Mark Joseph Daly

List of Publications by Year
in descending order

589	314,033	29 195	4 519
papers	citations	h-index	g-index

759	759	759	209204
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	PLINK: A Tool Set for Whole-Genome Association and Population-Based Linkage Analyses. American Journal of Human Genetics, 2007, 81, 559-575.	2.6	26,761
2	The Genome Analysis Toolkit: A MapReduce framework for analyzing next-generation DNA sequencing data. Genome Research, 2010, 20, 1297-1303.	2.4	21,358
3	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
4	Haploview: analysis and visualization of LD and haplotype maps. Bioinformatics, 2005, 21, 263-265.	1.8	13,223
5	A framework for variation discovery and genotyping using next-generation DNA sequencing data. Nature Genetics, 2011, 43, 491-498.	9.4	10,018
6	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	13.7	9,051
7	PGC-1Î±-responsive genes involved in oxidative phosphorylation are coordinately downregulated in human diabetes. Nature Genetics, 2003, 34, 267-273.	9.4	8,185
8	Biological insights from 108 schizophrenia-associated genetic loci. Nature, 2014, 511, 421-427.	13.7	6,934
9	MAPMAKER: An interactive computer package for constructing primary genetic linkage maps of experimental and natural populations. Genomics, 1987, 1, 174-181.	1.3	6,607
10	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
11	The Structure of Haplotype Blocks in the Human Genome. Science, 2002, 296, 2225-2229.	6.0	5,300
12	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	13.7	4,137
13	Hostâ€“microbe interactions have shaped the genetic architecture of inflammatory bowel disease. Nature, 2012, 491, 119-124.	13.7	4,038
14	LD Score regression distinguishes confounding from polygenicity in genome-wide association studies. Nature Genetics, 2015, 47, 291-295.	9.4	3,905
15	The landscape of somatic copy-number alteration across human cancers. Nature, 2010, 463, 899-905.	13.7	3,331
16	An atlas of genetic correlations across human diseases and traits. Nature Genetics, 2015, 47, 1236-1241.	9.4	3,145
17	A map of human genome sequence variation containing 1.42 million single nucleotide polymorphisms. Nature, 2001, 409, 928-933.	13.7	2,794
18	A Genome-Wide Association Study Identifies IL23R as an Inflammatory Bowel Disease Gene. Science, 2006, 314, 1461-1463.	6.0	2,739

#	Article	IF	Citations
19	Genome-wide association studies for common diseases and complex traits. Nature Reviews Genetics, 2005, 6, 95-108.	7.7	2,717
20	Identification of risk loci with shared effects on five major psychiatric disorders: a genome-wide analysis. Lancet, The, 2013, 381, 1371-1379.	6.3	2,643
21	Integrating common and rare genetic variation in diverse human populations. Nature, 2010, 467, 52-58.	13.7	2,625
22	Genome-Wide Association Analysis Identifies Loci for Type 2 Diabetes and Triglyceride Levels. Science, 2007, 316, 1331-1336.	6.0	2,623
23	Genome-wide association defines more than 30 distinct susceptibility loci for Crohn's disease. Nature Genetics, 2008, 40, 955-962.	9.4	2,422
24	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	13.7	2,400
25	Genome-wide meta-analysis increases to 71 the number of confirmed Crohn's disease susceptibility loci. Nature Genetics, 2010, 42, 1118-1125.	9.4	2,284
26	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	13.7	2,254
27	Genome sequence, comparative analysis and haplotype structure of the domestic dog. Nature, 2005, 438, 803-819.	13.7	2,215
28	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
29	Partitioning heritability by functional annotation using genome-wide association summary statistics. Nature Genetics, 2015, 47, 1228-1235.	9.4	2,045
30	Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations. Nature Genetics, 2015, 47, 979-986.	9.4	1,965
31	Plasma HDL cholesterol and risk of myocardial infarction: a mendelian randomisation study. Lancet, The, 2012, 380, 572-580.	6.3	1,937
32	Schizophrenia risk from complex variation of complement component 4. Nature, 2016, 530, 177-183.	13.7	1,915
33	Genome-wide association study identifies five new schizophrenia loci. Nature Genetics, 2011, 43, 969-976.	9.4	1,758
34	Efficient Control of Population Structure in Model Organism Association Mapping. Genetics, 2008, 178, 1709-1723.	1.2	1,752
35	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. Nature Genetics, 2008, 40, 638-645.	9.4	1,683
36	The common PPARÎ³ Pro12Ala polymorphism is associated with decreased risk of type 2 diabetes. Nature Genetics, 2000, 26, 76-80.	9.4	1,672

