

Julian Knight

List of Publications by Year in descending order

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Version: 2024-02-01

202
papers

34,276
citations

8732

75
h-index

4978

167
g-index

241
all docs

241
docs citations

241
times ranked

58514
citing authors

#	ARTICLE	IF	CITATIONS
1	Blood leukocyte transcriptomes in Gram-positive and Gram-negative community-acquired pneumonia. <i>European Respiratory Journal</i> , 2022, 59, 2101856.	3.1	3
2	Priority index: database of genetic targets in immune-mediated disease. <i>Nucleic Acids Research</i> , 2022, 50, D1358-D1367.	6.5	16
3	Serum calprotectin is not an independent predictor of severe COVID-19 in ambulatory adult patients. <i>Journal of Infection</i> , 2022, 84, e27-e29.	1.7	7
4	An immunodominant NP105â€“113-B*07:02 cytotoxic T cell response controls viral replication and is associated with less severe COVID-19 disease. <i>Nature Immunology</i> , 2022, 23, 50-61.	7.0	110
5	Response to Letter to the Editor by Ish et al. entitled â€“COVID-19 vaccine equityâ€”the need of the hourâ€™. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2022, , .	0.2	0
6	Human Leukocyte Antigen Association Study Reveals DRB1*04:02 Effects Additional to DRB1*07:01 in Anti-LGI1 Encephalitis. <i>Neurology: Neuroimmunology and Neuroinflammation</i> , 2022, 9, .	3.1	13
7	A blood atlas of COVID-19 defines hallmarks of disease severity and specificity. <i>Cell</i> , 2022, 185, 916-938.e58.	13.5	164
8	Fine-Scale Genetic Structure in the United Arab Emirates Reflects Endogamous and Consanguineous Culture, Population History, and Geography. <i>Molecular Biology and Evolution</i> , 2022, 39, .	3.5	3
9	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. <i>Human Genetics</i> , 2022, 141, 147-173.	1.8	22
10	Implementation and Extended Evaluation of the Euroimmun Anti-SARS-CoV-2 IgG Assay and Its Contribution to the United Kingdomâ€™s COVID-19 Public Health Response. <i>Microbiology Spectrum</i> , 2022, 10, e0228921.	1.2	2
11	SARS-CoV-2-specific antibody and T-cell responses 1 year after infection in people recovered from COVID-19: a longitudinal cohort study. <i>Lancet Microbe, The</i> , 2022, 3, e348-e356.	3.4	107
12	Whole-genome sequencing reveals host factors underlying critical COVID-19. <i>Nature</i> , 2022, 607, 97-103.	13.7	174
13	Divergent trajectories of antiviral memory after SARS-CoV-2 infection. <i>Nature Communications</i> , 2022, 13, 1251.	5.8	20
14	Identification of hostâ€“pathogen-disease relationships using a scalable multiplex serology platform in UK Biobank. <i>Nature Communications</i> , 2022, 13, 1818.	5.8	28
15	Why do breakthrough COVID-19 infections occur in the vaccinated?. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2022, 115, 67-68.	0.2	2
16	Redefining critical illness. <i>Nature Medicine</i> , 2022, 28, 1141-1148.	15.2	136
17	Epigenomic analysis reveals a dynamic and context-specific macrophage enhancer landscape associated with innate immune activation and tolerance. <i>Genome Biology</i> , 2022, 23, .	3.8	9
18	Natural Killer cells demonstrate distinct eQTL and transcriptome-wide disease associations, highlighting their role in autoimmunity. <i>Nature Communications</i> , 2022, 13, .	5.8	10

