Marshall S Horwitz

List of Publications by Year
in descending order

100 papers	8,582 citations	87888 38 h-index	45317 90 g-index
103 all docs	103 docs citations	103 times ranked	11604 citing authors

#	Article	IF	Citations
1	Simultaneous brain cell type and lineage determined by scRNA-seq reveals stereotyped cortical development. Cell Systems, 2022, 13, 438-453.e5.	6.2	2
2	Restoring RUNX1 deficiency in RUNX1 familial platelet disorder by inhibiting its degradation. Blood Advances, 2021, 5, 687-699.	5.2	12
3	Neutrophil elastase: Nonsense lost in translation. Cell Stem Cell, 2021, 28, 790-792.	11.1	1
4	RUNX1-mutated families show phenotype heterogeneity and a somatic mutation profile unique to germline predisposed AML. Blood Advances, 2020, 4, 1131-1144.	5.2	102
5	Inducible expression of a disease-associated ELANE mutation impairs granulocytic differentiation, without eliciting an unfolded protein response. Journal of Biological Chemistry, 2020, 295, 7492-7500.	3.4	16
6	Normal peripheral blood neutrophil numbers accompanying ELANE whole gene deletion mutation. Blood Advances, 2019, 3, 2470-2473.	5.2	10
7	Activating PAX gene family paralogs to complement PAX5 leukemia driver mutations. PLoS Genetics, 2018, 14, e1007642.	3.5	3
8	Acute lymphoblastic leukemia in a child with Leri-Weill syndrome and complete SHOX gene deletion: A Case Report. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2018, 162, 65-70.	0.6	2
9	GATA factor mutations in hematologic disease. Blood, 2017, 129, 2103-2110.	1.4	149
10	Prolonged pharmacological inhibition of cathepsin C results in elimination of neutrophil serine proteases. Biochemical Pharmacology, 2017, 131, 52-67.	4.4	34
11	GATA2 deficiency and related myeloid neoplasms. Seminars in Hematology, 2017, 54, 81-86.	3.4	125
12	Case Report of an Adolescent Male With Unexplained Pancytopenia: GATA2-Associated Bone Marrow Failure and Genetic Testing. Global Pediatric Health, 2017, 4, 2333794X1774494.	0.7	2
13	Genome sequencing in a case of Niemannâ€“Pick type C. Journal of Physical Education and Sports Management, 2016, 2, a001222.	1.2	10
14	Whole-organism lineage tracing by combinatorial and cumulative genome editing. Science, 2016, 353, aaf7907.	12.6	570
15	Expanded Phenotypic and Genetic Heterogeneity in the Clinical Spectrum of FPD-AML: Lymphoid Malignancies and Skin Disorders Are Common Features in Carriers of Germline RUNX1 Mutations. Blood, 2016, 128, 1212-1212.	1.4	2
16	Mechanisms and clinical applications of chromosomal instability in lymphoid malignancy. British Journal of Haematology, 2015, 171, 13-28.	2.5	27
17	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. Nature Genetics, 2015, 47, 180-185.	21.4	299
18	Pathogenesis of ELANE-mutant severe neutropenia revealed by induced pluripotent stem cells. Journal of Clinical Investigation, 2015, 125, 3103-3116.	8.2	62