#	Article	IF	Citations
37	Genetic and epigenetic fine mapping of causal autoimmune disease variants. Nature, 2015, 518, 337-343.	13.7	1,669
38	Clinical use of current polygenic risk scores may exacerbate health disparities. Nature Genetics, 2019, 51, 584-591.	9.4	1,664
39	Genome-wide association study identifies new susceptibility loci for Crohn disease and implicates autophagy in disease pathogenesis. Nature Genetics, 2007, 39, 596-604.	9.4	1,633
40	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. Nature Genetics, 2010, 42, 579-589.	9.4	1,631
41	Efficiency and power in genetic association studies. Nature Genetics, 2005, 37, 1217-1223.	9.4	1,597
42	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	13.7	1,597
43	High-resolution haplotype structure in the human genome. Nature Genetics, 2001, 29, 229-232.	9.4	1,596
44	Discovery of the first genome-wide significant risk loci for attention deficit/hyperactivity disorder. Nature Genetics, 2019, 51, 63-75.	9.4	1,594
45	Identification of common genetic risk variants for autism spectrum disorder. Nature Genetics, 2019, 51, 431-444.	9.4	1,538
46	Risk Alleles for Multiple Sclerosis Identified by a Genomewide Study. New England Journal of Medicine, 2007, 357, 851-862.	13.9	1,529
47	Replicating genotypeâ€“phenotype associations. Nature, 2007, 447, 655-660.	13.7	1,509
48	Association between Microdeletion and Microduplication at 16p11.2 and Autism. New England Journal of Medicine, 2008, 358, 667-675.	13.9	1,476
49	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
50	Genetic Mapping in Human Disease. Science, 2008, 322, 881-888.	6.0	1,289
51	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
52	Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47. Nature Genetics, 2011, 43, 246-252.	9.4	1,201
53	Genome-wide association study identifies 30 loci associated with bipolar disorder. Nature Genetics, 2019, 51, 793-803.	9.4	1,191
54	Guidelines for investigating causality of sequence variants in human disease. Nature, 2014, 508, 469-476.	13.7	1,130

#	Article	IF	Citations
55	Human Demographic History Impacts Genetic Risk Prediction across Diverse Populations. American Journal of Human Genetics, 2017, 100, 635-649.	2.6	1,120
56	Collaborative genome-wide association analysis supports a role for ANK3 and CACNA1C in bipolar disorder. Nature Genetics, 2008, 40, 1056-1058.	9.4	1,102
57	Modeling Linkage Disequilibrium Increases Accuracy of Polygenic Risk Scores. American Journal of Human Genetics, 2015, 97, 576-592.	2.6	1,098
58	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
59	Genetic architectures of psychiatric disorders: the emerging picture and its implications. Nature Reviews Genetics, 2012, 13, 537-551.	7.7	1,025
60	A mega-analysis of genome-wide association studies for major depressive disorder. Molecular Psychiatry, 2013, 18, 497-511.	4.1	1,002
61	High-throughput discovery of novel developmental phenotypes. Nature, 2016, 537, 508-514.	13.7	1,001
62	Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. Nature Genetics, 2009, 41, 334-341.	9.4	990
63	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	13.7	952
64	HLA-B*5701 genotype is a major determinant of drug-induced liver injury due to flucloxacillin. Nature Genetics, 2009, 41, 816-819.	9.4	950
65	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	9.4	943
66	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	13.7	929
67	A comprehensive genetic map of the mouse genome. Nature, 1996, 380, 149-152.	13.7	853
68	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. Science, 2018, 359, 693-697.	6.0	851
69	Integrated detection and population-genetic analysis of SNPs and copy number variation. Nature Genetics, 2008, 40, 1166-1174.	9.4	838
70	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	9.4	838
71	Intra- and Inter-cellular Rewiring of the Human Colon during Ulcerative Colitis. Cell, 2019, 178, 714-730.e22.	13.5	806
72	Common variants associated with plasma triglycerides and risk for coronary artery disease. Nature Genetics, 2013, 45, 1345-1352.	9.4	754

#	Article	IF	Citations
73	Genetic variation in the 5q31 cytokine gene cluster confers susceptibility to Crohn disease. Nature Genetics, 2001, 29, 223-228.	9.4	730
74	Meta-analysis of genome scans and replication identify CD6, IRF8 and TNFRSF1A as new multiple sclerosis susceptibility loci. Nature Genetics, 2009, 41, 776-782.	9.4	729
75	Genome Sequence and Comparative Analysis of the Solvent-Producing Bacterium Clostridium acetobutylicum. Journal of Bacteriology, 2001, 183, 4823-4838.	1.0	725
76	Integrated genotype calling and association analysis of SNPs, common copy number polymorphisms and rare CNVs. Nature Genetics, 2008, 40, 1253-1260.	9.4	712
77	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. Nature Neuroscience, 2015, 18, 199-209.	7.1	701
78	Estimation of the multiple testing burden for genomewide association studies of nearly all common variants. Genetic Epidemiology, 2008, 32, 381-385.	0.6	699
79	Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. Nature Genetics, 2011, 43, 1066-1073.	9.4	698
80	Seven new loci associated with age-related macular degeneration. Nature Genetics, 2013, 45, 433-439.	9.4	687
81	A high-resolution HLA and SNP haplotype map for disease association studies in the extended human MHC. Nature Genetics, 2006, 38, 1166-1172.	9.4	686
82	Genome-wide association analysis of metabolic traits in a birth cohort from a founder population. Nature Genetics, 2009, 41, 35-46.	9.4	676
83	Common deletion polymorphisms in the human genome. Nature Genetics, 2006, 38, 86-92.	9.4	656
84	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	6.0	646
85	Whole-genome association study of bipolar disorder. Molecular Psychiatry, 2008, 13, 558-569.	4.1	642
86	Exome Sequencing,ANGPTL3Mutations, and Familial Combined Hypolipidemia. New England Journal of Medicine, 2010, 363, 2220-2227.	13.9	640
87	Mapping the human genetic architecture of COVID-19. Nature, 2021, 600, 472-477.	13.7	640
88	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. Cell, 2018, 173, 1705-1715.e16.	13.5	623
89	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614
90	Inherited determinants of Crohn's disease and ulcerative colitis phenotypes: a genetic association study. Lancet, The, 2016, 387, 156-167.	6.3	607