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19	The suboptimal fibrinolytic response in COVID-19 is dictated by high PAI-1. <i>Journal of Thrombosis and Haemostasis</i> , 2022, 20, 2394-2406.	1.9	30
20	Host genetics and infectious disease: new tools, insights and translational opportunities. <i>Nature Reviews Genetics</i> , 2021, 22, 137-153.	7.7	98
21	Safety and efficacy of the ChAdOx1 nCoV-19 vaccine (AZD1222) against SARS-CoV-2: an interim analysis of four randomised controlled trials in Brazil, South Africa, and the UK. <i>Lancet, The</i> , 2021, 397, 99-111.	6.3	3,887
22	Phase 1/2 trial of SARS-CoV-2 vaccine ChAdOx1 nCoV-19 with a booster dose induces multifunctional antibody responses. <i>Nature Medicine</i> , 2021, 27, 279-288.	15.2	265
23	Early childhood epilepsies: epidemiology, classification, aetiology, and socio-economic determinants. <i>Brain</i> , 2021, 144, 2879-2891.	3.7	64
24	Sepsis Subclasses: A Framework for Development and Interpretation*. <i>Critical Care Medicine</i> , 2021, 49, 748-759.	0.4	81
25	Genome-wide CRISPR/Cas9-knockout in human induced Pluripotent Stem Cell (iPSC)-derived macrophages. <i>Scientific Reports</i> , 2021, 11, 4245.	1.6	25
26	Single-dose administration and the influence of the timing of the booster dose on immunogenicity and efficacy of ChAdOx1 nCoV-19 (AZD1222) vaccine: a pooled analysis of four randomised trials. <i>Lancet, The</i> , 2021, 397, 881-891.	6.3	979
27	A haemagglutination test for rapid detection of antibodies to SARS-CoV-2. <i>Nature Communications</i> , 2021, 12, 1951.	5.8	54
28	Transcriptomic Analysis of Inflammatory Cardiomyopathy Identifies Molecular Signatures of Disease and Informs in silico Prediction of a Network-Based Rationale for Therapy. <i>Frontiers in Immunology</i> , 2021, 12, 640837.	2.2	3
29	The antigenic anatomy of SARS-CoV-2 receptor binding domain. <i>Cell</i> , 2021, 184, 2183-2200.e22.	13.5	331
30	Evidence of escape of SARS-CoV-2 variant B.1.351 from natural and vaccine-induced sera. <i>Cell</i> , 2021, 184, 2348-2361.e6.	13.5	936
31	Reduced neutralization of SARS-CoV-2 B.1.1.7 variant by convalescent and vaccine sera. <i>Cell</i> , 2021, 184, 2201-2211.e7.	13.5	442
32	Functional Genomic Analysis of a RUNX3 Polymorphism Associated With Ankylosing Spondylitis. <i>Arthritis and Rheumatology</i> , 2021, 73, 980-990.	2.9	10
33	Decreased ATM Function Causes Delayed DNA Repair and Apoptosis in Common Variable Immunodeficiency Disorders. <i>Journal of Clinical Immunology</i> , 2021, 41, 1315-1330.	2.0	6
34	Antibody evasion by the P.1 strain of SARS-CoV-2. <i>Cell</i> , 2021, 184, 2939-2954.e9.	13.5	519
35	Pre-existing asthma as a comorbidity does not modify cytokine responses and severity of COVID-19. <i>Allergy, Asthma and Clinical Immunology</i> , 2021, 17, 67.	0.9	3
36	Ex vivo mass cytometry analysis reveals a profound myeloid proinflammatory signature in psoriatic arthritis synovial fluid. <i>Annals of the Rheumatic Diseases</i> , 2021, 80, 1559-1567.	0.5	16

