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List of Publications by Year in descending order

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Version: 2024-02-01

118
papers

24,378
citations

17440

63
h-index

19190

118
g-index

133
all docs

133
docs citations

133
times ranked

40181
citing authors

#	ARTICLE	IF	CITATIONS
1	Genomewide Association Analysis of Coronary Artery Disease. <i>New England Journal of Medicine</i> , 2007, 357, 443-453.	27.0	1,865
2	Defining the role of common variation in the genomic and biological architecture of adult human height. <i>Nature Genetics</i> , 2014, 46, 1173-1186.	21.4	1,818
3	Large-scale association analysis identifies new risk loci for coronary artery disease. <i>Nature Genetics</i> , 2013, 45, 25-33.	21.4	1,439
4	Genomic atlas of the human plasma proteome. <i>Nature</i> , 2018, 558, 73-79.	27.8	1,180
5	The Allelic Landscape of Human Blood Cell Trait Variation and Links to Common Complex Disease. <i>Cell</i> , 2016, 167, 1415-1429.e19.	28.9	1,052
6	Lineage-Specific Genome Architecture Links Enhancers and Non-coding Disease Variants to Target Gene Promoters. <i>Cell</i> , 2016, 167, 1369-1384.e19.	28.9	863
7	The Human Phenotype Ontology in 2017. <i>Nucleic Acids Research</i> , 2017, 45, D865-D876.	14.5	699
8	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. <i>Nucleic Acids Research</i> , 2014, 42, D966-D974.	14.5	698
9	DNA methylation and body-mass index: a genome-wide analysis. <i>Lancet</i> , The, 2014, 383, 1990-1998.	13.7	686
10	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. <i>Nature Genetics</i> , 2021, 53, 1300-1310.	21.4	590
11	Genetic Drivers of Epigenetic and Transcriptional Variation in Human Immune Cells. <i>Cell</i> , 2016, 167, 1398-1414.e24.	28.9	573
12	A catalog of genetic loci associated with kidney function from analyses of a million individuals. <i>Nature Genetics</i> , 2019, 51, 957-972.	21.4	549
13	Common genetic variation drives molecular heterogeneity in human iPSCs. <i>Nature</i> , 2017, 546, 370-375.	27.8	491
14	Bayesian refinement of association signals for 14 loci in 3 common diseases. <i>Nature Genetics</i> , 2012, 44, 1294-1301.	21.4	469
15	Leukemia-Associated Somatic Mutations Drive Distinct Patterns of Age-Related Clonal Hemopoiesis. <i>Cell Reports</i> , 2015, 10, 1239-1245.	6.4	443
16	New gene functions in megakaryopoiesis and platelet formation. <i>Nature</i> , 2011, 480, 201-208.	27.8	401
17	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. <i>Nature Neuroscience</i> , 2016, 19, 571-577.	14.8	388
18	The Polygenic and Monogenic Basis of Blood Traits and Diseases. <i>Cell</i> , 2020, 182, 1214-1231.e11.	28.9	388