#	Article	IF	Citations
91	Deletion polymorphism upstream of IRGM associated with altered IRGM expression and Crohn's disease. Nature Genetics, 2008, 40, 1107-1112.	9.4	604
92	A common haplotype of interferon regulatory factor 5 (IRF5) regulates splicing and expression and is associated with increased risk of systemic lupus erythematosus. Nature Genetics, 2006, 38, 550-555.	9.4	593
93	Demographic history and rare allele sharing among human populations. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11983-11988.	3.3	589
94	Fine mapping of type 1 diabetes susceptibility loci and evidence for colocalization of causal variants with lymphoid gene enhancers. Nature Genetics, 2015, 47, 381-386.	9.4	589
95	The ExAC browser: displaying reference data information from over 60 000 exomes. Nucleic Acids Research, 2017, 45, D840-D845.	6.5	587
96	Calibrating a coalescent simulation of human genome sequence variation. Genome Research, 2005, 15, 1576-1583.	2.4	581
97	Genome-wide association identifies multiple ulcerative colitis susceptibility loci. Nature Genetics, 2010, 42, 332-337.	9.4	572
98	Common variation in three genes, including a noncoding variant in CFH, strongly influences risk of age-related macular degeneration. Nature Genetics, 2006, 38, 1055-1059.	9.4	570
99	A genome-wide linkage and association scan reveals novel loci for autism. Nature, 2009, 461, 802-808.	13.7	570
100	Searching for missing heritability: Designing rare variant association studies. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E455-64.	3.3	570
101	Partitioning Heritability of Regulatory and Cell-Type-Specific Variants across 11 Common Diseases. American Journal of Human Genetics, 2014, 95, 535-552.	2.6	569
102	A cross-population atlas of genetic associations for 220 human phenotypes. Nature Genetics, 2021, 53, 1415-1424.	9.4	560
103	Disruption of Neurexin 1 Associated with Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 199-207.	2.6	545
104	Pervasive Sharing of Genetic Effects in Autoimmune Disease. PLoS Genetics, 2011, 7, e1002254.	1.5	540
105	Genetic variants near TNFAIP3 on 6q23 are associated with systemic lupus erythematosus. Nature Genetics, 2008, 40, 1059-1061.	9.4	534
106	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. Cell, 2012, 149, 525-537.	13.5	534
107	Testing for an Unusual Distribution of Rare Variants. PLoS Genetics, 2011, 7, e1001322.	1.5	530
108	Variation in genome-wide mutation rates within and between human families. Nature Genetics, 2011, 43, 712-714.	9.4	525

#	Article	IF	Citations
109	Meta-analysis of 375,000 individuals identifies 38 susceptibility loci for migraine. Nature Genetics, 2016, 48, 856-866.	9.4	520
110	Two independent alleles at 6q23 associated with risk of rheumatoid arthritis. Nature Genetics, 2007, 39, 1477-1482.	9.4	497
111	A genetic linkage map of the laboratory rat, Rattus norvegicus. Nature Genetics, 1995, 9, 63-69.	9.4	477
112	Common variants at CD40 and other loci confer risk of rheumatoid arthritis. Nature Genetics, 2008, 40, 1216-1223.	9.4	476
113	Fine-mapping inflammatory bowel disease loci to single-variant resolution. Nature, 2017, 547, 173-178.	13.7	473
114	Common variants at five new loci associated with early-onset inflammatory bowel disease. Nature Genetics, 2009, 41, 1335-1340.	9.4	459
115	Proteins Encoded in Genomic Regions Associated with Immune-Mediated Disease Physically Interact and Suggest Underlying Biology. PLoS Genetics, 2011, 7, e1001273.	1.5	450
116	Genomewide Search in Canadian Families with Inflammatory Bowel Disease Reveals Two Novel Susceptibility Loci. American Journal of Human Genetics, 2000, 66, 1863-1870.	2.6	449
117	The mosaic structure of variation in the laboratory mouse genome. Nature, 2002, 420, 574-578.	13.7	448
118	Comparative genetic architectures of schizophrenia in East Asian and European populations. Nature Genetics, 2019, 51, 1670-1678.	9.4	440
119	Methods for High-Density Admixture Mapping of Disease Genes. American Journal of Human Genetics, 2004, 74, 979-1000.	2.6	437
120	Genome-wide mapping of DNase hypersensitive sites using massively parallel signature sequencing (MPSS). Genome Research, 2005, 16, 123-131.	2.4	431
121	Common Inherited Variation in Mitochondrial Genes Is Not Enriched for Associations with Type 2 Diabetes or Related Glycemic Traits. PLoS Genetics, 2010, 6, e1001058.	1.5	429
122	Three functional variants of IFN regulatory factor 5 (IRF5) define risk and protective haplotypes for human lupus. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 6758-6763.	3.3	428
123	Ipr1 gene mediates innate immunity to tuberculosis. Nature, 2005, 434, 767-772.	13.7	425
124	Meta-Analysis of Genome-Wide Association Studies of Attention-Deficit/Hyperactivity Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2010, 49, 884-897.	0.3	423
125	Susceptibility to Amoxicillin-Clavulanate-Induced Liver Injury Is Influenced by Multiple HLA Class I and II Alleles. Gastroenterology, 2011, 141, 338-347.	0.6	412
126	Whole population, genome-wide mapping of hidden relatedness. Genome Research, 2009, 19, 318-326.	2.4	411

