434-444	5.7	15
139	Efficacy of neutrophil non-muscle myosin heavy chain-IIA immunofluorescence analysis in determining the pathogenicity of MYH9 variants. *Annals of Hematology*, **2017**, 96, 1065-1066	3	1
138	A combination of low-dose systemic etretinate and topical calcipotriol/betamethasone dipropionate treatment for hyperkeratosis and itching in Olmsted syndrome associated with a TRPV3 mutation. *Journal of Dermatological Science*, **2017**, 88, 144-146	4.3	5
137	Recurrent MYB rearrangement in blastic plasmacytoid dendritic cell neoplasm. *Leukemia*, **2017**, 31, 1629-1633	10.7	26
136	Autosomal dominant familial generalized pustular psoriasis caused by a CARD14 mutation. *British Journal of Dermatology*, **2017**, 177, e133-e135	4	22
135	Dynamics of clonal evolution in myelodysplastic syndromes. *Nature Genetics*, **2017**, 49, 204-212	36.3	228
134	Abnormal hematopoiesis and autoimmunity in human subjects with germline IKZF1 mutations. *Journal of Allergy and Clinical Immunology*, **2017**, 140, 223-231	11.5	63
133	Comprehensive detection of viruses in pediatric patients with acute liver failure using next-generation sequencing. *Journal of Clinical Virology*, **2017**, 96, 67-72	14.5	17
132	Loss of DNA Damage Response in Neuroblastoma and Utility of a PARP Inhibitor. *Journal of the National Cancer Institute*, **2017**, 109,	9.7	23
131	A case of lamellar ichthyosis due to a novel TGM1 mutation associated with Parkinson's disease. *European Journal of Dermatology*, **2017**, 27, 438-439	0.8	1
130	Congenital Ichthyosis and Recurrent Eczema Associated with a Novel ALOXE3 Mutation. *Acta Dermato-Venereologica*, **2017**, 97, 532-533	2.2	4
129	Striate Palmoplantar Keratoderma Showing Transgrediens in a Patient Harbouring Heterozygous Nonsense Mutations in Both DSG1 and SERPINB7. *Acta Dermato-Venereologica*, **2017**, 97, 399-401	2.2	4
128	Biallelic Mutations in KDSR Disrupt Ceramide Synthesis and Result in a Spectrum of Keratinization Disorders Associated with Thrombocytopenia. *Journal of Investigative Dermatology*, **2017**, 137, 2344-2353	4.3	35
127	Development of clinical paroxysmal nocturnal haemoglobinuria in children with aplastic anaemia. *British Journal of Haematology*, **2017**, 178, 954-958	4.5	11
126	Haploinsufficiency of TNFAIP3 (A20) by germline mutation is involved in autoimmune lymphoproliferative syndrome. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 1914-1922	11.5	69
125	Clinical utility of next-generation sequencing-based minimal residual disease in paediatric B-cell acute lymphoblastic leukaemia. *British Journal of Haematology*, **2017**, 176, 248-257	4.5	20
124	A newly revealed IL36RN mutation in sibling cases complements our IL36RN mutation statistics for generalized pustular psoriasis. *Journal of Dermatological Science*, **2017**, 85, 58-60	4.3	9

123	Diagnostic challenge of Diamond-Blackfan anemia in mothers and children by whole-exome sequencing. *International Journal of Hematology*, **2017**, 105, 515-520	2.3	14
122	Next-generation sequencing-guided identification of causative gene mutations. *Japanese Journal of Thrombosis and Hemostasis*, **2017**, 28, 3-8	0	
121	MEF2D-BCL9 Fusion Gene Is Associated With High-Risk Acute B-Cell Precursor Lymphoblastic Leukemia in Adolescents. *Journal of Clinical Oncology*, **2016**, 34, 3451-9	2.2	68
120	JAK2, MPL, and CALR mutations in children with essential thrombocythemia. *International Journal of Hematology*, **2016**, 104, 266-7	2.3	8
119	Phosphatase and tensin homolog (PTEN) mutation can cause activated phosphatidylinositol 3-kinase δ syndrome-like immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 1672-1680.e10	11.5	61
118	Successful T-cell reconstitution after unrelated cord blood transplantation in a patient with complete DiGeorge syndrome. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 1471-1473.e4	11.5	3
117	Identification of Viruses in Cases of Pediatric Acute Encephalitis and Encephalopathy Using Next-Generation Sequencing. *Scientific Reports*, **2016**, 6, 33452	4.9	51