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19	Compound inheritance of a low-frequency regulatory SNP and a rare null mutation in exon-junction complex subunit RBM8A causes TAR syndrome. <i>Nature Genetics</i> , 2012, 44, 435-439.	21.4	355
20	Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations. <i>Cell</i> , 2020, 182, 1198-1213.e14.	28.9	353
21	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 75-90.	6.2	343
22	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020, 583, 96-102.	27.8	338
23	Detection of Atherosclerotic Inflammation by ⁶⁸ Ga-DOTATATE PET Compared to [¹⁸ F]FDG PET Imaging. <i>Journal of the American College of Cardiology</i> , 2017, 69, 1774-1791.	2.8	321
24	Seventy-five genetic loci influencing the human red blood cell. <i>Nature</i> , 2012, 492, 369-375.	27.8	320
25	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. <i>Nature Communications</i> , 2018, 9, 1416.	12.8	279
26	Genome-wide Analysis of Simultaneous GATA1/2, RUNX1, FLI1, and SCL Binding in Megakaryocytes Identifies Hematopoietic Regulators. <i>Developmental Cell</i> , 2011, 20, 597-609.	7.0	255
27	Transcriptional diversity during lineage commitment of human blood progenitors. <i>Science</i> , 2014, 345, 1251033.	12.6	253
28	Cell-specific protein phenotypes for the autoimmune locus IL2RA using a genotype-selectable human bioresource. <i>Nature Genetics</i> , 2009, 41, 1011-1015.	21.4	249
29	Exome sequencing identifies NBEAL2 as the causative gene for gray platelet syndrome. <i>Nature Genetics</i> , 2011, 43, 735-737.	21.4	245
30	Large-Scale Gene-Centric Meta-analysis across 32 Studies Identifies Multiple Lipid Loci. <i>American Journal of Human Genetics</i> , 2012, 91, 823-838.	6.2	227
31	Efficiency and safety of varying the frequency of whole blood donation (INTERVAL): a randomised trial of 45 000 donors. <i>Lancet, The</i> , 2017, 390, 2360-2371.	13.7	222
32	DNA Methylation Dynamics of Human Hematopoietic Stem Cell Differentiation. <i>Cell Stem Cell</i> , 2016, 19, 808-822.	11.1	216
33	Large-scale production of megakaryocytes from human pluripotent stem cells by chemically defined forward programming. <i>Nature Communications</i> , 2016, 7, 11208.	12.8	199
34	Novel loci affecting iron homeostasis and their effects in individuals at risk for hemochromatosis. <i>Nature Communications</i> , 2014, 5, 4926.	12.8	192
35	Identification of novel risk loci for restless legs syndrome in genome-wide association studies in individuals of European ancestry: a meta-analysis. <i>Lancet Neurology, The</i> , 2017, 16, 898-907.	10.2	191
36	Loss-of-function nuclear factor κ B subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 1285-1296.	2.9	185

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37	Human Antibody Fragments Specific for Human Blood Group Antigens from a Phage Display Library. <i>Nature Biotechnology</i> , 1993, 11, 1145-1149.	17.5	172
38	Activation during preparation of therapeutic platelets affects deterioration during storage: a comparative flow cytometric study of different production methods. <i>British Journal of Haematology</i> , 1997, 98, 86-95.	2.5	157
39	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2016, 127, 2791-2803.	1.4	157
40	Comparative gene expression profiling of in vitro differentiated megakaryocytes and erythroblasts identifies novel activatory and inhibitory platelet membrane proteins. <i>Blood</i> , 2007, 109, 3260-3269.	1.4	153
41	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , 2020, 583, 90-95.	27.8	148
42	Platelet genomics and proteomics in human health and disease. <i>Journal of Clinical Investigation</i> , 2005, 115, 3370-3377.	8.2	146
43	Polygenic basis and biomedical consequences of telomere length variation. <i>Nature Genetics</i> , 2021, 53, 1425-1433.	21.4	145
44	Increased DNA methylation variability in type 1 diabetes across three immune effector cell types. <i>Nature Communications</i> , 2016, 7, 13555.	12.8	142
45	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2019, 134, 2082-2091.	1.4	131
46	The low-frequency allele of the platelet collagen signaling receptor glycoprotein VI is associated with reduced functional responses and expression. <i>Blood</i> , 2003, 101, 4372-4379.	1.4	124
47	A gain-of-function variant in <i>DIAPH1</i> causes dominant macrothrombocytopenia and hearing loss. <i>Blood</i> , 2016, 127, 2903-2914.	1.4	121
48	Human phenotype ontology annotation and cluster analysis to unravel genetic defects in 707 cases with unexplained bleeding and platelet disorders. <i>Genome Medicine</i> , 2015, 7, 36.	8.2	119
49	Inherited platelet disorders: toward DNA-based diagnosis. <i>Blood</i> , 2016, 127, 2814-2823.	1.4	119
50	A novel variant on chromosome 7q22.3 associated with mean platelet volume, counts, and function. <i>Blood</i> , 2009, 113, 3831-3837.	1.4	117
51	The INTERVAL trial to determine whether intervals between blood donations can be safely and acceptably decreased to optimise blood supply: study protocol for a randomised controlled trial. <i>Trials</i> , 2014, 15, 363.	1.6	112
52	Phenotypic Characterization of <i>EIF2AK4</i> Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. <i>Circulation</i> , 2017, 136, 2022-2033.	1.6	111
53	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. <i>Genome Medicine</i> , 2018, 10, 95.	8.2	111
54	Dynamics of Transcription Regulation in Human Bone Marrow Myeloid Differentiation to Mature Blood Neutrophils. <i>Cell Reports</i> , 2018, 24, 2784-2794.	6.4	104

