

# Akiko Shimamura

## List of Publications by Year in descending order

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Version: 2024-02-01

100  
papers

6,578  
citations

76326

40  
h-index

66911

78  
g-index

102  
all docs

102  
docs citations

102  
times ranked

6563  
citing authors

#	ARTICLE	IF	CITATIONS
1	International Consensus Classification of Myeloid Neoplasms and Acute Leukemias: integrating morphologic, clinical, and genomic data. <i>Blood</i> , 2022, 140, 1200-1228.	1.4	814
2	Pathophysiology and management of inherited bone marrow failure syndromes. <i>Blood Reviews</i> , 2010, 24, 101-122.	5.7	432
3	Diagnosing and treating Diamond Blackfan anaemia: results of an international clinical consensus conference. <i>British Journal of Haematology</i> , 2008, 142, 859-876.	2.5	408
4	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. <i>Nature Genetics</i> , 2015, 47, 180-185.	21.4	299
5	Stable long-term risk of leukaemia in patients with severe congenital neutropenia maintained on G-CSF therapy. <i>British Journal of Haematology</i> , 2010, 150, 196-199.	2.5	211
6	Draft consensus guidelines for diagnosis and treatment of Shwachman-Diamond syndrome. <i>Annals of the New York Academy of Sciences</i> , 2011, 1242, 40-55.	3.8	183
7	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. <i>Blood</i> , 2016, 127, 1017-1023.	1.4	179
8	Shwachman-Diamond Syndrome: A Review of the Clinical Presentation, Molecular Pathogenesis, Diagnosis, and Treatment. <i>Hematology/Oncology Clinics of North America</i> , 2009, 23, 233-248.	2.2	145
9	Genetic predisposition to hematologic malignancies: management and surveillance. <i>Blood</i> , 2017, 130, 424-432.	1.4	145
10	The human Shwachman-Diamond syndrome protein, SBDS, associates with ribosomal RNA. <i>Blood</i> , 2007, 110, 1458-1465.	1.4	136
11	Ataxia-Pancytopenia Syndrome Is Caused by Missense Mutations in SAMD9L. <i>American Journal of Human Genetics</i> , 2016, 98, 1146-1158.	6.2	136
12	Germline ETV6 Mutations Confer Susceptibility to Acute Lymphoblastic Leukemia and Thrombocytopenia. <i>PLoS Genetics</i> , 2015, 11, e1005262.	3.5	128
13	Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamond-like features. <i>Journal of Clinical Investigation</i> , 2017, 127, 4090-4103.	8.2	126
14	Genetic features of myelodysplastic syndrome and aplastic anemia in pediatric and young adult patients. <i>Haematologica</i> , 2016, 101, 1343-1350.	3.5	124
15	The Shwachman-Diamond SBDS protein localizes to the nucleolus. <i>Blood</i> , 2005, 106, 1253-1258.	1.4	123
16	The diversity of mutations and clinical outcomes for ELANE-associated neutropenia. <i>Current Opinion in Hematology</i> , 2015, 22, 3-11.	2.5	123
17	Variable Clinical Presentation of Shwachman-Diamond Syndrome: Update from the North American Shwachman-Diamond Syndrome Registry. <i>Journal of Pediatrics</i> , 2014, 164, 866-870.	1.8	121
18	Mitotic spindle destabilization and genomic instability in Shwachman-Diamond syndrome. <i>Journal of Clinical Investigation</i> , 2008, 118, 1511-1518.	8.2	115