116	Markedly High Plasma Thrombopoietin (TPO) Level is a Predictor of Poor Response to Immunosuppressive Therapy in Children With Acquired Severe Aplastic Anemia. *Pediatric Blood and Cancer*, **2016**, 63, 659-64	3	10
115	PIEZO1 gene mutation in a Japanese family with hereditary high phosphatidylcholine hemolytic anemia and hemochromatosis-induced diabetes mellitus. *International Journal of Hematology*, **2016**, 104, 125-9	2.3	18
114	Extrapulmonary tuberculosis mimicking Mendelian susceptibility to mycobacterial disease in a patient with signal transducer and activator of transcription 1 (STAT1) gain-of-function mutation. *Journal of Allergy and Clinical Immunology*, **2016**, 137, 619-622.e1	11.5	11
113	Development of Paroxysmal Nocturnal Hemoglobinuria in Children with Aplastic Anemia. *Blood*, **2016**, 128, 1499-1499	2.2	1
112	Novel and recurrent mutations in Japanese patients with Darier's disease. *Nagoya Journal of Medical Science*, **2016**, 78, 485-492	0.7	2
111	Plakin Family Autoantibodies in Bronchiolitis Obliterans Following Hematopoietic Stem Cell Transplantation As Useful Biomarkers and the Target for Rituximab Therapy. *Blood*, **2016**, 128, 3432-3432	2.2	
110	Genetic Landscape and Clonal Evolution Following 5-Aza Therapy in Patients with High-Risk Myelodysplastic Syndromes. *Blood*, **2016**, 128, 4304-4304	2.2	
109	Comprehensive Genetic Analysis in Cases of Juvenile Myelomonocytic Leukemia for Prognostic Estimation. *Blood*, **2016**, 128, 3159-3159	2.2	
108	the Impact of Clonal Dynamics on Prognosis and Outcome in Myelodysplastic Syndromes. *Blood*, **2016**, 128, 4287-4287	2.2	
107	Whole-Genome Sequencing of Primary Central Nervous System Lymphoma and Diffuse Large B-Cell Lymphoma. *Blood*, **2016**, 128, 4112-4112	2.2	0
106	A Cytokine-Based Diagnostic Program in Pediatric Aplastic Anemia and Hypocellular Refractory Cytopenia of Childhood. *Pediatric Blood and Cancer*, **2016**, 63, 652-8	3	4

105	Functional characterization of a novel GFI1B mutation causing congenital macrothrombocytopenia. *Journal of Thrombosis and Haemostasis*, **2016**, 14, 1462-9	15.4	28
104	Immunosuppressive therapy for patients with Down syndrome and idiopathic aplastic anemia. *International Journal of Hematology*, **2016**, 104, 130-3	2.3	2
103	Application of extensively targeted next-generation sequencing for the diagnosis of primary immunodeficiencies. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 303-305.e3	11.5	7
102	The phenotype and clinical course of Japanese Fanconi Anaemia infants is influenced by patient, but not maternal ALDH2 genotype. *British Journal of Haematology*, **2016**, 175, 457-461	4.5	8
101	Whole-exome sequencing reveals the spectrum of gene mutations and the clonal evolution patterns in paediatric acute myeloid leukaemia. *British Journal of Haematology*, **2016**, 175, 476-489	4.5	42
100	Correlation of rabbit antithymocyte globulin serum levels and clinical outcomes in children who received hematopoietic stem cell transplantation from an alternative donor. *Pediatric Transplantation*, **2016**, 20, 105-13	1.8	8
99	X-linked agammaglobulinemia associated with B-precursor acute lymphoblastic leukemia. *Journal of Clinical Immunology*, **2015**, 35, 108-11	5.7	17
98	A female patient with incomplete hemophagocytic lymphohistiocytosis caused by a heterozygous XIAP mutation associated with non-random X-chromosome inactivation skewed towards the wild-type XIAP allele. *Journal of Clinical Immunology*, **2015**, 35, 244-8	5.7	24
97	Integrated genetic and epigenetic analysis defines novel molecular subgroups in rhabdomyosarcoma. *Nature Communications*, **2015**, 6, 7557	17.4	110
96	Somatic Mutations and Clonal Hematopoiesis in Aplastic Anemia. *New England Journal of Medicine*, **2015**, 373, 35-47	59.2	361
95	Somatic mosaicism for a NRAS mutation associates with disparate clinical features in RAS-associated leukoproliferative disease: a report of two cases. *Journal of Clinical Immunology*, **2015**, 35, 454-8	5.7	26
94	Clinical and genetic features of dyskeratosis congenita, cryptic dyskeratosis congenita, and Hoyeraal-Hreidarsson syndrome in Japan. *International Journal of Hematology*, **2015**, 102, 544-52	2.3	14