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55	Treatment of COVID-19 with remdesivir in the absence of humoral immunity: a case report. <i>Nature Communications</i> , 2020, 11, 6385.	12.8	103
56	eFORGE: A Tool for Identifying Cell Type-Specific Signal in Epigenomic Data. <i>Cell Reports</i> , 2016, 17, 2137-2150.	6.4	102
57	A nonsynonymous SNP in the ITGB3 gene disrupts the conserved membrane-proximal cytoplasmic salt bridge in the α IIb β 3 integrin and cosegregates dominantly with abnormal proplatelet formation and macrothrombocytopenia. <i>Blood</i> , 2008, 111, 3407-3414.	1.4	94
58	Transcription profiling in human platelets reveals LRRFIP1 as a novel protein regulating platelet function. <i>Blood</i> , 2010, 116, 4646-4656.	1.4	90
59	A dominant gain-of-function mutation in universal tyrosine kinase <i>SRC</i> causes thrombocytopenia, myelofibrosis, bleeding, and bone pathologies. <i>Science Translational Medicine</i> , 2016, 8, 328ra30.	12.4	87
60	Gray platelet syndrome: proinflammatory megakaryocytes and α -granule loss cause myelofibrosis and confer metastasis resistance in mice. <i>Blood</i> , 2014, 124, 3624-3635.	1.4	79
61	A genome-wide meta-analysis yields 46 new loci associating with biomarkers of iron homeostasis. <i>Communications Biology</i> , 2021, 4, 156.	4.4	72
62	Germline variants in <i>ETV6</i> underlie reduced platelet formation, platelet dysfunction and increased levels of circulating CD34 ⁺ progenitors. <i>Haematologica</i> , 2017, 102, 282-294.	3.5	70
63	Automated typing of red blood cell and platelet antigens: a whole-genome sequencing study. <i>Lancet Haematology</i> , 2018, 5, e241-e251.	4.6	70
64	Functional genomics in zebrafish permits rapid characterization of novel platelet membrane proteins. <i>Blood</i> , 2009, 113, 4754-4762.	1.4	69
65	Chromosome contacts in activated T cells identify autoimmune disease candidate genes. <i>Genome Biology</i> , 2017, 18, 165.	8.8	68
66	Microarray-based genotyping for blood groups: comparison of gene array and 5 α -nuclease assay techniques with human platelet antigen as a model. <i>Transfusion</i> , 2005, 45, 654-659.	1.6	64
67	Mutations in tropomyosin 4 underlie a rare form of human macrothrombocytopenia. <i>Journal of Clinical Investigation</i> , 2017, 127, 814-829.	8.2	57
68	Curated disease-causing genes for bleeding, thrombotic, and platelet disorders: Communication from the SSC of the ISTH. <i>Journal of Thrombosis and Haemostasis</i> , 2019, 17, 1253-1260.	3.8	56
69	Distinct Trends of DNA Methylation Patterning in the Innate and Adaptive Immune Systems. <i>Cell Reports</i> , 2016, 17, 2101-2111.	6.4	54
70	Platelet function is modified by common sequence variation in megakaryocyte super enhancers. <i>Nature Communications</i> , 2017, 8, 16058.	12.8	50
71	GNE variants causing autosomal recessive macrothrombocytopenia without associated muscle wasting. <i>Blood</i> , 2018, 132, 1851-1854.	1.4	48
72	Characterization of human variable domain antibody fragments against the U1 RNA-associated A protein, selected from a synthetic and a patient-derived combinatorial V gene library. <i>European Journal of Immunology</i> , 1996, 26, 629-639.	2.9	46