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19	A novel diagnostic screen for defects in the Fanconi anemia pathway. <i>Blood</i> , 2002, 100, 4649-4654.	1.4	109
20	H/ACA Small RNA Dysfunctions in Disease Reveal Key Roles for Noncoding RNA Modifications in Hematopoietic Stem Cell Differentiation. <i>Cell Reports</i> , 2013, 3, 1493-1502.	6.4	109
21	Recent discoveries in the molecular pathogenesis of the inherited bone marrow failure syndrome Fanconi anemia. <i>Blood Reviews</i> , 2017, 31, 93-99.	5.7	109
22	Genomic analysis of bone marrow failure and myelodysplastic syndromes reveals phenotypic and diagnostic complexity. <i>Haematologica</i> , 2015, 100, 42-48.	3.5	108
23	Marrow failure: a window into ribosome biology. <i>Blood</i> , 2014, 124, 2784-2792.	1.4	105
24	Distinct genetic pathways define pre-malignant versus compensatory clonal hematopoiesis in Shwachman-Diamond syndrome. <i>Nature Communications</i> , 2021, 12, 1334.	12.8	103
25	Genetic predisposition to MDS: clinical features and clonal evolution. <i>Blood</i> , 2019, 133, 1071-1085.	1.4	100
26	Clinical and Molecular Pathophysiology of Shwachman-Diamond Syndrome. <i>Hematology/Oncology Clinics of North America</i> , 2013, 27, 117-128.	2.2	97
27	Somatic mutations and clonal hematopoiesis in congenital neutropenia. <i>Blood</i> , 2018, 131, 408-416.	1.4	91
28	ETV6 in hematopoiesis and leukemia predisposition. <i>Seminars in Hematology</i> , 2017, 54, 98-104.	3.4	87
29	Constitutional <i>SAMD9L</i> mutations cause familial myelodysplastic syndrome and transient monosomy 7. <i>Haematologica</i> , 2018, 103, 427-437.	3.5	83
30	Shwachman-Diamond Syndrome. <i>Seminars in Hematology</i> , 2006, 43, 178-188.	3.4	80
31	Germline Genetic Predisposition to Hematologic Malignancy. <i>Journal of Clinical Oncology</i> , 2017, 35, 1018-1028.	1.6	80
32	Congenital sideroblastic anemia due to mutations in the mitochondrial HSP70 homologue HSPA9. <i>Blood</i> , 2015, 126, 2734-2738.	1.4	78
33	Inherited thrombocytopenia associated with mutation of UDP-galactose-4-epimerase (GALE). <i>Human Molecular Genetics</i> , 2019, 28, 133-142.	2.9	73
34	Clinical features and outcomes of patients with Shwachman-Diamond syndrome and myelodysplastic syndrome or acute myeloid leukaemia: a multicentre, retrospective, cohort study. <i>Lancet Haematology</i> , 2020, 7, e238-e246.	4.6	73
35	Delayed globin synthesis leads to excess heme and the macrocytic anemia of Diamond Blackfan anemia and del(5q) myelodysplastic syndrome. <i>Science Translational Medicine</i> , 2016, 8, 338ra67.	12.4	68
36	Impaired ribosomal subunit association in Shwachman-Diamond syndrome. <i>Blood</i> , 2012, 120, 5143-5152.	1.4	66

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37	Pluripotent Stem Cell Models of Shwachman-Diamond Syndrome Reveal a Common Mechanism for Pancreatic and Hematopoietic Dysfunction. <i>Cell Stem Cell</i> , 2013, 12, 727-736.	11.1	66
38	Ribosomal dysfunction and inherited marrow failure. <i>British Journal of Haematology</i> , 2008, 141, 376-387.	2.5	53
39	Treosulfan-Based Conditioning and Hematopoietic Cell Transplantation for Nonmalignant Diseases: A Prospective Multicenter Trial. <i>Biology of Blood and Marrow Transplantation</i> , 2014, 20, 1996-2003.	2.0	51
40	Allogeneic Hematopoietic Cell Transplantation Using Treosulfan-Based Conditioning for Treatment of Marrow Failure Disorders. <i>Biology of Blood and Marrow Transplantation</i> , 2017, 23, 1669-1677.	2.0	45
41	Immunosuppressive therapy for pediatric aplastic anemia: a North American Pediatric Aplastic Anemia Consortium study. <i>Haematologica</i> , 2019, 104, 1974-1983.	3.5	43
42	Identifying Inherited and Acquired Genetic Factors Involved in Poor Stem Cell Mobilization and Donor-Derived Malignancy. <i>Biology of Blood and Marrow Transplantation</i> , 2016, 22, 2100-2103.	2.0	42
43	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. <i>European Respiratory Journal</i> , 2017, 49, 1601640.	6.7	41
44	Inherited Bone Marrow Failure Syndromes: Molecular Features. <i>Hematology American Society of Hematology Education Program</i> , 2006, 2006, 63-71.	2.5	36
45	Clinical spectrum and molecular pathophysiology of Shwachman-Diamond syndrome. <i>Current Opinion in Hematology</i> , 2011, 18, 30-35.	2.5	35
46	Aplastic anemia and clonal evolution: germ line and somatic genetics. <i>Hematology American Society of Hematology Education Program</i> , 2016, 2016, 74-82.	2.5	35
47	Long-Term Effects of G-CSF Therapy in Cyclic Neutropenia. <i>New England Journal of Medicine</i> , 2017, 377, 2290-2292.	27.0	35
48	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. <i>JAMA Cardiology</i> , 2021, 6, 457.	6.1	34
49	Diagnosis and treatment of pediatric acquired aplastic anemia (AAA): An initial survey of the North American Pediatric Aplastic Anemia Consortium (NAPAAC). <i>Pediatric Blood and Cancer</i> , 2014, 61, 869-874.	1.5	31
50	MYC Promotes Bone Marrow Stem Cell Dysfunction in Fanconi Anemia. <i>Cell Stem Cell</i> , 2021, 28, 33-47.e8.	11.1	31
51	Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene <i>BRCA1</i> . <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 5241-5246.	7.1	29
52	Clinical approach to marrow failure. <i>Hematology American Society of Hematology Education Program</i> , 2009, 2009, 329-337.	2.5	26
53	TGF- $\beta$ signaling underlies hematopoietic dysfunction and bone marrow failure in Shwachman-Diamond syndrome. <i>Journal of Clinical Investigation</i> , 2019, 129, 3821-3826.	8.2	25
54	Bone Marrow Morphology Associated With Germline <i>RUNX1</i> Mutations in Patients With Familial Platelet Disorder With Associated Myeloid Malignancy. <i>Pediatric and Developmental Pathology</i> , 2019, 22, 315-328.	1.0	23