93	GATA2 and secondary mutations in familial myelodysplastic syndromes and pediatric myeloid malignancies. *Haematologica*, **2015**, 100, e398-401	6.6	38
92	Late-Onset Combined Immunodeficiency with a Novel IL2RG Mutation and Probable Revertant Somatic Mosaicism. *Journal of Clinical Immunology*, **2015**, 35, 610-4	5.7	18
91	Fulminant adenovirus hepatitis after hematopoietic stem cell transplant: Retrospective real-time PCR analysis for adenovirus DNA in two cases. *Journal of Infection and Chemotherapy*, **2015**, 21, 857-63	2.2	7
90	Profiling of somatic mutations in acute myeloid leukemia with FLT3-ITD at diagnosis and relapse. *Blood*, **2015**, 126, 2491-501	2.2	134
89	Mutations in the gene encoding the E2 conjugating enzyme UBE2T cause Fanconi anemia. *American Journal of Human Genetics*, **2015**, 96, 1001-7	11	90
88	Paroxysmal nocturnal hemoglobinuria and telomere length predicts response to immunosuppressive therapy in pediatric aplastic anemia. *Haematologica*, **2015**, 100, 1546-52	6.6	46

87	BRCC3 mutations in myeloid neoplasms. *Haematologica*, **2015**, 100, 1051-7	6.6	17
86	Choreito formula for BK virus-associated hemorrhagic cystitis after allogeneic hematopoietic stem cell transplantation. *Biology of Blood and Marrow Transplantation*, **2015**, 21, 319-25	4.7	13
85	Loss of function mutations in RPL27 and RPS27 identified by whole-exome sequencing in Diamond-Blackfan anaemia. *British Journal of Haematology*, **2015**, 168, 854-64	4.5	67
84	Two Novel Distinct Subtypes of Myeloid Neoplasms Molecularly Associated with Histone H3K36 Methylations. *Blood*, **2015**, 126, 2841-2841	2.2	1
83	Genetic Predispositions to Myeloid Neoplasms Caused By Germline DDX41 Mutations. *Blood*, **2015**, 126, 2843-2843	2.2	5
82	Genetic Background of Idiopathic Bone Marrow Failure Syndromes in Children. *Blood*, **2015**, 126, 3610-3610	2.2	1
81	Serial Sequencing in Myelodysplastic Syndromes Reveals Dynamic Changes in Clonal Architecture and Allows for a New Prognostic Assessment of Mutations Detected in Cross-Sectional Testing. *Blood*, **2015**, 126, 709-709	2.2	2
80	Aberrant DNA Methylation Is Associated with a Poor Outcome in Juvenile Myelomonocytic Leukemia. *PLoS ONE*, **2015**, 10, e0145394	3.7	18
79	COL4A1 is a Novel Causative Gene Responsible for Congenital Hemolytic Anemia, Representing Characteristic Clinical Course in Infants. *Blood*, **2015**, 126, 934-934	2.2	
78	Landscape of DNA Methylation and Genetic Profiles in 291 Patients with Myelodysplastic Syndromes. *Blood*, **2015**, 126, 5205-5205	2.2	
77	Functional Characterization of a Novel GFI1B Mutation Causing Congenital Macrothrombocytopenia. *Blood*, **2015**, 126, 75-75	2.2	
76	Myelodysplastic Syndrome Patients Show Mutation-Specific DNA Methylation Patterns. *Blood*, **2015**, 126, 1646-1646	2.2	
75	Evaluation of Cytokines after Platelet-Lysate-Expanded Mesenchymal Stromal Cell Therapy for Steroid Refractory Acute Graft-Versus-Host Disease. *Blood*, **2015**, 126, 1892-1892	2.2	
74	Clinical Utility of Next-Generation Sequencing-Based Minimal Residual Disease in Childhood Acute Lymphoblastic Leukemia. *Blood*, **2015**, 126, 2611-2611	2.2	
73	Correction of Fanconi Anemia Mutation Using the Crispr/Cas9 System. *Blood*, **2015**, 126, 3622-3622	2.2	0
72	Whole-Exome Analysis of Autoimmune Lymphoproliferative Syndrome-like Diseases. *Blood*, **2015**, 126, 1022-1022	2.2	
71	Biallelic DICER1 mutations in sporadic pleuropulmonary blastoma. *Cancer Research*, **2014**, 74, 2742-9	10.1	56
70	Genomic and molecular characterization of esophageal squamous cell carcinoma. *Nature Genetics*, **2014**, 46, 467-73	36.3	398

69	Somatic RHOA mutation in angioimmunoblastic T cell lymphoma. *Nature Genetics*, **2014**, 46, 171-5	36.3	411
68	Whole-exome sequence analysis of ataxia telangiectasia-like phenotype. *Journal of the Neurological Sciences*, **2014**, 340, 86-90	3.2	11
67	Landscape of genetic lesions in 944 patients with myelodysplastic syndromes. *Leukemia*, **2014**, 28, 241-7	10.7	957
66	Clonal leukemic evolution in myelodysplastic syndromes with TET2 and IDH1/2 mutations. *Haematologica*, **2014**, 99, 28-36	6.6	37
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