#	Article	IF	Citations
127	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. American Journal of Psychiatry, 2017, 174, 850-858.	4.0	410
128	A sequence-based variation map of 8.27 million SNPs in inbred mouse strains. Nature, 2007, 448, 1050-1053.	13.7	406
129	Genome-wide association study of advanced age-related macular degeneration identifies a role of the hepatic lipase gene (LIPC). Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7395-7400.	3.3	406
130	Variation in complement factor 3 is associated with risk of age-related macular degeneration. Nature Genetics, 2007, 39, 1200-1201.	9.4	405
131	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. Nature Genetics, 2017, 49, 978-985.	9.4	401
132	Large-scale discovery and genotyping of single-nucleotide polymorphisms in the mouse. Nature Genetics, 2000, 24, 381-386.	9.4	395
133	Genomewide Association Studies: History, Rationale, and Prospects for Psychiatric Disorders. American Journal of Psychiatry, 2009, 166, 540-556.	4.0	391
134	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. Nature Neuroscience, 2016, 19, 571-577.	7.1	388
135	Genetics of 35 blood and urine biomarkers in the UK Biobank. Nature Genetics, 2021, 53, 185-194.	9.4	377
136	Largest GWAS of PTSD (N=20â€‰070) yields genetic overlap with schizophrenia and sex differences in heritability. Molecular Psychiatry, 2018, 23, 666-673.	4.1	374
137	Identifying Relationships among Genomic Disease Regions: Predicting Genes at Pathogenic SNP Associations and Rare Deletions. PLoS Genetics, 2009, 5, e1000534.	1.5	371
138	Ulcerative colitisâ€“risk loci on chromosomes 1p36 and 12q15 found by genome-wide association study. Nature Genetics, 2009, 41, 216-220.	9.4	364
139	International meta-analysis of PTSD genome-wide association studies identifies sex- and ancestry-specific genetic risk loci. Nature Communications, 2019, 10, 4558.	5.8	363
140	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. PLoS Genetics, 2014, 10, e1004494.	1.5	351
141	LRRK2 Is Involved in the IFN-Î³ Response and Host Response to Pathogens. Journal of Immunology, 2010, 185, 5577-5585.	0.4	350
142	Common Single Nucleotide Polymorphisms in TCF7L2 Are Reproducibly Associated With Type 2 Diabetes and Reduce the Insulin Response to Glucose in Nondiabetic Individuals. Diabetes, 2006, 55, 2890-2895.	0.3	346
143	Validating, augmenting and refining genome-wide association signals. Nature Reviews Genetics, 2009, 10, 318-329.	7.7	339
144	Genome-wide meta-analysis identifies new susceptibility loci for migraine. Nature Genetics, 2013, 45, 912-917.	9.4	338

#	Article	IF	Citations
145	Genomeâ€wide association scan of quantitative traits for attention deficit hyperactivity disorder identifies novel associations and confirms candidate gene associations. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1345-1354.	1.1	335
146	Genome-wide association study of migraine implicates a common susceptibility variant on 8q22.1. Nature Genetics, 2010, 42, 869-873.	9.4	332
147	High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. Nature Genetics, 2010, 42, 851-858.	9.4	332
148	Genome-wide enhancer maps link risk variants to disease genes. Nature, 2021, 593, 238-243.	13.7	332
149	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. Nature Genetics, 2016, 48, 552-555.	9.4	326
150	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	13.7	326
151	Variation near complement factor I is associated with risk of advanced AMD. European Journal of Human Genetics, 2009, 17, 100-104.	1.4	324
152	Complex host genetics influence the microbiome in inflammatory bowel disease. Genome Medicine, 2014, 6, 107.	3.6	322
153	Rare variants in CFI, C3 and C9 are associated with high risk of advanced age-related macular degeneration. Nature Genetics, 2013, 45, 1366-1370.	9.4	311
154	Genetic variants at CD28, PRDM1 and CD2/CD58 are associated with rheumatoid arthritis risk. Nature Genetics, 2009, 41, 1313-1318.	9.4	306
155	A genome-wide scan for linkage to human exceptional longevity identifies a locus on chromosome 4. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 10505-10508.	3.3	300
156	Microdeletion/duplication at 15q13.2q13.3 among individuals with features of autism and other neuropsychiatric disorders. Journal of Medical Genetics, 2009, 46, 242-248.	1.5	300
157	Impaired Autophagy of an Intracellular Pathogen Induced by a Crohn's Disease Associated ATG16L1 Variant. PLoS ONE, 2008, 3, e3391.	1.1	299
158	Atg16L1 T300A variant decreases selective autophagy resulting in altered cytokine signaling and decreased antibacterial defense. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 7741-7746.	3.3	298
159	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	9.4	298
160	Human knockouts and phenotypic analysis in a cohort with a high rate of consanguinity. Nature, 2017, 544, 235-239.	13.7	292
161	A rare penetrant mutation in CFH confers high risk of age-related macular degeneration. Nature Genetics, 2011, 43, 1232-1236.	9.4	291
162	Abundant contribution of short tandem repeats to gene expression variation in humans. Nature Genetics, 2016, 48, 22-29.	9.4	291