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19	Co-Opting the Genetic Redundancy of the Pax Family to Treat PAX5 Mutated Pre-B ALL. Blood, 2015, 126, 2445-2445.	1.4	0
20	GATA2 deficiency: flesh and blood. Blood, 2014, 123, 799-800.	1.4	8
21	Heritable GATA2 mutations associated with familial AML-MDS: a case report and review of literature. Journal of Hematology and Oncology, 2014, 7, 36.	17.0	25
22	Neutropenia-associated ELANE mutations disrupting translation initiation produce novel neutrophil elastase isoforms. Blood, 2014, 123, 562-569.	1.4	38
23	Targeted correction of RUNX1 mutation in FPD patient-specific induced pluripotent stem cells rescues megakaryopoietic defects. Blood, 2014, 124, 1926-1930.	1.4	67
24	Genome Sequencing of Idiopathic Pulmonary Fibrosis in Conjunction with a Medical School Human Anatomy Course. PLoS ONE, 2014, 9, e106744.	2.5	9
25	Use of somatic mutations to quantify random contributions to mouse development. BMC Genomics, 2013, 14, 39.	2.8	13
26	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 1226-1231.	21.4	270
27	ELANE Mutations in Cyclic and Severe Congenital Neutropenia. Hematology/Oncology Clinics of North America, 2013, 27, 19-41.	2.2	105
28	Support for the N -Methyl-D-Aspartate Receptor Hypofunction Hypothesis of Schizophrenia From Exome Sequencing in Multiplex Families. JAMA Psychiatry, 2013, 70, 582.	11.0	119
29	Clonal Expansions and Short Telomeres Are Associated with Neoplasia in Early-onset, but not Late-onset, Ulcerative Colitis. Inflammatory Bowel Diseases, 2013, 19, 2593-2602.	1.9	23
30	Mitotic errors, aneuploidy and micronuclei in Hodgkin lymphoma pathogenesis. Communicative and Integrative Biology, 2013, 6, e23544.	1.4	4
31	The Kelch Protein KLHDC8B Guards against Mitotic Errors, Centrosomal Amplification, and Chromosomal Instability. Journal of Biological Chemistry, 2012, 287, 39083-39093.	3.4	18
32	Loss-of-function germline GATA2 mutations in patients with MDS/AML or MonoMAC syndrome and primary lymphedema reveal a key role for GATA2 in the lymphatic vasculature. Blood, 2012, 119, 1283-1291.	1.4	244
33	Decoding cell lineage from acquired mutations using arbitrary deep sequencing. Nature Methods, 2012, 9, 78-80.	19.0	39
34	Target protein interactions of indoleâ€3â€carbinol and the highly potent derivative 1â€benzylâ€3C with the Câ€terminal domain of human elastase uncouples cell cycle arrest from apoptotic signaling. Molecular Carcinogenesis, 2012, 51, 881-894.	2.7	11
35	HIF1Î± induced switch from bivalent to exclusively glycolytic metabolism during ESC-to-EpiSC/hESC transition. EMBO Journal, 2012, 31, 2103-2116.	7.8	480
36	Novel Isoforms of Neutrophil Elastase Produced by Neutropenia-Associated Mutations of the Initiation Codon and an Internal Ribosomal Entry Site (IRES) of ELANE. Blood, 2012, 120, 9-9.	1.4	15

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37	Towards Cell Therapy for FPD/AML with Patient-Specific IPS Cells. Blood, 2012, 120, 4677-4677.	1.4	0
38	Heritable GATA2 mutations associated with familial myelodysplastic syndrome and acute myeloid leukemia. Nature Genetics, 2011, 43, 1012-1017.	21.4	524
39	HIF Induces Human Embryonic Stem Cell Markers in Cancer Cells. Cancer Research, 2011, 71, 4640-4652.	0.9	473
40	Assessment of Hypoxia Inducible Factor Levels in Cancer Cell Lines upon Hypoxic Induction Using a Novel Reporter Construct. PLoS ONE, 2011, 6, e27460.	2.5	36
41	Passenger mutations as a marker of clonal cell lineages in emerging neoplasia. Seminars in Cancer Biology, 2010, 20, 294-303.	9.6	27
42	Phylogenetic analysis of developmental and postnatal mouse cell lineages. Evolution & Development, 2010, 12, 84-94.	2.0	34
43	Poor prognosis in familial acute myeloid leukaemia with combined biallelic CEBPA mutations and downstream events affecting the ATM, FLT3 and CDX2 genes. British Journal of Haematology, 2010, 150, 382-385.	2.5	17
44	KLHDC8Bin Hodgkin lymphoma and possibly twinning. Communicative and Integrative Biology, 2010, 3, 154-158.	1.4	4
45	Mutations in a gene encoding a midbody protein in binucleated Reed-Sternberg cells of Hodgkin lymphoma. Cell Cycle, 2010, 9, 670-675.	2.6	10
46	Neutrophil Elastase, Proteinase 3, and Cathepsin G as Therapeutic Targets in Human Diseases. Pharmacological Reviews, 2010, 62, 726-759.	16.0	676
47	GATA2 is a New Predisposition Gene for Familial Myelodysplastic Syndrome (MDS) and Acute Myeloid Leukemia (AML). Blood, 2010, 116, LBA-3-LBA-3.	1.4	10
48	Clonal expansions in ulcerative colitis identify patients with neoplasia. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 20871-20876.	7.1	58
49	Contributions to Neutropenia from PFAAP5 (N4BP2L2), a Novel Protein Mediating Transcriptional Repressor Cooperation between Gfi1 and Neutrophil Elastase. Molecular and Cellular Biology, 2009, 29, 4394-4405.	2.3	35
50	Mutations in a gene encoding a midbody kelch protein in familial and sporadic classical Hodgkin lymphoma lead to binucleated cells. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 14920-14925.	7.1	59
51	Lymphadenopathy as the primary manifestation of malignant transformation in two patients with severe congenital neutropenia. Pediatric Blood and Cancer, 2008, 50, 1072-1075.	1.5	0
52	Neutropenia in 6 ethnic groups from the Caribbean and the U.S.. Cancer, 2008, 113, 854-860.	4.1	29
53	Duffy (Fy), DARC, and neutropenia among women from the United States, Europe and the Caribbean. British Journal of Haematology, 2008, 143, 288-293.	2.5	55
54	Integrative analysis of RUNX1 downstream pathways and target genes. BMC Genomics, 2008, 9, 363.	2.8	116