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37	Using de novo assembly to identify structural variation of eight complex immune system gene regions. <i>PLoS Computational Biology</i> , 2021, 17, e1009254.	1.5	22
38	Reduced neutralization of SARS-CoV-2 B.1.617 by vaccine and convalescent serum. <i>Cell</i> , 2021, 184, 4220-4236.e13.	13.5	630
39	Two doses of SARS-CoV-2 vaccination induce robust immune responses to emerging SARS-CoV-2 variants of concern. <i>Nature Communications</i> , 2021, 12, 5061.	5.8	150
40	Genomic Insights into Myasthenia Gravis Identify Distinct Immunological Mechanisms in Early and Late Onset Disease. <i>Annals of Neurology</i> , 2021, 90, 455-463.	2.8	8
41	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.	9.4	590
42	Genetic mechanisms of critical illness in COVID-19. <i>Nature</i> , 2021, 591, 92-98.	13.7	1,014
43	The impact of viral mutations on recognition by SARS-CoV-2 specific T _H cells. <i>iScience</i> , 2021, 24, 103353.	1.9	57
44	Identification of LZTFL1 as a candidate effector gene at a COVID-19 risk locus. <i>Nature Genetics</i> , 2021, 53, 1606-1615.	9.4	93
45	Maternal immune activation downregulates schizophrenia genes in the foetal mouse brain. <i>Brain Communications</i> , 2021, 3, fcab275.	1.5	10
46	Musical Activities, Prosocial Behaviors, and Executive Function Skills of Kindergarten Children. <i>Music & Science</i> , 2021, 4, 205920432110548.	0.6	4
47	Disruption of c-MYC Binding and Chromosomal Looping Involving Genetic Variants Associated With Ankylosing Spondylitis Upstream of the RUNX3 Promoter. <i>Frontiers in Genetics</i> , 2021, 12, 741867.	1.1	3
48	Elevated risk of invasive group A streptococcal disease and host genetic variation in the human leucocyte antigen locus. <i>Genes and Immunity</i> , 2020, 21, 63-70.	2.2	5
49	From genome-wide association studies to rational drug target prioritisation in inflammatory arthritis. <i>Lancet Rheumatology</i> , The, 2020, 2, e50-e62.	2.2	17
50	The Polygenic and Monogenic Basis of Blood Traits and Diseases. <i>Cell</i> , 2020, 182, 1214-1231.e11.	13.5	388
51	Early-onset autoimmunity associated with SOCS1 haploinsufficiency. <i>Nature Communications</i> , 2020, 11, 5341.	5.8	74
52	Safety and immunogenicity of the ChAdOx1 nCoV-19 vaccine against SARS-CoV-2: a preliminary report of a phase 1/2, single-blind, randomised controlled trial. <i>Lancet</i> , The, 2020, 396, 467-478.	6.3	2,080
53	Safety and immunogenicity of ChAdOx1 nCoV-19 vaccine administered in a prime-boost regimen in young and old adults (COV002): a single-blind, randomised, controlled, phase 2/3 trial. <i>Lancet</i> , The, 2020, 396, 1979-1993.	6.3	1,196
54	Identifying collagen VI as a target of fibrotic diseases regulated by CREBBP/EP300. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 20753-20763.	3.3	45

