

Marshall S Horwitz

List of Publications by Year in descending order

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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	Neutrophil Elastase, Proteinase 3, and Cathepsin G as Therapeutic Targets in Human Diseases. <i>Pharmacological Reviews</i> , 2010, 62, 726-759.	16.0	676
2	Whole-organism lineage tracing by combinatorial and cumulative genome editing. <i>Science</i> , 2016, 353, aaf7907.	12.6	570
3	Mutations in the gene encoding neutrophil elastase in congenital and cyclic neutropenia. <i>Blood</i> , 2000, 96, 2317-2322.	1.4	529
4	Heritable GATA2 mutations associated with familial myelodysplastic syndrome and acute myeloid leukemia. <i>Nature Genetics</i> , 2011, 43, 1012-1017.	21.4	524
5	HIF1 α induced switch from bivalent to exclusively glycolytic metabolism during ESC-to-EpiSC/hESC transition. <i>EMBO Journal</i> , 2012, 31, 2103-2116.	7.8	480
6	HIF Induces Human Embryonic Stem Cell Markers in Cancer Cells. <i>Cancer Research</i> , 2011, 71, 4640-4652.	0.9	473
7	Mutations in ELA2, encoding neutrophil elastase, define a 21-day biological clock in cyclic haematopoiesis. <i>Nature Genetics</i> , 1999, 23, 433-436.	21.4	444
8	Mutations in proto-oncogene GFI1 cause human neutropenia and target ELA2. <i>Nature Genetics</i> , 2003, 34, 308-312.	21.4	350
9	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. <i>Blood</i> , 2002, 99, 1364-1372.	1.4	348
10	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. <i>Nature Genetics</i> , 2015, 47, 180-185.	21.4	299
11	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. <i>Nature Genetics</i> , 2013, 45, 1226-1231.	21.4	270
12	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. <i>Blood</i> , 2012, 119, 1283-1291.	1.4	244
13	Neutrophil elastase in cyclic and severe congenital neutropenia. <i>Blood</i> , 2007, 109, 1817-1824.	1.4	221
14	Mutations associated with neutropenia in dogs and humans disrupt intracellular transport of neutrophil elastase. <i>Nature Genetics</i> , 2003, 35, 90-96.	21.4	163
15	Cf1 Coordinates Epigenetic Repression of <i>p21^{Cip/WAF1}</i> by Recruitment of Histone Lysine Methyltransferase G9a and Histone Deacetylase 1. <i>Molecular and Cellular Biology</i> , 2005, 25, 10338-10351.	2.3	157
16	GATA factor mutations in hematologic disease. <i>Blood</i> , 2017, 129, 2103-2110.	1.4	149
17	GATA2 deficiency and related myeloid neoplasms. <i>Seminars in Hematology</i> , 2017, 54, 81-86.	3.4	125
18	Support for the N-Methyl-D-Aspartate Receptor Hypofunction Hypothesis of Schizophrenia From Exome Sequencing in Multiplex Families. <i>JAMA Psychiatry</i> , 2013, 70, 582.	11.0	119

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19	Integrative analysis of RUNX1 downstream pathways and target genes. BMC Genomics, 2008, 9, 363.	2.8	116
20	Congenital and Acquired Neutropenia. Hematology American Society of Hematology Education Program, 2004, 2004, 63-79.	2.5	112
21	ELANE Mutations in Cyclic and Severe Congenital Neutropenia. Hematology/Oncology Clinics of North America, 2013, 27, 19-41.	2.2	105
22	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
23	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
24	Phylogenetic fate mapping. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 5448-5453.	7.1	90
25	Characterization of Mutant Neutrophil Elastase in Severe Congenital Neutropenia. Journal of Biological Chemistry, 2001, 276, 14230-14241.	3.4	81
26	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
27	Targeted transcriptional repression of Gfi1 by GFI1 and GFI1B in lymphoid cells. Nucleic Acids Research, 2004, 32, 2508-2519.	14.5	74
28	Hereditary neutropenia: dogs explain human neutrophil elastase mutations. Trends in Molecular Medicine, 2004, 10, 163-170.	6.7	72
29	Targeted correction of RUNX1 mutation in FPD patient-specific induced pluripotent stem cells rescues megakaryopoietic defects. Blood, 2014, 124, 1926-1930.	1.4	67
30	Mice expressing a neutrophil elastase mutation derived from patients with severe congenital neutropenia have normal granulopoiesis. Blood, 2002, 100, 3221-3228.	1.4	65
31	Pathogenesis of ELANE-mutant severe neutropenia revealed by induced pluripotent stem cells. Journal of Clinical Investigation, 2015, 125, 3103-3116.	8.2	62
32	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
33	Pseudoautosomal Linkage of Hodgkin Disease. American Journal of Human Genetics, 1999, 65, 1413-1422.	6.2	58
34	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
35	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
36	Duffy (Fy), <i>DARC</i> , and neutropenia among women from the United States, Europe and the Caribbean. British Journal of Haematology, 2008, 143, 288-293.	2.5	55