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55	Phylogenetic Fate Mapping: Theoretical and Experimental Studies Applied to the Development of Mouse Fibroblasts. Genetics, 2008, 178, 967-977.	2.9	26
56	A Phylogenetic Approach to Mapping Cell Fate. Current Topics in Developmental Biology, 2007, 79, 157-184.	2.2	27
57	Epigenetic Regulation of Protein-Coding and MicroRNA Genes by the Gfi1-Interacting Tumor Suppressor PRDM5. Molecular and Cellular Biology, 2007, 27, 6889-6902.	2.3	79
58	Neutrophil elastase in cyclic and severe congenital neutropenia. Blood, 2007, 109, 1817-1824.	1.4	221
59	Double de novo mutations ofELA2 in cyclic and severe congenital neutropenia. Human Mutation, 2007, 28, 874-881.	2.5	33
60	The Clinical, Immunohematological, and Molecular Study of Iranian Patients with Severe Congenital Neutropenia. Journal of Clinical Immunology, 2007, 27, 525-533.	3.8	38
61	Familial leukemia. Best Practice and Research in Clinical Haematology, 2006, 19, 269-279.	1.7	4
62	Phylogenetic fate mapping. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 5448-5453.	7.1	90
63	KLHDC8B Is a Novel, Mitotically-Regulated Classical Hodgkinâ€™s Lymphoma Candidate Susceptibility Gene.. Blood, 2006, 108, 473-473.	1.4	4
64	Neutropenia-Associated Mutations in PRDM5, a Novel Epigenetic Regulator of Hematopoiesis.. Blood, 2006, 108, 503-503.	1.4	1
65	A 17q25.3 Duplication Defines a New Dosage-Sensitive Congenital Neutropenia Locus and Implicates SOCS3 as a Candidate Gene for Cases Unexplained by ELA2 Mutation.. Blood, 2006, 108, 1277-1277.	1.4	0
66	Neutrophil elastase unleashed. Blood, 2005, 105, 3392-3393.	1.4	3
67	G-CSF receptor mutations mix it up. Blood, 2005, 105, 440-440.	1.4	0
68	Gfi1 Coordinates Epigenetic Repression of p21^{Cip/WAF1} by Recruitment of Histone Lysine Methyltransferase G9a and Histone Deacetylase 1. Molecular and Cellular Biology, 2005, 25, 10338-10351.	2.3	157
69	Gfi-1 takes center stage in hematopoietic stem cells. Trends in Molecular Medicine, 2005, 11, 49-52.	6.7	30
70	Mechanisms of dominant congenital neutropenias. Drug Discovery Today Disease Mechanisms, 2005, 2, 471-477.	0.8	1
71	Targeted transcriptional repression of Gfi1 by GFI1 and GFI1B in lymphoid cells. Nucleic Acids Research, 2004, 32, 2508-2519.	14.5	74
72	A Novel Notch Protein, N2N, Targeted by Neutrophil Elastase and Implicated in Hereditary Neutropenia. Molecular and Cellular Biology, 2004, 24, 58-70.	2.3	57