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55	Dissecting ELANE neutropenia pathogenicity by human HSC gene editing. <i>Cell Stem Cell</i> , 2021, 28, 833-845.e5.	11.1	23
56	Hematologic complications with age in Shwachman-Diamond syndrome. <i>Blood Advances</i> , 2022, 6, 297-306.	5.2	23
57	The eIF2-alpha kinase HRI is a novel therapeutic target in multiple myeloma. <i>Leukemia Research</i> , 2017, 55, 23-32.	0.8	22
58	Breast cancer in a case of Shwachman Diamond syndrome. <i>Pediatric Blood and Cancer</i> , 2012, 59, 945-946.	1.5	21
59	Targeted mass spectrometry enables robust quantification of FANCD2 mono-ubiquitination in response to DNA damage. <i>DNA Repair</i> , 2018, 65, 47-53.	2.8	18
60	Neutropenia in the age of genetic testing: Advances and challenges. <i>American Journal of Hematology</i> , 2019, 94, 384-393.	4.1	18
61	Lessons From Pediatric MDS: Approaches to Germline Predisposition to Hematologic Malignancies. <i>Frontiers in Oncology</i> , 2022, 12, 813149.	2.8	16
62	Spindle Microtubule Dysfunction and Cancer Predisposition. <i>Journal of Cellular Physiology</i> , 2014, 229, 1881-1883.	4.1	14
63	Diagnostic workup for severe aplastic anemia in children: Consensus of the <sc>North American Pediatric Aplastic Anemia Consortium</sc>. <i>American Journal of Hematology</i> , 2021, 96, 1491-1504.	4.1	14
64	Genetic predisposition to MDS: diagnosis and management. <i>Hematology American Society of Hematology Education Program</i> , 2019, 2019, 110-119.	2.5	13
65	Pregnancy outcomes in inherited bone marrow failure syndromes. <i>Blood</i> , 2017, 130, 1671-1674.	1.4	12
66	Diagnosis and treatment of pediatric myelodysplastic syndromes: A survey of the North American Pediatric Aplastic Anemia Consortium. <i>Pediatric Blood and Cancer</i> , 2020, 67, e28652.	1.5	12
67	Association of unbalanced translocation der(1;7) with germline GATA2 mutations. <i>Blood</i> , 2021, 138, 2441-2445.	1.4	12
68	A study assessing the feasibility of randomization of pediatric and young adult patients between matched unrelated donor bone marrow transplantation and immune-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. <i>Pediatric Blood and Cancer</i> , 2020, 67, e28444.	1.5	11
69	SLC25A38 congenital sideroblastic anemia: Phenotypes and genotypes of 31 individuals from 24 families, including 11 novel mutations, and a review of the literature. <i>Human Mutation</i> , 2021, 42, 1367-1383.	2.5	11
70	Therapeutic discovery for marrow failure with MDS predisposition using pluripotent stem cells. <i>JCI Insight</i> , 2019, 4, .	5.0	10
71	The frequent and clinically benign anomalies of chromosomes 7 and 20 in Shwachman-diamond syndrome may be subject to further clonal variations. <i>Molecular Cytogenetics</i> , 2021, 14, 54.	0.9	10
72	Inflammatory manifestations in patients with Shwachmanâ€Diamond syndrome: A novel phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1754-1760.	1.2	8