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73	Introduction to Antibody Engineering and Phage Display. <i>Vox Sanguinis</i> , 2000, 78, 72-79.	1.5	45
74	Integrative analysis of the plasma proteome and polygenic risk of cardiometabolic diseases. <i>Nature Metabolism</i> , 2021, 3, 1476-1483.	11.9	43
75	Rare variants in GP1BB are responsible for autosomal dominant macrothrombocytopenia. <i>Blood</i> , 2017, 129, 520-524.	1.4	42
76	Large genome-wide association study identifies three novel risk variants for restless legs syndrome. <i>Communications Biology</i> , 2020, 3, 703.	4.4	40
77	Mutational and phenotypic characterization of hereditary hemorrhagic telangiectasia. <i>Blood</i> , 2020, 136, 1907-1918.	1.4	40
78	Mitochondrial DNA variants modulate N-formylmethionine, proteostasis and risk of late-onset human diseases. <i>Nature Medicine</i> , 2021, 27, 1564-1575.	30.7	40
79	Expanded repertoire of RASGRP2 variants responsible for platelet dysfunction and severe bleeding. <i>Blood</i> , 2017, 130, 1026-1030.	1.4	38
80	Congenital macrothrombocytopenia with focal myelofibrosis due to mutations in human G6b-B is rescued in humanized mice. <i>Blood</i> , 2018, 132, 1399-1412.	1.4	37
81	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018, 103, 144-153.	6.2	36
82	Threonine-145/Methionine-145 variants of baculovirus produced recombinant ligand binding domain of GPIb \pm express HPA-2 epitopes and show equal binding of von Willebrand factor. <i>Blood</i> , 2000, 95, 205-211.	1.4	34
83	Silencing of RhoA nucleotide exchange factor, ARHGEF3, reveals its unexpected role in iron uptake. <i>Blood</i> , 2011, 118, 4967-4976.	1.4	34
84	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. <i>Blood</i> , 2020, 136, 1956-1967.	1.4	34
85	Phenotype description and response to thrombopoietin receptor agonist in DIAPH1-related disorder. <i>Blood Advances</i> , 2018, 2, 2341-2346.	5.2	33
86	Development and validation of a universal blood donor genotyping platform: a multinational prospective study. <i>Blood Advances</i> , 2020, 4, 3495-3506.	5.2	31
87	Effects of adiposity on the human plasma proteome: observational and Mendelian randomisation estimates. <i>International Journal of Obesity</i> , 2021, 45, 2221-2229.	3.4	31
88	Nbeal2 interacts with Dock7, Sec16a, and Vac14. <i>Blood</i> , 2018, 131, 1000-1011.	1.4	29
89	Germline mutations in the transcription factor IKZF5 cause thrombocytopenia. <i>Blood</i> , 2019, 134, 2070-2081.	1.4	29
90	Sphingolipid dysregulation due to lack of functional KDSR impairs proplatelet formation causing thrombocytopenia. <i>Haematologica</i> , 2019, 104, 1036-1045.	3.5	28