#	Article	IF	Citations
163	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	5.8	289
164	New models of collaboration in genome-wide association studies: the Genetic Association Information Network. Nature Genetics, 2007, 39, 1045-1051.	9.4	288
165	Genomewide Scan of Multiple Sclerosis in Finnish Multiplex Families. American Journal of Human Genetics, 1997, 61, 1379-1387.	2.6	284
166	Haplotype Structure and Genotype-Phenotype Correlations of the Sulfonylurea Receptor and the Islet ATP-Sensitive Potassium Channel Gene Region. Diabetes, 2004, 53, 1360-1368.	0.3	284
167	The Evaluation of Tools Used to Predict the Impact of Missense Variants Is Hindered by Two Types of Circularity. Human Mutation, 2015, 36, 513-523.	1.1	283
168	Loss-of-heterozygosity analysis of small-cell lung carcinomas using single-nucleotide polymorphism arrays. Nature Biotechnology, 2000, 18, 1001-1005.	9.4	282
169	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	9.4	281
170	Polygenic and clinical risk scores and their impact on age at onset and prediction of cardiometabolic diseases and common cancers. Nature Medicine, 2020, 26, 549-557.	15.2	281
171	High-density mapping of the MHC identifies a shared role for HLA-DRB1*01:03 in inflammatory bowel diseases and heterozygous advantage in ulcerative colitis. Nature Genetics, 2015, 47, 172-179.	9.4	280
172	Human genome sequence variation and the influence of gene history, mutation and recombination. Nature Genetics, 2002, 32, 135-142.	9.4	278
173	Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. ELife, 2019, 8, .	2.8	276
174	Evaluating and improving power in whole-genome association studies using fixed marker sets. Nature Genetics, 2006, 38, 663-667.	9.4	274
175	Functional variants in the LRRK2 gene confer shared effects on risk for Crohnâ€™s disease and Parkinsonâ€™s disease. Science Translational Medicine, 2018, 10, .	5.8	273
176	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 1226-1231.	9.4	270
177	Renal disease susceptibility and hypertension are under independent genetic control in the fawn-hooded rat. Nature Genetics, 1996, 12, 44-51.	9.4	257
178	Prediction Model for Prevalence and Incidence of Advanced Age-Related Macular Degeneration Based on Genetic, Demographic, and Environmental Variables. , 2009, 50, 2044.		257
179	The iPSYCH2012 caseâ€“cohort sample: new directions for unravelling genetic and environmental architectures of severe mental disorders. Molecular Psychiatry, 2018, 23, 6-14.	4.1	257
180	Genetic architecture of complex traits: Large phenotypic effects and pervasive epistasis. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 19910-19914.	3.3	254

#	Article	IF	Citations
181	CARD15 Genetic Variation in a Quebec Population: Prevalence, Genotype-Phenotype Relationship, and Haplotype Structure. American Journal of Human Genetics, 2002, 71, 74-83.	2.6	253
182	Integrated Model of De Novo and Inherited Genetic Variants Yields Greater Power to Identify Risk Genes. PLoS Genetics, 2013, 9, e1003671.	1.5	253
183	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
184	Predicting Polygenic Risk of Psychiatric Disorders. Biological Psychiatry, 2019, 86, 97-109.	0.7	252
185	Exome array analysis identifies new loci and low-frequency variants influencing insulin processing and secretion. Nature Genetics, 2013, 45, 197-201.	9.4	247
186	Discovery of stimulation-responsive immune enhancers with CRISPR activation. Nature, 2017, 549, 111-115.	13.7	247
187	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	9.4	245
188	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	3.8	242
189	Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. Nature Genetics, 2012, 44, 631-635.	9.4	239
190	Mutations causing medullary cystic kidney disease type 1 lie in a large VNTR in MUC1 missed by massively parallel sequencing. Nature Genetics, 2013, 45, 299-303.	9.4	237
191	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	2.6	237
192	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	9.4	235
193	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	6.0	234
194	Common variants near FRK/COL10A1 and VEGFA are associated with advanced age-related macular degeneration. Human Molecular Genetics, 2011, 20, 3699-3709.	1.4	232
195	Mapping of multiple susceptibility variants within the MHC region for 7 immune-mediated diseases. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 18680-18685.	3.3	231
196	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	9.4	230
197	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nature Genetics, 2012, 44, 390-397.	9.4	229
198	Genomeâ€wide association scan of attention deficit hyperactivity disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1337-1344.	1.1	228

