

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

82
papers

3,370
citations

33
h-index

57
g-index

87
ext. papers

3,902
ext. citations

7.5
avg, IF

4.47
L-index

#	Paper	IF	Citations
82	Heritable GATA2 mutations associated with familial myelodysplastic syndrome and acute myeloid leukemia. <i>Nature Genetics</i> , 2011 , 43, 1012-7	36.3	424
81	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, 1283-91 ²	2.2	216
80	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. <i>Nature Genetics</i> , 2013 , 45, 1226-1231	36.3	205
79	Sphingosine kinase-1 enhances endothelial cell survival through a PECAM-1-dependent activation of PI-3K/Akt and regulation of Bcl-2 family members. <i>Blood</i> , 2005 , 105, 3169-77	2.2	147
78	GATA2 is required for lymphatic vessel valve development and maintenance. <i>Journal of Clinical Investigation</i> , 2015 , 125, 2979-94	15.9	136
77	Generalized CNS disease and massive GM1-ganglioside accumulation in mice defective in lysosomal acid beta-galactosidase. <i>Human Molecular Genetics</i> , 1997 , 6, 205-11	5.6	135
76	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. <i>Blood</i> , 2016 , 127, 1017-23	2.2	117
75	Transcriptional synergism between vitamin D-responsive elements in the rat 25-hydroxyvitamin D3 24-hydroxylase (CYP24) promoter. <i>Journal of Biological Chemistry</i> , 1996 , 271, 29715-21	5.4	102
74	PPARgamma agonists ameliorate endothelial cell activation via inhibition of diacylglycerol-protein kinase C signaling pathway: role of diacylglycerol kinase. <i>Circulation Research</i> , 2004 , 94, 1515-22	15.7	89
73	Basal and angiopoietin-1-mediated endothelial permeability is regulated by sphingosine kinase-1. <i>Blood</i> , 2008 , 111, 3489-97	2.2	78
72	Systemic and neurologic abnormalities distinguish the lysosomal disorders sialidosis and galactosialidosis in mice. <i>Human Molecular Genetics</i> , 2002 , 11, 1455-64	5.6	78
71	Ozz-E3, a muscle-specific ubiquitin ligase, regulates beta-catenin degradation during myogenesis. <i>Developmental Cell</i> , 2004 , 6, 269-82	10.2	76
70	ARMC5 mutations are common in familial bilateral macronodular adrenal hyperplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014 , 99, E1784-92	5.6	73
69	Novel RUNX1 mutations in familial platelet disorder with enhanced risk for acute myeloid leukemia: clues for improved identification of the FPD/AML syndrome. <i>Leukemia</i> , 2010 , 24, 242-6	10.7	73
68	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. <i>Molecular Psychiatry</i> , 2012 , 17, 1103-15	15.1	71
67	Phenoxodiol, an experimental anticancer drug, shows potent antiangiogenic properties in addition to its antitumour effects. <i>International Journal of Cancer</i> , 2006 , 118, 2412-20	7.5	67
66	Effect of disrupted SOX18 transcription factor function on tumor growth, vascularization, and endothelial development. <i>Journal of the National Cancer Institute</i> , 2006 , 98, 1060-7	9.7	65