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55	Broad and strong memory CD4+ and CD8+ T cells induced by SARS-CoV-2 in UK convalescent individuals following COVID-19. <i>Nature Immunology</i> , 2020, 21, 1336-1345.	7.0	1,066
56	Performance characteristics of five immunoassays for SARS-CoV-2: a head-to-head benchmark comparison. <i>Lancet Infectious Diseases</i> , The, 2020, 20, 1390-1400.	4.6	336
57	Epstein-Barr virus reactivation in sepsis due to community-acquired pneumonia is associated with increased morbidity and an immunosuppressed host transcriptomic endotype. <i>Scientific Reports</i> , 2020, 10, 9838.	1.6	13
58	Peripheral CD8+ T cell characteristics associated with durable responses to immune checkpoint blockade in patients with metastatic melanoma. <i>Nature Medicine</i> , 2020, 26, 193-199.	15.2	211
59	Interferon-Induced Transmembrane Protein 3 Genetic Variant rs12252-C Associated With Disease Severity in Coronavirus Disease 2019. <i>Journal of Infectious Diseases</i> , 2020, 222, 34-37.	1.9	140
60	Multi-level evidence of an allelic hierarchy of USH2A variants in hearing, auditory processing and speech/language outcomes. <i>Communications Biology</i> , 2020, 3, 180.	2.0	6
61	Longitudinal COVID-19 profiling associates IL-1RA and IL-10 with disease severity and RANTES with mild disease. <i>JCI Insight</i> , 2020, 5, .	2.3	310
62	Antibody testing for COVID-19: A report from the National COVID Scientific Advisory Panel. <i>Wellcome Open Research</i> , 2020, 5, 139.	0.9	179
63	SARS-CoV-2 RNA detected in blood products from patients with COVID-19 is not associated with infectious virus. <i>Wellcome Open Research</i> , 2020, 5, 181.	0.9	81
64	SARS-CoV-2 RNA detected in blood products from patients with COVID-19 is not associated with infectious virus. <i>Wellcome Open Research</i> , 2020, 5, 181.	0.9	122
65	Defective tubulin detyrosination causes structural brain abnormalities with cognitive deficiency in humans and mice. <i>Human Molecular Genetics</i> , 2019, 28, 3391-3405.	1.4	43
66	Implementation of a genomic medicine multi-disciplinary team approach for rare disease in the clinical setting: a prospective exome sequencing case series. <i>Genome Medicine</i> , 2019, 11, 46.	3.6	25
67	A genetics-led approach defines the drug target landscape of 30 immune-related traits. <i>Nature Genetics</i> , 2019, 51, 1082-1091.	9.4	157
68	Context-specific regulation of surface and soluble IL7R expression by an autoimmune risk allele. <i>Nature Communications</i> , 2019, 10, 4575.	5.8	37
69	ANKK1 Ankylosing spondylitis associated polymorphism of the IL7R controls expression surface and soluble IL7R-alpha during inflammation. <i>Rheumatology</i> , 2019, 58, .	0.9	0
70	The association between endometriosis and autoimmune diseases: a systematic review and meta-analysis. <i>Human Reproduction Update</i> , 2019, 25, 486-503.	5.2	179
71	Novel ways to identify new treatment targets for inflammatory arthritis. <i>Rheumatology</i> , 2019, 58, .	0.9	0
72	Joint sequencing of human and pathogen genomes reveals the genetics of pneumococcal meningitis. <i>Nature Communications</i> , 2019, 10, 2176.	5.8	83

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73	Use of gene expression studies to investigate the human immunological response to malaria infection. <i>Malaria Journal</i> , 2019, 18, 418.	0.8	11
74	Transcriptomic Signatures in Sepsis and a Differential Response to Steroids. From the VANISH Randomized Trial. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2019, 199, 980-986.	2.5	178
75	Assessment of an Antibody-in-Lymphocyte Supernatant Assay for the Etiological Diagnosis of Pneumococcal Pneumonia in Children. <i>Frontiers in Cellular and Infection Microbiology</i> , 2019, 9, 459.	1.8	3
76	AltHapAlignR: improved accuracy of RNA-seq analyses through the use of alternative haplotypes. <i>Bioinformatics</i> , 2018, 34, 2401-2408.	1.8	27
77	Genomic Response to Vitamin D Supplementation in the Setting of a Randomized, Placebo-Controlled Trial. <i>EBioMedicine</i> , 2018, 31, 133-142.	2.7	29
78	A community approach to mortality prediction in sepsis via gene expression analysis. <i>Nature Communications</i> , 2018, 9, 694.	5.8	178
79	NOX1 loss-of-function genetic variants in patients with inflammatory bowel disease. <i>Mucosal Immunology</i> , 2018, 11, 562-574.	2.7	71
80	Evidence for a second ankylosing spondylitis-associated <i>RUNX3</i> regulatory polymorphism. <i>RMD Open</i> , 2018, 4, e000628.	1.8	16
81	Risk of nontyphoidal <i>Salmonella</i> bacteraemia in African children is modified by STAT4. <i>Nature Communications</i> , 2018, 9, 1014.	5.8	29
82	“Not pathogenic until proven otherwise”: perspectives of UK clinical genomics professionals toward secondary findings in context of a Genomic Medicine Multidisciplinary Team and the 100,000 Genomes Project. <i>Genetics in Medicine</i> , 2018, 20, 320-328.	1.1	56
83	Microvesicle Subsets in Sepsis Due to Community Acquired Pneumonia Compared to Faecal Peritonitis. <i>Shock</i> , 2018, 49, 393-401.	1.0	25
84	Investigating the Impact of a Musical Intervention on Preschool Children’s Executive Function. <i>Frontiers in Psychology</i> , 2018, 9, 2389.	1.1	25
85	Distinct HLA associations of LGI1 and CASPR2-antibody diseases. <i>Brain</i> , 2018, 141, 2263-2271.	3.7	100
86	Translating GWAS in rheumatic disease: approaches to establishing mechanism and function for genetic associations with ankylosing spondylitis. <i>Briefings in Functional Genomics</i> , 2018, 17, 308-318.	1.3	6
87	MicroRNA profiles are associated with lymphocyte count after a major traumatic injury: a potential role for immunometabolism?. <i>British Journal of Anaesthesia</i> , 2018, 121, e23-e24.	1.5	0
88	miR-10b-5p is a novel Th17 regulator present in Th17 cells from ankylosing spondylitis. <i>Annals of the Rheumatic Diseases</i> , 2017, 76, 620-625.	0.5	61
89	A functional SNP associated with atopic dermatitis controls cell type-specific methylation of the VSTM1 gene locus. <i>Genome Medicine</i> , 2017, 9, 18.	3.6	30
90	Enhanced understanding of the host-pathogen interaction in sepsis: new opportunities for omic approaches. <i>Lancet Respiratory Medicine</i> , 2017, 5, 212-223.	5.2	33