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91	How common are single gene mutations as a cause for lacunar stroke?. <i>Neurology</i> , 2019, 93, e2007-e2020.	1.1	26
92	NBEAL2 is required for neutrophil and NK cell function and pathogen defense. <i>Journal of Clinical Investigation</i> , 2017, 127, 3521-3526.	8.2	25
93	Introduction to Antibody Engineering and Phage Display. <i>Vox Sanguinis</i> , 2000, 78, 72-79.	1.5	24
94	Dawning of the age of genomics for platelet granule disorders: improving insight, diagnosis and management. <i>British Journal of Haematology</i> , 2017, 176, 705-720.	2.5	22
95	Neutrophil specific granule and NETosis defects in gray platelet syndrome. <i>Blood Advances</i> , 2021, 5, 549-564.	5.2	18
96	Elevated levels of tissue factor pathway inhibitor in patients with mild to moderate bleeding tendency. <i>Blood Advances</i> , 2021, 5, 391-398.	5.2	18
97	Recruitment and representativeness of blood donors in the INTERVAL randomised trial assessing varying inter-donation intervals. <i>Trials</i> , 2016, 17, 458.	1.6	17
98	Longer-term efficiency and safety of increasing the frequency of whole blood donation (INTERVAL): extension study of a randomised trial of 20â€757 blood donors. <i>Lancet Haematology</i> , 2019, 6, e510-e520.	4.6	17
99	A coagulation defect arising from heterozygous premature termination of tissue factor. <i>Journal of Clinical Investigation</i> , 2020, 130, 5302-5312.	8.2	17
100	Whole-exome sequencing identifies rare genetic variants associated with human plasma metabolites. <i>American Journal of Human Genetics</i> , 2022, 109, 1038-1054.	6.2	17
101	Paired rRNA-depleted and polyA-selected RNA sequencing data and supporting multi-omics data from human T cells. <i>Scientific Data</i> , 2020, 7, 376.	5.3	15
102	Transcriptional characterization of human megakaryocyte polyploidization and lineage commitment. <i>Journal of Thrombosis and Haemostasis</i> , 2021, 19, 1236-1249.	3.8	15
103	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. <i>Molecular Psychiatry</i> , 2021, 26, 5239-5250.	7.9	15
104	Comparison of four methods to measure haemoglobin concentrations in whole blood donors (<sc>COMPARE</sc>): A diagnostic accuracy study. <i>Transfusion Medicine</i> , 2021, 31, 94-103.	1.1	13
105	Megakaryocytes in Myeloproliferative Neoplasms Have Unique Somatic Mutations. <i>American Journal of Pathology</i> , 2017, 187, 1512-1522.	3.8	12
106	Cell type-specific novel long non-coding RNA and circular RNA in the BLUEPRINT hematopoietic transcriptomes atlas. <i>Haematologica</i> , 2021, 106, 2613-2623.	3.5	12
107	The Natural History of Fetomaternal Alloimmunization to the Platelet-Specific Antigen HPA-1a (PIA1), Tj ETQq1 1 0.784314 rgBT /Overdo	1.4	10
108	Identification of variation in the platelet transcriptome associated with Glycoprotein 6 haplotype. <i>Platelets</i> , 2008, 19, 258-267.	2.3	9

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109	The influence of rare variants in circulating metabolic biomarkers. PLoS Genetics, 2020, 16, e1008605.	3.5	9
110	Machine learning optimized polygenic scores for blood cell traits identify sex-specific trajectories and genetic correlations with disease. Cell Genomics, 2022, 2, 100086.	6.5	9
111	Long-Read Sequencing Identifies the First Retrotransposon Insertion and Resolves Structural Variants Causing Antithrombin Deficiency. Thrombosis and Haemostasis, 2022, 122, 1369-1378.	3.4	9
112	G protein-coupled receptor kinase 5 regulates thrombin signaling in platelets via PAR-1. Blood Advances, 2022, 6, 2319-2330.	5.2	8
113	Image-based characterization of thrombus formation in time-lapse DIC microscopy. Medical Image Analysis, 2012, 16, 915-931.	11.6	6
114	Single Nucleotide Variants in the Protein C Pathway and Mortality in Dialysis Patients. PLoS ONE, 2014, 9, e97251.	2.5	6
115	Platelet Genomics. , 2013, , 67-89.		2
116	Common genetic variants do not associate with CAD in familial hypercholesterolemia. European Journal of Human Genetics, 2014, 22, 809-813.	2.8	2
117	Joint Thrombus and Vessel Segmentation Using Dynamic Texture Likelihoods and Shape Prior. Lecture Notes in Computer Science, 2011, 14, 579-586.	1.3	2
118	Automatic Event Detection within Thrombus Formation Based on Integer Programming. Lecture Notes in Computer Science, 2013, , 215-224.	1.3	0