Akiko Shimamura

List of Publications by Year
in descending order

100	6,578	76326	66911
papers	citations	40 h-index	78 g-index

102	102	102	6563
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Low frequency of treatable pediatric disease alleles in gnomAD: An opportunity for future genomic screening of newborns. Human Genetics and Genomics Advances, 2022, 3, 100059.	1.7	3
2	Autophagy in mesenchymal progenitors protects mice against bone marrow failure after severe intermittent stress. Blood, 2022, 139, 690-703.	1.4	8
3	Hematologic complications with age in Shwachman-Diamond syndrome. Blood Advances, 2022, 6, 297-306.	5.2	23
4	Lessons From Pediatric MDS: Approaches to Germline Predisposition to Hematologic Malignancies. Frontiers in Oncology, 2022, 12, 813149.	2.8	16
5	Telomere biology disorders: ends and (genetic) means. Blood, 2022, 139, 1776-1777.	1.4	0
6	Coronavirus disease 2019 and vaccination in patients with Shwachmanâ€Diamond syndrome. Pediatric Blood and Cancer, 2022, 69, e29647.	1.5	3
7	Metformin for treatment of cytopenias in children and young adults with Fanconi anemia. Blood Advances, 2022, 6, 3803-3811.	5.2	4
8	International Consensus Classification of Myeloid Neoplasms and Acute Leukemias: integrating morphologic, clinical, and genomic data. Blood, 2022, 140, 1200-1228.	1.4	814
9	MYC Promotes Bone Marrow Stem Cell Dysfunction in Fanconi Anemia. Cell Stem Cell, 2021, 28, 33-47.e8.	11.1	31
10	Repolarization of HSC attenuates HSCs failure in Shwachmanâ€“Diamond syndrome. Leukemia, 2021, 35, 1751-1762.	7.2	5
11	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	6.1	34
12	Inhibition of TGFÎ²1 and TGFÎ²3 promotes hematopoiesis in Fanconi anemia. Experimental Hematology, 2021, 93, 70-84.e4.	0.4	8
13	Distinct genetic pathways define pre-malignant versus compensatory clonal hematopoiesis in Shwachman-Diamond syndrome. Nature Communications, 2021, 12, 1334.	12.8	103
14	Molecular alterations governing predisposition to myelodysplastic syndromes: Insights from Shwachman-Diamond syndrome. Best Practice and Research in Clinical Haematology, 2021, 34, 101252.	1.7	2
15	Dissecting ELANE neutropenia pathogenicity by human HSC gene editing. Cell Stem Cell, 2021, 28, 833-845.e5.	11.1	23
16	SLC25A38 congenital sideroblastic anemia: Phenotypes and genotypes of 31 individuals from 24 families, including 11 novel mutations, and a review of the literature. Human Mutation, 2021, 42, 1367-1383.	2.5	11
17	Diagnostic workâ€up for severe aplastic anemia in children: Consensus of the <scp>North American Pediatric Aplastic Anemia Consortium</scp>. American Journal of Hematology, 2021, 96, 1491-1504.	4.1	14
18	Association of unbalanced translocation der(1;7) with germline GATA2 mutations. Blood, 2021, 138, 2441-2445.	1.4	12