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37	Genetic Heterogeneity in Familial Acute Myelogenous Leukemia: Evidence for a Second Locus at Chromosome 16q21-23.2. <i>American Journal of Human Genetics</i> , 1997, 61, 873-881.	6.2	42
38	Gfi-1 Oncoproteins in Hematopoiesis. <i>Hematology</i> , 2003, 8, 339-344.	1.5	41
39	Decoding cell lineage from acquired mutations using arbitrary deep sequencing. <i>Nature Methods</i> , 2012, 9, 78-80.	19.0	39
40	The Clinical, Immunohematological, and Molecular Study of Iranian Patients with Severe Congenital Neutropenia. <i>Journal of Clinical Immunology</i> , 2007, 27, 525-533.	3.8	38
41	Neutropenia-associated ELANE mutations disrupting translation initiation produce novel neutrophil elastase isoforms. <i>Blood</i> , 2014, 123, 562-569.	1.4	38
42	Lymphoid Enhancer Factor-1 Links Two Hereditary Leukemia Syndromes through Core-binding Factor $\hat{1}\pm$ Regulation of ELA2. <i>Journal of Biological Chemistry</i> , 2004, 279, 2873-2884.	3.4	36
43	Assessment of Hypoxia Inducible Factor Levels in Cancer Cell Lines upon Hypoxic Induction Using a Novel Reporter Construct. <i>PLoS ONE</i> , 2011, 6, e27460.	2.5	36
44	Role of neutrophil elastase in bone marrow failure syndromes: molecular genetic revival of the chalone hypothesis. <i>Current Opinion in Hematology</i> , 2003, 10, 49-54.	2.5	35
45	Contributions to Neutropenia from PFAAP5 (N4BP2L2), a Novel Protein Mediating Transcriptional Repressor Cooperation between Gfi1 and Neutrophil Elastase. <i>Molecular and Cellular Biology</i> , 2009, 29, 4394-4405.	2.3	35
46	Phylogenetic analysis of developmental and postnatal mouse cell lineages. <i>Evolution & Development</i> , 2010, 12, 84-94.	2.0	34
47	Prolonged pharmacological inhibition of cathepsin C results in elimination of neutrophil serine proteases. <i>Biochemical Pharmacology</i> , 2017, 131, 52-67.	4.4	34
48	Double de novo mutations of ELA2 in cyclic and severe congenital neutropenia. <i>Human Mutation</i> , 2007, 28, 874-881.	2.5	33
49	Selection of a Dominant Negative Retinoblastoma Protein (RB) Inhibiting Satellite Myoblast Differentiation Implies an Indirect Interaction between MyoD and RB. <i>Molecular and Cellular Biology</i> , 2000, 20, 5129-5139.	2.3	30
50	Gfi-1 takes center stage in hematopoietic stem cells. <i>Trends in Molecular Medicine</i> , 2005, 11, 49-52.	6.7	30
51	Neutropenia in 6 ethnic groups from the Caribbean and the U.S.. <i>Cancer</i> , 2008, 113, 854-860.	4.1	29
52	A Phylogenetic Approach to Mapping Cell Fate. <i>Current Topics in Developmental Biology</i> , 2007, 79, 157-184.	2.2	27
53	Passenger mutations as a marker of clonal cell lineages in emerging neoplasia. <i>Seminars in Cancer Biology</i> , 2010, 20, 294-303.	9.6	27
54	Mechanisms and clinical applications of chromosomal instability in lymphoid malignancy. <i>British Journal of Haematology</i> , 2015, 171, 13-28.	2.5	27