#	Article	IF	Citations
199	Transferability of tag SNPs in genetic association studies in multiple populations. Nature Genetics, 2006, 38, 1298-1303.	9.4	224
200	Inflammatory bowel disease susceptibility loci defined by genome scan meta-analysis of 1952 affected relative pairs. Human Molecular Genetics, 2004, 13, 763-770.	1.4	219
201	Combined Analysis from Eleven Linkage Studies of Bipolar Disorder Provides Strong Evidence of Susceptibility Loci on Chromosomes 6q and 8q. American Journal of Human Genetics, 2005, 77, 582-595.	2.6	218
202	Histocompatible Embryonic Stem Cells by Parthenogenesis. Science, 2007, 315, 482-486.	6.0	217
203	Genome-wide meta-analysis in alopecia areata resolves HLA associations and reveals two new susceptibility loci. Nature Communications, 2015, 6, 5966.	5.8	213
204	Myosin IXB variant increases the risk of celiac disease and points toward a primary intestinal barrier defect. Nature Genetics, 2005, 37, 1341-1344.	9.4	211
205	Genome-wide Association Study in a High-Risk Isolate for Multiple Sclerosis Reveals Associated Variants in STAT3 Gene. American Journal of Human Genetics, 2010, 86, 285-291.	2.6	210
206	Ataxia, Dementia, and Hypogonadotropism Caused by Disordered Ubiquitination. New England Journal of Medicine, 2013, 368, 1992-2003.	13.9	208
207	Genetic Adaptation of Fatty-Acid Metabolism: A Human-Specific Haplotype Increasing the Biosynthesis of Long-Chain Omega-3 and Omega-6 Fatty Acids. American Journal of Human Genetics, 2012, 90, 809-820.	2.6	205
208	RICOPILI: Rapid Imputation for COnsortias PIpeLIne. Bioinformatics, 2020, 36, 930-933.	1.8	201
209	Genetic Analysis of Human Traits In Vitro: Drug Response and Gene Expression in Lymphoblastoid Cell Lines. PLoS Genetics, 2008, 4, e1000287.	1.5	200
210	zCall: a rare variant caller for array-based genotyping. Bioinformatics, 2012, 28, 2543-2545.	1.8	195
211	The vibrator Mutation Causes Neurodegeneration via Reduced Expression of PITPÎ±: Positional Complementation Cloning and Extragenic Suppression. Neuron, 1997, 18, 711-722.	3.8	193
212	Genetic mapping of a murine locus controlling development of T helper 1/T helper 2 type responses.. Proceedings of the National Academy of Sciences of the United States of America, 1996, 93, 12467-12472.	3.3	191
213	Whole exome sequencing study identifies novel rare and common Alzheimerâ€™s-Associated variants involved in immune response and transcriptional regulation. Molecular Psychiatry, 2020, 25, 1859-1875.	4.1	191
214	A Meta-Analysis of Genome-Wide Association Scans Identifies IL18RAP, PTPN2, TAGAP, and PUS10 As Shared Risk Loci for Crohn's Disease and Celiac Disease. PLoS Genetics, 2011, 7, e1001283.	1.5	187
215	The role of the CD58 locus in multiple sclerosis. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 5264-5269.	3.3	185
216	Deep Resequencing of GWAS Loci Identifies Rare Variants in CARD9, IL23R and RNF186 That Are Associated with Ulcerative Colitis. PLoS Genetics, 2013, 9, e1003723.	1.5	185

#	Article	IF	Citations
217	Evaluation of Common Variants in the Six Known Maturity-Onset Diabetes of the Young (MODY) Genes for Association With Type 2 Diabetes. Diabetes, 2007, 56, 685-693.	0.3	178
218	Genomewide Linkage Analysis of Stature in Multiple Populations Reveals Several Regions with Evidence of Linkage to Adult Height. American Journal of Human Genetics, 2001, 69, 106-116.	2.6	177
219	Mismatch Repair Genes Mlh1 and Mlh3 Modify CAG Instability in Huntington's Disease Mice: Genome-Wide and Candidate Approaches. PLoS Genetics, 2013, 9, e1003930.	1.5	175
220	Association of Liver Injury From Specific Drugs, or Groups ofÂDrugs, With Polymorphisms in HLA and Other Genes in aÂGenome-Wide Association Study. Gastroenterology, 2017, 152, 1078-1089.	0.6	174
221	Comprehensive Association Testing of Common Mitochondrial DNA Variation in Metabolic Disease. American Journal of Human Genetics, 2006, 79, 54-61.	2.6	173
222	The functional spectrum of low-frequency coding variation. Genome Biology, 2011, 12, R84.	13.9	173
223	A genome-wide association study of shared risk across psychiatric disorders implicates gene regulation during fetal neurodevelopment. Nature Neuroscience, 2019, 22, 353-361.	7.1	173
224	A susceptibility locus for asthma-related traits on chromosome 7 revealed by genome-wide scan in a founder population. Nature Genetics, 2001, 28, 87-91.	9.4	168
225	Support for involvement of neuregulin 1 in schizophrenia pathophysiology. Molecular Psychiatry, 2005, 10, 366-374.	4.1	168
226	Genomewide Linkage Analysis of Bipolar Disorder by Use of a High-Density Single-Nucleotideâ€“Polymorphism (SNP) Genotyping Assay: A Comparison with Microsatellite Marker Assays and Finding of Significant Linkage to Chromosome 6q22. American Journal of Human Genetics, 2004, 74, 886-897.	2.6	167
227	Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. Nature Genetics, 2016, 48, 1107-1111.	9.4	167
228	Additional support for schizophrenia linkage on chromosomes 6 and 8: A multicenter study. , 1996, 67, 580-594.		166
229	Genome scan of schizophrenia. American Journal of Psychiatry, 1998, 155, 741-50.	4.0	166
230	Risk Models for Progression to Advanced Age-Related Macular Degeneration Using Demographic, Environmental, Genetic, and Ocular Factors. Ophthalmology, 2011, 118, 2203-2211.	2.5	161
231	A genome-wide scan for common variants affecting the rate of age-related cognitive decline. Neurobiology of Aging, 2012, 33, 1017.e1-1017.e15.	1.5	160
232	Integrating Autoimmune Risk Loci with Gene-Expression Data Identifies Specific Pathogenic Immune Cell Subsets. American Journal of Human Genetics, 2011, 89, 496-506.	2.6	159
233	Estimating the selective effects of heterozygous protein-truncating variants from human exome data. Nature Genetics, 2017, 49, 806-810.	9.4	157
234	A second major histocompatibility complex susceptibility locus for multiple sclerosis. Annals of Neurology, 2007, 61, 228-236.	2.8	156