65	Role of protein kinase Czeta in thrombin-induced endothelial permeability changes: inhibition by angiotensin-1. <i>Blood</i> , 2004 , 104, 1716-24	2.2	59
64	Genomic subtyping and therapeutic targeting of acute erythroleukemia. <i>Nature Genetics</i> , 2019 , 51, 694-704	36.3	54
63	Identification of a vitamin D responsive element in the promoter of the rat cytochrome P450(24) gene. <i>Nucleic Acids Research</i> , 1994 , 22, 2410-6	20.1	53
62	Regulation of vascular leak and recovery from ischemic injury by general and VE-cadherin-restricted miRNA antagonists of miR-27. <i>Blood</i> , 2013 , 122, 2911-9	2.2	48
61	Deep sequencing analysis of the developing mouse brain reveals a novel microRNA. <i>BMC Genomics</i> , 2011 , 12, 176	4.5	46
60	Spliceosome mutations in hematopoietic malignancies. <i>Nature Genetics</i> , 2011 , 44, 9-10	36.3	46
59	Myeloid neoplasms with germline DDX41 mutation. <i>International Journal of Hematology</i> , 2017 , 106, 163-174	1.34	44
58	Correction of murine galactosialidosis by bone marrow-derived macrophages overexpressing human protective protein/cathepsin A under control of the colony-stimulating factor-1 receptor promoter. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1998 , 95, 14880-5	11.5	42
57	Revealing Missing Human Protein Isoforms Based on Ab Initio Prediction, RNA-seq and Proteomics. <i>Scientific Reports</i> , 2015 , 5, 10940	4.9	38
56	RUNX1-mutated families show phenotype heterogeneity and a somatic mutation profile unique to germline predisposed AML. <i>Blood Advances</i> , 2020 , 4, 1131-1144	7.8	37
55	Stress-induced premature senescence mediated by a novel gene, SENEX, results in an anti-inflammatory phenotype in endothelial cells. <i>Blood</i> , 2010 , 116, 4016-24	2.2	36
54	A vascular cell-restricted RhoGAP, p73RhoGAP, is a key regulator of angiogenesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004 , 101, 12212-7	11.5	36
53	Secondary leukemia in patients with germline transcription factor mutations (RUNX1, GATA2, CEBPA). <i>Blood</i> , 2020 , 136, 24-35	2.2	35
52	JAM-C induces endothelial cell permeability through its association and regulation of β 3 integrins. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2009 , 29, 1200-6	9.4	35
51	Osteoblast gene expression in rat long bones: effects of ovariectomy and dihydrotestosterone on mRNA levels. <i>Calcified Tissue International</i> , 2000 , 67, 75-9	3.9	35
50	GATA2 monoallelic expression underlies reduced penetrance in inherited GATA2-mutated MDS/AML. <i>Leukemia</i> , 2018 , 32, 2502-2507	10.7	33
49	Differential effects on gene transcription and hematopoietic differentiation correlate with GATA2 mutant disease phenotypes. <i>Leukemia</i> , 2018 , 32, 194-202	10.7	32
48	Molecular networks involved in mouse cerebral corticogenesis and spatio-temporal regulation of Sox4 and Sox11 novel antisense transcripts revealed by transcriptome profiling. <i>Genome Biology</i> , 2009 , 10, R104	18.3	31