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55	Phylogenetic Fate Mapping: Theoretical and Experimental Studies Applied to the Development of Mouse Fibroblasts. <i>Genetics</i> , 2008, 178, 967-977.	2.9	26
56	Heritable GATA2 mutations associated with familial AML-MDS: a case report and review of literature. <i>Journal of Hematology and Oncology</i> , 2014, 7, 36.	17.0	25
57	Leukemia in Severe Congenital Neutropenia: Defective Proteolysis Suggests New Pathways to Malignancy and Opportunities for Therapy. <i>Cancer Investigation</i> , 2003, 21, 579-587.	1.3	24
58	Clonal Expansions and Short Telomeres Are Associated with Neoplasia in Early-onset, but not Late-onset, Ulcerative Colitis. <i>Inflammatory Bowel Diseases</i> , 2013, 19, 2593-2602.	1.9	23
59	A family inheriting different subtypes of acute myelogenous leukemia. , 1996, 52, 295-304.		22
60	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. <i>Nucleic Acids Research</i> , 2004, 32, 6327-6333.	14.5	21
61	Evidence for the involvement of cathepsin B in skeletal myoblast differentiation. <i>Journal of Cellular Biochemistry</i> , 2002, 84, 520-531.	2.6	20
62	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
63	The Kelch Protein KLHDC8B Guards against Mitotic Errors, Centrosomal Amplification, and Chromosomal Instability. <i>Journal of Biological Chemistry</i> , 2012, 287, 39083-39093.	3.4	18
64	Poor prognosis in familial acute myeloid leukaemia with combined biallelic <i>CEBPA</i> mutations and downstream events affecting the <i>ATM</i> , <i>FLT3</i> and <i>CDX2</i> genes. <i>British Journal of Haematology</i> , 2010, 150, 382-385.	2.5	17
65	Inducible expression of a disease-associated ELANE mutation impairs granulocytic differentiation, without eliciting an unfolded protein response. <i>Journal of Biological Chemistry</i> , 2020, 295, 7492-7500.	3.4	16
66	Novel Isoforms of Neutrophil Elastase Produced by Neutropenia-Associated Mutations of the Initiation Codon and an Internal Ribosomal Entry Site (IRES) of ELANE. <i>Blood</i> , 2012, 120, 9-9.	1.4	15
67	Use of somatic mutations to quantify random contributions to mouse development. <i>BMC Genomics</i> , 2013, 14, 39.	2.8	13
68	Restoring RUNX1 deficiency in <i>RUNX1</i> familial platelet disorder by inhibiting its degradation. <i>Blood Advances</i> , 2021, 5, 687-699.	5.2	12
69	Target protein interactions of indole-3-carbinol and the highly potent derivative 1-benzyl-3C with the C-terminal domain of human elastase uncouples cell cycle arrest from apoptotic signaling. <i>Molecular Carcinogenesis</i> , 2012, 51, 881-894.	2.7	11
70	Mutations in the gene encoding neutrophil elastase in congenital and cyclic neutropenia. <i>Blood</i> , 2000, 96, 2317-2322.	1.4	11
71	Enteritis Necroticans with Recurrent Enterocutaneous Fistulae Caused by <i>Clostridium perfringens</i> in a Child With Cyclic Neutropenia. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2004, 38, 213-215.	1.8	10
72	Mutations in a gene encoding a midbody protein in binucleated Reed-Sternberg cells of Hodgkin lymphoma. <i>Cell Cycle</i> , 2010, 9, 670-675.	2.6	10