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73	Congenital and Acquired Neutropenia. Hematology American Society of Hematology Education Program, 2004, 2004, 63-79.	2.5	112
74	Lymphoid Enhancer Factor-1 Links Two Hereditary Leukemia Syndromes through Core-binding Factor Î± Regulation of ELA2. Journal of Biological Chemistry, 2004, 279, 2873-2884.	3.4	36
75	Enteritis Necroticans with Recurrent Enterocutaneous Fistulae Caused by Clostridium perfringens in a Child With Cyclic Neutropenia. Journal of Pediatric Gastroenterology and Nutrition, 2004, 38, 213-215.	1.8	10
76	Paradoxical homozygous expression from heterozygotes and heterozygous expression from homozygotes as a consequence of transcriptional infidelity through a polyadenine tract in the AP3B1 gene responsible for canine cyclic neutropenia. Nucleic Acids Research, 2004, 32, 6327-6333.	14.5	21
77	Chromosome band 16q22-linked familial AML: Exclusion of candidate genes, and possible disease risk modification byNQO1 polymorphisms. Genes Chromosomes and Cancer, 2004, 41, 278-282.	2.8	4
78	Hereditary neutropenia: dogs explain human neutrophil elastase mutations. Trends in Molecular Medicine, 2004, 10, 163-170.	6.7	72
79	Acquired Mutation and Reversion of the Neutrophil Elastase Gene, ELA2, in Kostmann Syndrome and Cyclic Neutropenia Suggest that Some Mutations Cause Neutropenia while Others Paradoxically Suppress It.. Blood, 2004, 104, 781-781.	1.4	0
80	Mutations in proto-oncogene GFI1 cause human neutropenia and target ELA2. Nature Genetics, 2003, 34, 308-312.	21.4	350
81	Mutations associated with neutropenia in dogs and humans disrupt intracellular transport of neutrophil elastase. Nature Genetics, 2003, 35, 90-96.	21.4	163
82	Leukemia in Severe Congenital Neutropenia: Defective Proteolysis Suggests New Pathways to Malignancy and Opportunities for Therapy. Cancer Investigation, 2003, 21, 579-587.	1.3	24
83	Targets of the transcriptional repressor oncoprotein Gfi-1. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 5932-5937.	7.1	93
84	Gfi-1 Oncoproteins in Hematopoiesis. Hematology, 2003, 8, 339-344.	1.5	41
85	Role of neutrophil elastase in bone marrow failure syndromes: molecular genetic revival of the chalone hypothesis. Current Opinion in Hematology, 2003, 10, 49-54.	2.5	35
86	Low levels of serum elastase are not associated with mutations in ELA-2 elastase encoding gene in chronic idiopathic neutropenia. Blood, 2003, 101, 2898-2898.	1.4	8
87	Possibility of somatic mosaicism of ELA2 mutation overlooked in an asymptomatic father transmitting severe congenital neutropenia to two offspring. British Journal of Haematology, 2002, 118, 923-923.	2.5	6
88	In vitro analyses of known and novel RUNX1/AML1 mutations in dominant familial platelet disorder with predisposition to acute myelogenous leukemia: implications for mechanisms of pathogenesis. Blood, 2002, 99, 1364-1372.	1.4	348
89	Mice expressing a neutrophil elastase mutation derived from patients with severe congenital neutropenia have normal granulopoiesis. Blood, 2002, 100, 3221-3228.	1.4	65
90	Evidence for the involvement of cathepsin B in skeletal myoblast differentiation. Journal of Cellular Biochemistry, 2002, 84, 520-531.	2.6	20

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91	Characterization of Mutant Neutrophil Elastase in Severe Congenital Neutropenia. Journal of Biological Chemistry, 2001, 276, 14230-14241.	3.4	81
92	Susceptibility gene for familial acute myeloid leukemia associated with loss of 5q and/or 7q is not localized on the commonly deleted portion of 5q. , 2000, 28, 164-172.		18
93	Mutations in the gene encoding neutrophil elastase in congenital and cyclic neutropenia. Blood, 2000, 96, 2317-2322.	1.4	529
94	Selection of a Dominant Negative Retinoblastoma Protein (RB) Inhibiting Satellite Myoblast Differentiation Implies an Indirect Interaction between MyoD and RB. Molecular and Cellular Biology, 2000, 20, 5129-5139.	2.3	30
95	Mutations in the gene encoding neutrophil elastase in congenital and cyclic neutropenia. Blood, 2000, 96, 2317-2322.	1.4	11
96	Mutations in ELA2, encoding neutrophil elastase, define a 21-day biological clock in cyclic haematopoiesis. Nature Genetics, 1999, 23, 433-436.	21.4	444
97	Pseudoautosomal Linkage of Hodgkin Disease. American Journal of Human Genetics, 1999, 65, 1413-1422.	6.2	58
98	Genetic Heterogeneity in Familial Acute Myelogenous Leukemia: Evidence for a Second Locus at Chromosome 16q21-23.2. American Journal of Human Genetics, 1997, 61, 873-881.	6.2	42
99	Hypermethylated Myoblasts Specifically Deficient in MyoD Autoactivation as a Consequence of Instability of MyoD. Experimental Cell Research, 1996, 226, 170-182.	2.6	8
100	A family inheriting different subtypes of acute myelogenous leukemia. , 1996, 52, 295-304.		22