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19	Congenital Xâ€linked Neutropenia with Myelodysplasia and Somatic Tetraploidy due to a Germline Mutation in SEPT6. American Journal of Hematology, 2021, , .	4.1	1
20	The frequent and clinically benign anomalies of chromosomes 7 and 20 in Shwachman-diamond syndrome may be subject to further clonal variations. Molecular Cytogenetics, 2021, 14, 54.	0.9	10
21	Translational research for bone marrow failure patients. Experimental Hematology, 2021, , .	0.4	3
22	Clinical features and outcomes of patients with Shwachman-Diamond syndrome and myelodysplastic syndrome or acute myeloid leukaemia: a multicentre, retrospective, cohort study. Lancet Haematology,the, 2020, 7, e238-e246.	4.6	73
23	A study assessing the feasibility of randomization of pediatric and young adult patients between matched unrelated donor bone marrow transplantation and immuneâ€suppressive therapy for newly diagnosed severe aplastic anemia: A joint pilot trial of the North American Pediatric Aplastic Anemia Consortium and the Pediatric Transplantation and Cellular Therapy Consortium. Pediatric Blood and Cancer, 2020, 67, e28444.	1.5	11
24	Diagnosis and treatment of pediatric myelodysplastic syndromes: A survey of the North American Pediatric Aplastic Anemia Consortium. Pediatric Blood and Cancer, 2020, 67, e28652.	1.5	12
25	Inflammatory manifestations in patients with Shwachmanâ€“Diamond syndrome: A novel phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 1754-1760.	1.2	8
26	An induced pluripotent stem cell model of Fanconi anemia reveals mechanisms of p53-driven progenitor cell differentiation. Blood Advances, 2020, 4, 4679-4692.	5.2	1
27	Towards Identifying the Target of Autoimmunity in Aplastic Anemia. Blood, 2020, 136, 2-2.	1.4	0
28	Neutropenia in the age of genetic testing: Advances and challenges. American Journal of Hematology, 2019, 94, 384-393.	4.1	18
29	Genetic predisposition to MDS: clinical features and clonal evolution. Blood, 2019, 133, 1071-1085.	1.4	100
30	Immunosuppressive therapy for pediatric aplastic anemia: a North American Pediatric Aplastic Anemia Consortium study. Haematologica, 2019, 104, 1974-1983.	3.5	43
31	Genetic predisposition to MDS: diagnosis and management. Hematology American Society of Hematology Education Program, 2019, 2019, 110-119.	2.5	13
32	Bone Marrow Morphology Associated With GermlineRUNX1Mutations in Patients With Familial Platelet Disorder With Associated Myeloid Malignancy. Pediatric and Developmental Pathology, 2019, 22, 315-328.	1.0	23
33	Inherited thrombocytopenia associated with mutation of UDP-galactose-4-epimerase (GALE). Human Molecular Genetics, 2019, 28, 133-142.	2.9	73
34	Therapeutic discovery for marrow failure with MDS predisposition using pluripotent stem cells. JCI Insight, 2019, 4, .	5.0	10
35	TGF-Î² signaling underlies hematopoietic dysfunction and bone marrow failure in Shwachman-Diamond syndrome. Journal of Clinical Investigation, 2019, 129, 3821-3826.	8.2	25
36	Gene Editing ELANE in Human Hematopoietic Stem and Progenitor Cells Reveals Disease Mechanisms and Therapeutic Strategies for Severe Congenital Neutropenia. Blood, 2019, 134, 3-3.	1.4	8

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37	Constitutional SAMD9L mutations cause familial myelodysplastic syndrome and transient monosomy 7. Haematologica, 2018, 103, 427-437.	3.5	83
38	Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene BRCA1. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 5241-5246.	7.1	29
39	Targeted mass spectrometry enables robust quantification of FANCD2 mono-ubiquitination in response to DNA damage. DNA Repair, 2018, 65, 47-53.	2.8	18
40	Somatic mutations and clonal hematopoiesis in congenital neutropenia. Blood, 2018, 131, 408-416.	1.4	91
41	Itâ€™s ALL in the Family: IKZF1 and Hereditary Leukemia. Cancer Cell, 2018, 33, 798-800.	16.8	4
42	Pancreatic lipomatosis in Diamondâ€“Blackfan anemia: The importance of genetic testing in bone marrow failure disorders. American Journal of Hematology, 2018, 93, 1194-1195.	4.1	1
43	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. European Respiratory Journal, 2017, 49, 1601640.	6.7	41
44	The eIF2-alpha kinase HRI is a novel therapeutic target in multiple myeloma. Leukemia Research, 2017, 55, 23-32.	0.8	22
45	Germline Genetic Predisposition to Hematologic Malignancy. Journal of Clinical Oncology, 2017, 35, 1018-1028.	1.6	80
46	Allogeneic Hematopoietic Cell Transplantation Using Treosulfan-Based Conditioning for Treatment of Marrow Failure Disorders. Biology of Blood and Marrow Transplantation, 2017, 23, 1669-1677.	2.0	45
47	Genetic predisposition to hematologic malignancies: management and surveillance. Blood, 2017, 130, 424-432.	1.4	145
48	ETV6 in hematopoiesis and leukemia predisposition. Seminars in Hematology, 2017, 54, 98-104.	3.4	87
49	Pregnancy outcomes in inherited bone marrow failure syndromes. Blood, 2017, 130, 1671-1674.	1.4	12
50	Long-Term Effects of G-CSF Therapy in Cyclic Neutropenia. New England Journal of Medicine, 2017, 377, 2290-2292.	27.0	35
51	Recent discoveries in the molecular pathogenesis of the inherited bone marrow failure syndrome Fanconi anemia. Blood Reviews, 2017, 31, 93-99.	5.7	109
52	Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamondâ€“like features. Journal of Clinical Investigation, 2017, 127, 4090-4103.	8.2	126
53	The Activated TGFÎ² Pathway in Shwachman Diamond Syndrome Impairs Hematopoiesis and Is Down-Regulated By Deletion of 7q. Blood, 2017, 130, 875-875.	1.4	3
54	Aplastic anemia and clonal evolution: germ line and somatic genetics. Hematology American Society of Hematology Education Program, 2016, 2016, 74-82.	2.5	35