47	Splice factor mutations and alternative splicing as drivers of hematopoietic malignancy. <i>Immunological Reviews</i> , 2015 , 263, 257-78	11.3	29
46	Transcriptional regulation of the chicken CYP2H1 gene. Localization of a phenobarbital-responsive enhancer domain. <i>Journal of Biological Chemistry</i> , 1991 , 266, 17031-9	5.4	28
45	Ectrodactyly and Lethal Pulmonary Acinar Dysplasia Associated with Homozygous FGFR2 Mutations Identified by Exome Sequencing. <i>Human Mutation</i> , 2016 , 37, 955-63	4.7	28
44	A tale of two siblings: two cases of AML arising from a single pre-leukemic DNMT3A mutant clone. <i>Leukemia</i> , 2015 , 29, 2101-4	10.7	26
43	Spatiotemporal regulation of multiple overlapping sense and novel natural antisense transcripts at the Nrgn and Camk2n1 gene loci during mouse cerebral corticogenesis. <i>Cerebral Cortex</i> , 2011 , 21, 683-97	5.1	26
42	A four-gene LincRNA expression signature predicts risk in multiple cohorts of acute myeloid leukemia patients. <i>Leukemia</i> , 2018 , 32, 263-272	10.7	25
41	Chronic increases in sphingosine kinase-1 activity induce a pro-inflammatory, pro-angiogenic phenotype in endothelial cells. <i>Cellular and Molecular Biology Letters</i> , 2009 , 14, 424-41	8.1	23
40	Localization of the human vitamin D 24-hydroxylase gene (CYP24) to chromosome 20q13.2-->q13.3. <i>Cytogenetic and Genome Research</i> , 1993 , 62, 192-3	1.9	22
39	The mutational burden of therapy-related myeloid neoplasms is similar to primary myelodysplastic syndrome but has a distinctive distribution. <i>Leukemia</i> , 2019 , 33, 2842-2853	10.7	19
38	Apolipoprotein E enhances hepatic lipase-mediated hydrolysis of reconstituted high-density lipoprotein phospholipid and triacylglycerol in an isoform-dependent manner. <i>Biochemistry</i> , 2004 , 43, 12306-14	3.2	19
37	Lack of PPCA expression only partially coincides with lysosomal storage in galactosialidosis mice: indirect evidence for spatial requirement of the catalytic rather than the protective function of PPCA. <i>Human Molecular Genetics</i> , 1998 , 7, 1787-94	5.6	19
36	Superinduction by cycloheximide of cytochrome P4502H1 and 5-aminolevulinate synthase gene transcription in chick embryo liver. <i>Archives of Biochemistry and Biophysics</i> , 1993 , 300, 531-4	4.1	19
35	Self-reverting mutations partially correct the blood phenotype in a Diamond Blackfan anemia patient. <i>Haematologica</i> , 2017 , 102, e506-e509	6.6	17
34	Sphingosine kinase-1 associates with integrin $\alpha V\beta 3$ to mediate endothelial cell survival. <i>American Journal of Pathology</i> , 2009 , 175, 2217-25	5.8	17
33	Expression profiling reveals functionally important genes and coordinately regulated signaling pathway genes during in vitro angiogenesis. <i>Physiological Genomics</i> , 2005 , 22, 57-69	3.6	16
32	Derivation of an endogenous small RNA from double-stranded Sox4 sense and natural antisense transcripts in the mouse brain. <i>Genomics</i> , 2016 , 107, 88-99	4.3	14
31	Poor prognosis in familial acute myeloid leukaemia with combined biallelic CEBPA mutations and downstream events affecting the ATM, FLT3 and CDX2 genes. <i>British Journal of Haematology</i> , 2010 , 150, 382-5	4.5	13
30	A novel germline mutation in a family with ataxia-pancytopenia syndrome and pediatric acute lymphoblastic leukemia. <i>Haematologica</i> , 2019 , 104, e318-e321	6.6	11