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91	Insights from early experience of a Rare Disease Genomic Medicine Multidisciplinary Team: a qualitative study. <i>European Journal of Human Genetics</i> , 2017, 25, 680-686.	1.4	24
92	Shared and Distinct Aspects of the Sepsis Transcriptomic Response to Fecal Peritonitis and Pneumonia. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2017, 196, 328-339.	2.5	178
93	A point mutation in the ion conduction pore of AMPA receptor GRIA3 causes dramatically perturbed sleep patterns as well as intellectual disability. <i>Human Molecular Genetics</i> , 2017, 26, 3869-3882.	1.4	35
94	Classification of patients with sepsis according to blood genomic endotype: a prospective cohort study. <i>Lancet Respiratory Medicine</i> , 2017, 5, 816-826.	5.2	381
95	Unique transcriptome signatures and GM-CSF expression in lymphocytes from patients with spondyloarthritis. <i>Nature Communications</i> , 2017, 8, 1510.	5.8	118
96	Investigation of a possible extended risk haplotype in the IL23R region associated with ankylosing spondylitis. <i>Genes and Immunity</i> , 2017, 18, 105-108.	2.2	8
97	ReliableGenome: annotation of genomic regions with high/low variant calling concordance. <i>Bioinformatics</i> , 2017, 33, 155-160.	1.8	7
98	Pathogenic implications for autoimmune mechanisms derived by comparative eQTL analysis of CD4+ versus CD8+ T cells. <i>PLoS Genetics</i> , 2017, 13, e1006643.	1.5	110
99	High resolution HLA haplotyping by imputation for a British population bioresource. <i>Human Immunology</i> , 2017, 78, 242-251.	1.2	31
100	Whole-genome sequencing of spermatocytic tumors provides insights into the mutational processes operating in the male germline. <i>PLoS ONE</i> , 2017, 12, e0178169.	1.1	36
101	Characterisation of the global transcriptional response to heat shock and the impact of individual genetic variation. <i>Genome Medicine</i> , 2016, 8, 87.	3.6	4
102	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. <i>American Journal of Human Genetics</i> , 2016, 99, 125-138.	2.6	92
103	Tensor decomposition for multiple-tissue gene expression experiments. <i>Nature Genetics</i> , 2016, 48, 1094-1100.	9.4	142
104	Chronic mucocutaneous candidiasis: characterization of a family with STAT-1 gain-of-function and development of an <i>ex-vivo</i> assay for Th17 deficiency of diagnostic utility. <i>Clinical and Experimental Immunology</i> , 2016, 184, 216-227.	1.1	25
105	Gene panel sequencing improves the diagnostic work-up of patients with idiopathic erythrocytosis and identifies new mutations. <i>Haematologica</i> , 2016, 101, 1306-1318.	1.7	66
106	HLA-C Level Is Regulated by a Polymorphic Oct1 Binding Site in the HLA-C Promoter Region. <i>American Journal of Human Genetics</i> , 2016, 99, 1353-1358.	2.6	49
107	XGR software for enhanced interpretation of genomic summary data, illustrated by application to immunological traits. <i>Genome Medicine</i> , 2016, 8, 129.	3.6	137
108	Genomic landscape of the individual host response and outcomes in sepsis: a prospective cohort study. <i>Lancet Respiratory Medicine</i> , 2016, 4, 259-271.	5.2	536