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55	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. Blood, 2016, 127, 1017-1023.	1.4	179
56	Identifying Inherited and Acquired Genetic Factors Involved in Poor Stem Cell Mobilization and Donor-Derived Malignancy. Biology of Blood and Marrow Transplantation, 2016, 22, 2100-2103.	2.0	42
57	Delayed globin synthesis leads to excess heme and the macrocytic anemia of Diamond Blackfan anemia and del(5q) myelodysplastic syndrome. Science Translational Medicine, 2016, 8, 338ra67.	12.4	68
58	Genetic features of myelodysplastic syndrome and aplastic anemia in pediatric and young adult patients. Haematologica, 2016, 101, 1343-1350.	3.5	124
59	Ataxia-Pancytopenia Syndrome Is Caused by Missense Mutations in SAMD9L. American Journal of Human Genetics, 2016, 98, 1146-1158.	6.2	136
60	Modeling Bone Marrow Failure and MDS in Shwachman Diamond Syndrome Using Induced Pluripotent Stem Cells. Blood, 2016, 128, 1496-1496.	1.4	1
61	Placental Growth Factor (PlGF) Enhances TLR/MK2 Dependent TNF Gene Transcription and Dissociates miRNAs Targeting TNF Regulatory Transcripts from the Polysome. Blood, 2016, 128, 408-408.	1.4	0
62	Single Cell Transcriptional Profiling Reveals Activation of TNF-Alpha Signaling in Hematopoietic Stem and Progenitor Cells from Shwachman-Diamond Syndrome Patients. Blood, 2016, 128, 335-335.	1.4	0
63	Detection of Mutations in Inherited Bone Marrow Failure and Myelodysplastic Syndrome Genes Using Genomic Capture and Massively Parallel Sequencing in Clinical Diagnostics. Blood, 2016, 128, 1507-1507.	1.4	0
64	Germline and Somatic Genetic Characterization of Shwachman-Diamond Syndrome. Blood, 2016, 128, 2681-2681.	1.4	0
65	Mutation Burden in Hematopoietic Stem Cells Is Not Increased in Congenital Neutropenia. Blood, 2016, 128, 405-405.	1.4	2
66	Hyperactive Non-Canonical TGF-Î² Pathway Signaling in Fanconi Anemia Bone Marrow Stromal Cells Contributes to Growth Suppression. Blood, 2016, 128, 1039-1039.	1.4	0
67	Genomic analysis of bone marrow failure and myelodysplastic syndromes reveals phenotypic and diagnostic complexity. Haematologica, 2015, 100, 42-48.	3.5	108
68	Congenital sideroblastic anemia due to mutations in the mitochondrial HSP70 homologue HSPA9. Blood, 2015, 126, 2734-2738.	1.4	78
69	Germline ETV6 Mutations Confer Susceptibility to Acute Lymphoblastic Leukemia and Thrombocytopenia. PLoS Genetics, 2015, 11, e1005262.	3.5	128
70	The diversity of mutations and clinical outcomes for ELANE-associated neutropenia. Current Opinion in Hematology, 2015, 22, 3-11.	2.5	123
71	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. Nature Genetics, 2015, 47, 180-185.	21.4	299
72	The North American Shwachman-Diamond Syndrome Registry: Genetically Undefined Shwachman-Diamond Syndrome. Blood, 2015, 126, 3614-3614.	1.4	0