#	Article	IF	Citations
235	Next-Generation Sequencing Strategies Enable Routine Detection of Balanced Chromosome Rearrangements for Clinical Diagnostics and Genetic Research. American Journal of Human Genetics, 2011, 88, 469-481.	2.6	154
236	The Autism Sequencing Consortium: Large-Scale, High-Throughput Sequencing in Autism Spectrum Disorders. Neuron, 2012, 76, 1052-1056.	3.8	153
237	Clozapine-induced agranulocytosis is associated with rare HLA-DQB1 and HLA-B alleles. Nature Communications, 2014, 5, 4757.	5.8	153
238	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	3.9	153
239	An Integrated Haplotype Map of the Human Major Histocompatibility Complex. American Journal of Human Genetics, 2003, 73, 580-590.	2.6	151
240	Case-Control Genome-Wide Association Study of Attention-Deficit/Hyperactivity Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2010, 49, 906-920.	0.3	150
241	Association Between Variants of PRDM1 and NDP52 and Crohn's Disease, Based on Exome Sequencing and Functional Studies. Gastroenterology, 2013, 145, 339-347.	0.6	149
242	Autism spectrum disorder and attention deficit hyperactivity disorder have a similar burden of rare protein-truncating variants. Nature Neuroscience, 2019, 22, 1961-1965.	7.1	148
243	IBD5 is a General Risk Factor for Inflammatory Bowel Disease: Replication of Association with Crohn Disease and Identification of a Novel Association with Ulcerative Colitis. American Journal of Human Genetics, 2003, 73, 205-211.	2.6	147
244	Deletion of TOP3Î², a component of FMRP-containing mRNPs, contributes to neurodevelopmental disorders. Nature Neuroscience, 2013, 16, 1228-1237.	7.1	144
245	Genetic variation in laboratory mice. Nature Genetics, 2005, 37, 1175-1180.	9.4	143
246	WHAP: haplotype-based association analysis. Bioinformatics, 2007, 23, 255-256.	1.8	143
247	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	13.7	142
248	Haplotype-based identification of a microsomal transfer protein marker associated with the human lifespan. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 14115-14120.	3.3	141
249	Guilt beyond a reasonable doubt. Nature Genetics, 2007, 39, 813-815.	9.4	140
250	Identification of Small Exonic CNV from Whole-Exome Sequence Data and Application to Autism Spectrum Disorder. American Journal of Human Genetics, 2013, 93, 607-619.	2.6	136
251	An Unexpectedly Complex Architecture for Skin Pigmentation in Africans. Cell, 2017, 171, 1340-1353.e14.	13.5	134
252	Phenome-wide association studies across large population cohorts support drug target validation. Nature Communications, 2018, 9, 4285.	5.8	134

#	Article	IF	Citations
253	Host genetic variation and its microbiome interactions within the Human Microbiome Project. Genome Medicine, 2018, 10, 6.	3.6	134
254	Accurately Assessing the Risk of Schizophrenia Conferred by Rare Copy-Number Variation Affecting Genes with Brain Function. PLoS Genetics, 2010, 6, e1001097.	1.5	134
255	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. PLoS Genetics, 2013, 9, e1003443.	1.5	133
256	Genomewide Search for Type 2 Diabetes Mellitus Susceptibility Loci in Finnish Families: The Botnia Study. American Journal of Human Genetics, 2002, 70, 509-516.	2.6	132
257	The Genome-wide Patterns of Variation Expose Significant Substructure in a Founder Population. American Journal of Human Genetics, 2008, 83, 787-794.	2.6	132
258	Exact Multipoint Quantitative-Trait Linkage Analysis in Pedigrees by Variance Components. American Journal of Human Genetics, 2000, 66, 1153-1157.	2.6	130
259	Efficient Multipoint Linkage Analysis through Reduction of Inheritance Space. American Journal of Human Genetics, 2001, 68, 963-977.	2.6	130
260	The role of the Toll receptor pathway in susceptibility to inflammatory bowel diseases. Genes and Immunity, 2007, 8, 387-397.	2.2	129
261	Association of Genetic Variants in NUDT15 With Thiopurine-Induced Myelosuppression in Patients With Inflammatory Bowel Disease. JAMA - Journal of the American Medical Association, 2019, 321, 773.	3.8	129
262	DGAT1 mutation is linked to a congenital diarrheal disorder. Journal of Clinical Investigation, 2012, 122, 4680-4684.	3.9	127
263	Genetic variants in the region harbouring IL2/IL21 associated with ulcerative colitis. Gut, 2009, 58, 799-804.	6.1	126
264	Autism spectrum disorder severity reflects the average contribution of de novo and familial influences. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 15161-15165.	3.3	125
265	Exome sequencing in schizophrenia-affected parentâ€“offspring trios reveals risk conferred by protein-coding de novo mutations. Nature Neuroscience, 2020, 23, 185-193.	7.1	125
266	Tractor uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. Nature Genetics, 2021, 53, 195-204.	9.4	125
267	Scalable generalized linear mixed model for region-based association tests in large biobanks and cohorts. Nature Genetics, 2020, 52, 634-639.	9.4	124
268	Genetic analysis of ozone-induced acute lung injury in sensitive and resistant strains of mice. Nature Genetics, 1997, 17, 471-474.	9.4	122
269	Variant TREM2 as Risk Factor for Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 182-184.	13.9	122
270	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	7.1	122