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109	An ankylosing spondylitis-associated genetic variant in the <i>IL23R-IL12RB2</i> intergenic region modulates enhancer activity and is associated with increased Th1-cell differentiation. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 2150-2156.	0.5	45
110	The genetic association of <i>RUNX3</i> with ankylosing spondylitis can be explained by allele-specific effects on IRF4 recruitment that alter gene expression. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 1534-1540.	0.5	48
111	The Role of Micrnas in The Development of Hospital Acquired Infection in Polytrauma Patients. <i>Intensive Care Medicine Experimental</i> , 2015, 3, .	0.9	1
112	Distinct Transcriptional and Anti-Mycobacterial Profiles of Peripheral Blood Monocytes Dependent on the Ratio of Monocytes: Lymphocytes. <i>EBioMedicine</i> , 2015, 2, 1619-1626.	2.7	61
113	A functional AT/G polymorphism in the 5'UTR-untranslated region of SETDB2 in the IgE locus on human chromosome 13q14. <i>Genes and Immunity</i> , 2015, 16, 488-494.	2.2	6
114	Genome-wide association study of survival from sepsis due to pneumonia: an observational cohort study. <i>Lancet Respiratory Medicine</i> , 2015, 3, 53-60.	5.2	166
115	Reply to Pembrey et al: ZNF277 microdeletions, specific language impairment and the meiotic mismatch methylation (3M) hypothesis. <i>European Journal of Human Genetics</i> , 2015, 23, 1113-1115.	1.4	2
116	Preclinical target validation using patient-derived cells. <i>Nature Reviews Drug Discovery</i> , 2015, 14, 149-150.	21.5	46
117	Cell Specific eQTL Analysis without Sorting Cells. <i>PLoS Genetics</i> , 2015, 11, e1005223.	1.5	115
118	Genomic modulators of gene expression in human neutrophils. <i>Nature Communications</i> , 2015, 6, 7545.	5.8	120
119	Transcriptomic profiling facilitates classification of response to influenza challenge. <i>Journal of Molecular Medicine</i> , 2015, 93, 105-114.	1.7	38
120	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. <i>Nature Genetics</i> , 2015, 47, 717-726.	9.4	310
121	Genetic variants associated with non-typhoidal Salmonella bacteraemia in African children. <i>Lancet</i> , 2015, 385, S13.	6.3	5
122	Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus. <i>Nature Genetics</i> , 2015, 47, 1457-1464.	9.4	730
123	Application of whole genome and RNA sequencing to investigate the genomic landscape of common variable immunodeficiency disorders. <i>Clinical Immunology</i> , 2015, 160, 301-314.	1.4	100
124	Gain-of-Function Mutations in ZIC1 Are Associated with Coronal Craniosynostosis and Learning Disability. <i>American Journal of Human Genetics</i> , 2015, 97, 378-388.	2.6	56
125	Genome-wide analysis identifies a role for common copy number variants in specific language impairment. <i>European Journal of Human Genetics</i> , 2015, 23, 1370-1377.	1.4	46
126	Genomic mapping of the MHC transactivator CIITA using an integrated ChIP-seq and genetical genomics approach. <i>Genome Biology</i> , 2014, 15, 494.	3.8	32