29	A synonymous GATA2 variant underlying familial myeloid malignancy with striking intrafamilial phenotypic variability. <i>British Journal of Haematology</i> , 2020 , 190, e297-e301	4.5	10
28	Targeted gene panels identify a high frequency of pathogenic germline variants in patients diagnosed with a hematological malignancy and at least one other independent cancer. <i>Leukemia</i> , 2021 , 35, 3245-3256	10.7	10
27	Characterisation of a compound in-cis GATA2 germline mutation in a pedigree presenting with myelodysplastic syndrome/acute myeloid leukemia with concurrent thrombocytopenia. <i>Leukemia</i> , 2015 , 29, 1795-7	10.7	9
26	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	2.2	8
25	Allan-Herndon-Dudley syndrome with unusual profound sensorineural hearing loss. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 1872-6	2.5	7
24	A Method for Next-Generation Sequencing of Paired Diagnostic and Remission Samples to Detect Mitochondrial DNA Mutations Associated with Leukemia. <i>Journal of Molecular Diagnostics</i> , 2017 , 19, 711-721	5.1	7
23	Apparent TAK2-negative Tpolycythaemia vera due to compound mutations in exon 14. <i>British Journal of Haematology</i> , 2017 , 178, 333-336	4.5	7
22	Delayed diagnosis leading to accelerated-phase chronic eosinophilic leukemia due to a cytogenetically cryptic, imatinib-responsive TNIP1-PDFGRB fusion gene. <i>Leukemia</i> , 2016 , 30, 1402-5	10.7	6
21	T cell receptor assessment in autoimmune disease requires access to the most adjacent immunologically active organ. <i>Journal of Autoimmunity</i> , 2017 , 81, 24-33	15.5	5
20	Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in SLC5A6. <i>Npj Genomic Medicine</i> , 2019 , 4, 28	6.2	5
19	A novel, somatic, transforming mutation in the extracellular domain of Epidermal Growth Factor Receptor identified in myeloproliferative neoplasm. <i>Scientific Reports</i> , 2017 , 7, 2467	4.9	4
18	In depth analysis of the Sox4 gene locus that consists of sense and natural antisense transcripts. <i>Data in Brief</i> , 2016 , 7, 282-90	1.2	4
17	Genome-wide gene expression profiling identifies overlap with malignant adrenocortical tumours and novel mechanisms of inefficient steroidogenesis in familial ACTH-independent macronodular adrenal hyperplasia. <i>Endocrine-Related Cancer</i> , 2012 , 19, L19-23	5.7	4
16	GATA2 deficiency syndrome: A decade of discovery. <i>Human Mutation</i> , 2021 , 42, 1399-1421	4.7	4
15	ARMC5 is not implicated in familial hyperaldosteronism type II (FH-II). <i>Journal of Human Hypertension</i> , 2017 , 31, 857-859	2.6	3
14	High Incidence of Mutated Cancer-Associated Genes at Diagnosis in CML Patients with Early Transformation to Blast Crisis. <i>Blood</i> , 2015 , 126, 600-600	2.2	3
13	The RUNX1 database (RUNX1db): establishment of an expert curated RUNX1 registry and genomics database as a public resource for familial platelet disorder with myeloid malignancy. <i>Haematologica</i> , 2021 , 106, 3004-3007	6.6	3
12	Clinical implications of transient myeloproliferative disorder in a neonate without Down syndrome features. <i>Journal of Paediatrics and Child Health</i> , 2017 , 53, 1018-1020	1.3	2

11	Australian Familial Haematological Cancer Study - Findings from 15 Years of Aggregated Clinical, Genomic and Transcriptomic Data. <i>Blood</i> , 2019 , 134, 1439-1439	2.2	2
10	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	2.2	2
9	Rare and Common Germline Variants Contribute to Occurrence of Myelodysplastic Syndrome. <i>Blood</i> , 2015 , 126, 1644-1644	2.2	1
8	Two monogenic disorders masquerading as one: severe congenital neutropenia with monocytosis and non-syndromic sensorineural hearing loss. <i>BMC Medical Genetics</i> , 2020 , 21, 35	2.1	1
7	Childhood acute myeloid leukemia shows a high level of germline predisposition. <i>Blood</i> , 2021 , 138, 2293-2298	2.2	0
6	Pathogenic variants in cause recessive central conducting lymphatic anomaly with lymphedema.. <i>Science Translational Medicine</i> , 2022 , 14, eabm4869	17.5	0
5	Development of a Data Portal for Aggregation and Analysis of Genomics Data in Familial Platelet Disorder with Predisposition to Myeloid Malignancy - the RUNX1.DB. <i>Blood</i> , 2018 , 132, 5241-5241	2.2	
4	Familial Clustering of Hematological Malignancies: Harbingers of Wider Germline Cancer Susceptibility. <i>Blood</i> , 2019 , 134, 3794-3794	2.2	
3	Aberrant Activation of Epidermal Growth Factor Receptor in MPN May Respond to the Kinase Inhibitor Gefitinib. <i>Blood</i> , 2014 , 124, 1882-1882	2.2	
2	Clonal Diversity of Recurrently Mutated Genes in Myelodysplastic Syndromes. <i>Blood</i> , 2014 , 124, 4634-4634	2.2	
1	Mechanisms of Co-Operation of DNMT3A Mutations with JAK2 V617F Through Histone H4 Arginine 3 Provides New Insights in MPN Disease Pathogenesis. <i>Blood</i> , 2011 , 118, 2823-2823	2.2	