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73	Identifying Inherited and Acquired Genetic Factors Involved in Poor Stem Cell Mobilization and Donor-Derived Malignancy. Blood, 2015, 126, 3163-3163.	1.4	0
74	Spindle Microtubule Dysfunction and Cancer Predisposition. Journal of Cellular Physiology, 2014, 229, 1881-1883.	4.1	14
75	Variable Clinical Presentation of Shwachmanâ€“Diamond Syndrome: Update from the North American Shwachmanâ€“Diamond Syndrome Registry. Journal of Pediatrics, 2014, 164, 866-870.	1.8	121
76	Treosulfan-Based Conditioning and Hematopoietic Cell Transplantation for Nonmalignant Diseases: A Prospective Multicenter Trial. Biology of Blood and Marrow Transplantation, 2014, 20, 1996-2003.	2.0	51
77	Diagnosis and treatment of pediatric acquired aplastic anemia (AAA): An initial survey of the North American Pediatric Aplastic Anemia Consortium (NAPAAC). Pediatric Blood and Cancer, 2014, 61, 869-874.	1.5	31
78	Marrow failure: a window into ribosome biology. Blood, 2014, 124, 2784-2792.	1.4	105
79	H/ACA Small RNA Dysfunctions in Disease Reveal Key Roles for Noncoding RNA Modifications in Hematopoietic Stem Cell Differentiation. Cell Reports, 2013, 3, 1493-1502.	6.4	109
80	Clinical and Molecular Pathophysiology of Shwachmanâ€“Diamond Syndrome. Hematology/Oncology Clinics of North America, 2013, 27, 117-128.	2.2	97
81	Pluripotent Stem Cell Models of Shwachman-Diamond Syndrome Reveal a Common Mechanism for Pancreatic and Hematopoietic Dysfunction. Cell Stem Cell, 2013, 12, 727-736.	11.1	66
82	Impaired ribosomal subunit association in Shwachman-Diamond syndrome. Blood, 2012, 120, 5143-5152.	1.4	66
83	Breast cancer in a case of Shwachman Diamond syndrome. Pediatric Blood and Cancer, 2012, 59, 945-946.	1.5	21
84	Variable Clinical Presentation of Shwachman-Diamond Syndrome: Update From the North-American Shwachman-Diamond Syndrome Registry.. Blood, 2012, 120, 2367-2367.	1.4	1
85	Clinical spectrum and molecular pathophysiology of Shwachmanâ€“Diamond syndrome. Current Opinion in Hematology, 2011, 18, 30-35.	2.5	35
86	Draft consensus guidelines for diagnosis and treatment of Shwachmanâ€Diamond syndrome. Annals of the New York Academy of Sciences, 2011, 1242, 40-55.	3.8	183
87	Pathophysiology and management of inherited bone marrow failure syndromes. Blood Reviews, 2010, 24, 101-122.	5.7	432
88	Stable longâ€term risk of leukaemia in patients with severe congenital neutropenia maintained on Gâ€CSF therapy. British Journal of Haematology, 2010, 150, 196-199.	2.5	211
89	Clinical approach to marrow failure. Hematology American Society of Hematology Education Program, 2009, 2009, 329-337.	2.5	26
90	Shwachman-Diamond Syndrome: A Review of the Clinical Presentation, Molecular Pathogenesis, Diagnosis, and Treatment. Hematology/Oncology Clinics of North America, 2009, 23, 233-248.	2.2	145

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91	Ribosomal dysfunction and inherited marrow failure. British Journal of Haematology, 2008, 141, 376-387.	2.5	53
92	Diagnosing and treating Diamond Blackfan anaemia: results of an international clinical consensus conference. British Journal of Haematology, 2008, 142, 859-876.	2.5	408
93	Mitotic spindle destabilization and genomic instability in Shwachman-Diamond syndrome. Journal of Clinical Investigation, 2008, 118, 1511-1518.	8.2	115
94	Hematopoietic Stem Cell Defects in Novel Fanconi Anemia Mouse Models. Blood, 2008, 112, 440-440.	1.4	1
95	The human Shwachman-Diamond syndrome protein, SBDS, associates with ribosomal RNA. Blood, 2007, 110, 1458-1465.	1.4	136
96	Significance of Telomere Length Measurement in the Diagnosis of Dyskeratosis Congenita.. Blood, 2007, 110, 836-836.	1.4	0
97	Shwachman-Diamond Syndrome. Seminars in Hematology, 2006, 43, 178-188.	3.4	80
98	Inherited Bone Marrow Failure Syndromes: Molecular Features. Hematology American Society of Hematology Education Program, 2006, 2006, 63-71.	2.5	36
99	The Shwachman-Diamond SBDS protein localizes to the nucleolus. Blood, 2005, 106, 1253-1258.	1.4	123
100	A novel diagnostic screen for defects in the Fanconi anemia pathway. Blood, 2002, 100, 4649-4654.	1.4	109