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127	Approaches for establishing the function of regulatory genetic variants involved in disease. <i>Genome Medicine</i> , 2014, 6, 92.	3.6	34
128	Fine mapping genetic determinants of the highly variably expressed MHC gene ZFP57. <i>European Journal of Human Genetics</i> , 2014, 22, 568-571.	1.4	16
129	Homozygous microdeletion of exon 5 in ZNF277 in a girl with specific language impairment. <i>European Journal of Human Genetics</i> , 2014, 22, 1165-1171.	1.4	27
130	Increased prevalence of sex chromosome aneuploidies in specific language impairment and dyslexia. <i>Developmental Medicine and Child Neurology</i> , 2014, 56, 346-353.	1.1	42
131	Choice of transcripts and software has a large effect on variant annotation. <i>Genome Medicine</i> , 2014, 6, 26.	3.6	158
132	Whole-genome sequencing of bladder cancers reveals somatic CDKN1A mutations and clinicopathological associations with mutation burden. <i>Nature Communications</i> , 2014, 5, 3756.	5.8	81
133	Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. <i>Human Molecular Genetics</i> , 2014, 23, 3200-3211.	1.4	222
134	An Integrated Approach to Defining Genetic and Environmental Determinants for Major Clinical Outcomes Involving Vitamin D. <i>Molecular Diagnosis and Therapy</i> , 2014, 18, 261-272.	1.6	14
135	Associations of HLA alleles with specific language impairment. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 1.	1.5	67
136	Innate Immune Activity Conditions the Effect of Regulatory Variants upon Monocyte Gene Expression. <i>Science</i> , 2014, 343, 1246949.	6.0	706
137	Genetics of gene expression in immunity to infection. <i>Current Opinion in Immunology</i> , 2014, 30, 63-71.	2.4	54
138	Integrating mapping-, assembly- and haplotype-based approaches for calling variants in clinical sequencing applications. <i>Nature Genetics</i> , 2014, 46, 912-918.	9.4	937
139	Erythrocytosis associated with a novel missense mutation in the BPGM gene. <i>Haematologica</i> , 2014, 99, e201-e204.	1.7	35
140	Meta-analysis of genome-wide association studies identifies ten loci influencing allergic sensitization. <i>Nature Genetics</i> , 2013, 45, 902-906.	9.4	221
141	Major Histocompatibility Complex Genomics and Human Disease. <i>Annual Review of Genomics and Human Genetics</i> , 2013, 14, 301-323.	2.5	580
142	Systematic identification of trans eQTLs as putative drivers of known disease associations. <i>Nature Genetics</i> , 2013, 45, 1238-1243.	9.4	1,544
143	Genomic modulators of the immune response. <i>Trends in Genetics</i> , 2013, 29, 74-83.	2.9	52
144	Germline mutations affecting the proofreading domains of POLE and POLD1 predispose to colorectal adenomas and carcinomas. <i>Nature Genetics</i> , 2013, 45, 136-144.	9.4	851

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145	Congenital myasthenic syndromes due to mutations in <i>ALG2</i> and <i>ALG14</i> . <i>Brain</i> , 2013, 136, 944-956.	3.7	117
146	Gene-centric meta-analyses of 108 912 individuals confirm known body mass index loci and reveal three novel signals. <i>Human Molecular Genetics</i> , 2013, 22, 184-201.	1.4	82
147	Homozygous mutations in a predicted endonuclease are a novel cause of congenital dyserythropoietic anemia type I. <i>Haematologica</i> , 2013, 98, 1383-1387.	1.7	71
148	Genetics of gene expression in primary immune cells identifies cell type-specific master regulators and roles of HLA alleles. <i>Nature Genetics</i> , 2012, 44, 502-510.	9.4	445
149	Resolving the variable genome and epigenome in human disease. <i>Journal of Internal Medicine</i> , 2012, 271, 379-391.	2.7	14
150	Functional Differences Exist between TNF Promoters Encoding the Common \sim 237G SNP and the Rarer HLA-B*5701-Linked A Variant. <i>PLoS ONE</i> , 2012, 7, e40100.	1.1	11
151	Allele-specific transcription of the asthma-associated PHD finger protein 11 gene (PHF11) modulated by octamer-binding transcription factor 1 (Oct-1). <i>Journal of Allergy and Clinical Immunology</i> , 2011, 127, 1054-1062.e2.	1.5	15
152	Pervasive haplotypic variation in the spliceo-transcriptome of the human major histocompatibility complex. <i>Genome Research</i> , 2011, 21, 1042-1054.	2.4	63
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