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73	Genome sequencing in a case of Niemann-Pick type C. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a001222.	1.2	10
74	Normal peripheral blood neutrophil numbers accompanying ELANE whole gene deletion mutation. <i>Blood Advances</i> , 2019, 3, 2470-2473.	5.2	10
75	GATA2 is a New Predisposition Gene for Familial Myelodysplastic Syndrome (MDS) and Acute Myeloid Leukemia (AML). <i>Blood</i> , 2010, 116, LBA-3-LBA-3.	1.4	10
76	Genome Sequencing of Idiopathic Pulmonary Fibrosis in Conjunction with a Medical School Human Anatomy Course. <i>PLoS ONE</i> , 2014, 9, e106744.	2.5	9
77	Hypermethylated Myoblasts Specifically Deficient in MyoD Autoactivation as a Consequence of Instability of MyoD. <i>Experimental Cell Research</i> , 1996, 226, 170-182.	2.6	8
78	Low levels of serum elastase are not associated with mutations in ELA-2 elastase encoding gene in chronic idiopathic neutropenia. <i>Blood</i> , 2003, 101, 2898-2898.	1.4	8
79	GATA2 deficiency: flesh and blood. <i>Blood</i> , 2014, 123, 799-800.	1.4	8
80	Possibility of somatic mosaicism of ELA2 mutation overlooked in an asymptomatic father transmitting severe congenital neutropenia to two offspring. <i>British Journal of Haematology</i> , 2002, 118, 923-923.	2.5	6
81	Chromosome band 16q22-linked familial AML: Exclusion of candidate genes, and possible disease risk modification byNQO1 polymorphisms. <i>Genes Chromosomes and Cancer</i> , 2004, 41, 278-282.	2.8	4
82	Familial leukemia. <i>Best Practice and Research in Clinical Haematology</i> , 2006, 19, 269-279.	1.7	4
83	<i>KLHDC8B</i> in Hodgkin lymphoma and possibly twinning. <i>Communicative and Integrative Biology</i> , 2010, 3, 154-158.	1.4	4
84	Mitotic errors, aneuploidy and micronuclei in Hodgkin lymphoma pathogenesis. <i>Communicative and Integrative Biology</i> , 2013, 6, e23544.	1.4	4
85	<i>KLHDC8B</i> Is a Novel, Mitotically-Regulated Classical Hodgkin's Lymphoma Candidate Susceptibility Gene. <i>Blood</i> , 2006, 108, 473-473.	1.4	4
86	Neutrophil elastase unleashed. <i>Blood</i> , 2005, 105, 3392-3393.	1.4	3
87	Activating PAX gene family paralogs to complement PAX5 leukemia driver mutations. <i>PLoS Genetics</i> , 2018, 14, e1007642.	3.5	3
88	Case Report of an Adolescent Male With Unexplained Pancytopenia: <i>GATA2</i> -Associated Bone Marrow Failure and Genetic Testing. <i>Global Pediatric Health</i> , 2017, 4, 2333794X1774494.	0.7	2
89	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. <i>Blood</i> , 2016, 128, 1212-1212.	1.4	2
90	Acute lymphoblastic leukemia in a child with Leri-Weill syndrome and complete SHOX gene deletion: A Case Report. <i>Biomedical Papers of the Medical Faculty of the University Palacky&#x0301;, Olomouc, Czechoslovakia</i> , 2018, 162, 65-70.	0.6	2

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91	Simultaneous brain cell type and lineage determined by scRNA-seq reveals stereotyped cortical development. <i>Cell Systems</i> , 2022, 13, 438-453.e5.	6.2	2
92	Mechanisms of dominant congenital neutropenias. <i>Drug Discovery Today Disease Mechanisms</i> , 2005, 2, 471-477.	0.8	1
93	Neutrophil elastase: Nonsense lost in translation. <i>Cell Stem Cell</i> , 2021, 28, 790-792.	11.1	1
94	Neutropenia-Associated Mutations in PRDM5, a Novel Epigenetic Regulator of Hematopoiesis.. <i>Blood</i> , 2006, 108, 503-503.	1.4	1
95	G-CSF receptor mutations mix it up. <i>Blood</i> , 2005, 105, 440-440.	1.4	0
96	Lymphadenopathy as the primary manifestation of malignant transformation in two patients with severe congenital neutropenia. <i>Pediatric Blood and Cancer</i> , 2008, 50, 1072-1075.	1.5	0
97	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.. <i>Blood</i> , 2004, 104, 781-781.	1.4	0
98	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.. <i>Blood</i> , 2006, 108, 1277-1277.	1.4	0
99	Towards Cell Therapy for FPD/AML with Patient-Specific IPS Cells. <i>Blood</i> , 2012, 120, 4677-4677.	1.4	0
100	Co-Opting the Genetic Redundancy of the Pax Family to Treat PAX5 Mutated Pre-B ALL. <i>Blood</i> , 2015, 126, 2445-2445.	1.4	0