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73	Inhibition of TGF $\beta$ 21 and TGF $\beta$ 23 promotes hematopoiesis in Fanconi anemia. <i>Experimental Hematology</i> , 2021, 93, 70-84.e4.	0.4	8
74	Gene Editing ELANE in Human Hematopoietic Stem and Progenitor Cells Reveals Disease Mechanisms and Therapeutic Strategies for Severe Congenital Neutropenia. <i>Blood</i> , 2019, 134, 3-3.	1.4	8
75	Autophagy in mesenchymal progenitors protects mice against bone marrow failure after severe intermittent stress. <i>Blood</i> , 2022, 139, 690-703.	1.4	8
76	Repolarization of HSC attenuates HSCs failure in Shwachman-Diamond syndrome. <i>Leukemia</i> , 2021, 35, 1751-1762.	7.2	5
77	It's ALL in the Family: IKZF1 and Hereditary Leukemia. <i>Cancer Cell</i> , 2018, 33, 798-800.	16.8	4
78	Metformin for treatment of cytopenias in children and young adults with Fanconi anemia. <i>Blood Advances</i> , 2022, 6, 3803-3811.	5.2	4
79	Low frequency of treatable pediatric disease alleles in gnomAD: An opportunity for future genomic screening of newborns. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100059.	1.7	3
80	The Activated TGF $\beta$ 2 Pathway in Shwachman Diamond Syndrome Impairs Hematopoiesis and Is Down-Regulated By Deletion of 7q. <i>Blood</i> , 2017, 130, 875-875.	1.4	3
81	Translational research for bone marrow failure patients. <i>Experimental Hematology</i> , 2021, , .	0.4	3
82	Coronavirus disease 2019 and vaccination in patients with Shwachman-Diamond syndrome. <i>Pediatric Blood and Cancer</i> , 2022, 69, e29647.	1.5	3
83	Molecular alterations governing predisposition to myelodysplastic syndromes: Insights from Shwachman-Diamond syndrome. <i>Best Practice and Research in Clinical Haematology</i> , 2021, 34, 101252.	1.7	2
84	Mutation Burden in Hematopoietic Stem Cells Is Not Increased in Congenital Neutropenia. <i>Blood</i> , 2016, 128, 405-405.	1.4	2
85	Pancreatic lipomatosis in Diamond-Blackfan anemia: The importance of genetic testing in bone marrow failure disorders. <i>American Journal of Hematology</i> , 2018, 93, 1194-1195.	4.1	1
86	Hematopoietic Stem Cell Defects in Novel Fanconi Anemia Mouse Models. <i>Blood</i> , 2008, 112, 440-440.	1.4	1
87	Variable Clinical Presentation of Shwachman-Diamond Syndrome: Update From the North-American Shwachman-Diamond Syndrome Registry.. <i>Blood</i> , 2012, 120, 2367-2367.	1.4	1
88	Modeling Bone Marrow Failure and MDS in Shwachman Diamond Syndrome Using Induced Pluripotent Stem Cells. <i>Blood</i> , 2016, 128, 1496-1496.	1.4	1
89	Congenital X-linked Neutropenia with Myelodysplasia and Somatic Tetraploidy due to a Germline Mutation in SEPT6. <i>American Journal of Hematology</i> , 2021, , .	4.1	1
90	An induced pluripotent stem cell model of Fanconi anemia reveals mechanisms of p53-driven progenitor cell differentiation. <i>Blood Advances</i> , 2020, 4, 4679-4692.	5.2	1

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91	Significance of Telomere Length Measurement in the Diagnosis of Dyskeratosis Congenita.. Blood, 2007, 110, 836-836.	1.4	0
92	The North American Shwachman-Diamond Syndrome Registry: Genetically Undefined Shwachman-Diamond Syndrome. Blood, 2015, 126, 3614-3614.	1.4	0
93	Identifying Inherited and Acquired Genetic Factors Involved in Poor Stem Cell Mobilization and Donor-Derived Malignancy. Blood, 2015, 126, 3163-3163.	1.4	0
94	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
95	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
96	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
97	Germline and Somatic Genetic Characterization of Shwachman-Diamond Syndrome. Blood, 2016, 128, 2681-2681.	1.4	0
98	Hyperactive Non-Canonical TGF- $\beta$ 2 Pathway Signaling in Fanconi Anemia Bone Marrow Stromal Cells Contributes to Growth Suppression. Blood, 2016, 128, 1039-1039.	1.4	0
99	Towards Identifying the Target of Autoimmunity in Aplastic Anemia. Blood, 2020, 136, 2-2.	1.4	0
100	Telomere biology disorders: ends and (genetic) means. Blood, 2022, 139, 1776-1777.	1.4	0