#	Article	IF	Citations
271	BCL11A deletions result in fetal hemoglobin persistence and neurodevelopmental alterations. Journal of Clinical Investigation, 2015, 125, 2363-2368.	3.9	122
272	Common SNPs in HMGCR in Micronesians and Whites Associated With LDL-Cholesterol Levels Affect Alternative Splicing of Exon13. Arteriosclerosis, Thrombosis, and Vascular Biology, 2008, 28, 2078-2084.	1.1	120
273	Genetic architecture of human plasma lipidome and its link to cardiovascular disease. Nature Communications, 2019, 10, 4329.	5.8	120
274	A molecular marker based linkage map ofVitis. Genome, 1995, 38, 786-794.	0.9	117
275	Familial Eosinophilia Maps to the Cytokine Gene Cluster on Human Chromosomal Region 5q31-q33. American Journal of Human Genetics, 1998, 63, 1086-1094.	2.6	117
276	Genetic and environmental risk factors in congenital heart disease functionally converge in protein networks driving heart development. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 14035-14040.	3.3	117
277	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	13.7	115
278	Gender Disparity and Mutation Burden in Metastatic Melanoma. Journal of the National Cancer Institute, 2015, 107, djv221.	3.0	114
279	Survey of variation in human transcription factors reveals prevalent DNA binding changes. Science, 2016, 351, 1450-1454.	6.0	114
280	Genetic investigation of chromosome 5q GABAA receptor subunit genes in schizophrenia. Molecular Psychiatry, 2005, 10, 1074-1088.	4.1	112
281	Analysis of High-Resolution HapMap of DTNBP1 (Dysbindin) Suggests No Consistency between Reported Common Variant Associations and Schizophrenia. American Journal of Human Genetics, 2006, 79, 903-909.	2.6	111
282	Serrate2 is disrupted in the mouse limb-development mutant syndactylism. Nature, 1997, 389, 722-725.	13.7	110
283	A Pleiotropic Missense Variant in SLC39A8 Is Associated With Crohnâ€™s Disease and Human Gut Microbiome Composition. Gastroenterology, 2016, 151, 724-732.	0.6	109
284	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	9.4	109
285	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. Nature Medicine, 2021, 27, 1012-1024.	15.2	109
286	Association of Variants in theLIPCandABCA1Genes with Intermediate and Large Drusen and Advanced Age-Related Macular Degeneration. , 2011, 52, 4663.		108
287	ASD and schizophrenia show distinct developmental profiles in common genetic overlap with population-based social communication difficulties. Molecular Psychiatry, 2018, 23, 263-270.	4.1	107
288	Common body mass index-associated variants confer risk of extreme obesity. Human Molecular Genetics, 2009, 18, 3502-3507.	1.4	106

#	Article	IF	Citations
289	Genetic Coding Variant in GPR65 Alters Lysosomal pH and Links Lysosomal Dysfunction with Colitis Risk. Immunity, 2016, 44, 1392-1405.	6.6	106
290	Genetic influences on eight psychiatric disorders based on family data of 4 408 646 full and half-siblings, and genetic data of 333 748 cases and controls. Psychological Medicine, 2019, 49, 1166-1173.	2.7	106
291	Genome-wide scan in Portuguese Island families identifies 5q31â€“5q35 as a susceptibility locus for schizophrenia and psychosis. Molecular Psychiatry, 2004, 9, 213-218.	4.1	105
292	Genomeâ€wide association scan of the time to onset of attention deficit hyperactivity disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1355-1358.	1.1	103
293	Prospective Assessment of Genetic Effects on Progression to Different Stages of Age-Related Macular Degeneration Using Multistate Markov Models. , 2012, 53, 1548.		102
294	Genetic Differences in the Immediate Transcriptome Response to Stress Predict Risk-Related Brain Function and Psychiatric Disorders. Neuron, 2015, 86, 1189-1202.	3.8	102
295	Ubiquitin Ligase TRIM62 Regulates CARD9-Mediated Anti-fungal Immunity and Intestinal Inflammation. Immunity, 2015, 43, 715-726.	6.6	102
296	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. American Journal of Human Genetics, 2018, 102, 1204-1211.	2.6	102
297	Prevalence of rearrangements in the 22q11.2 region and population-based risk of neuropsychiatric and developmental disorders in a Danish population: a case-cohort study. Lancet Psychiatry,the, 2018, 5, 573-580.	3.7	102
298	Autosomal monoallelic expression in the mouse. Genome Biology, 2012, 13, R10.	13.9	101
299	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. Nature Neuroscience, 2019, 22, 1966-1974.	7.1	101
300	Inherited myeloproliferative neoplasm risk affects haematopoietic stem cells. Nature, 2020, 586, 769-775.	13.7	101
301	The role of polygenic risk and susceptibility genes in breast cancer over the course of life. Nature Communications, 2020, 11, 6383.	5.8	101
302	A missense variant in Mitochondrial Amidoxime Reducing Component 1 gene and protection against liver disease. PLoS Genetics, 2020, 16, e1008629.	1.5	101
303	Rare genetic variants in the CFI gene are associated with advanced age-related macular degeneration and commonly result in reduced serum factor I levels. Human Molecular Genetics, 2015, 24, 3861-70.	1.4	100
304	Exome Sequencing Analysis Reveals Variants in Primary Immunodeficiency Genes in Patients With Very Early Onset Inflammatory Bowel Disease. Gastroenterology, 2015, 149, 1415-1424.	0.6	99
305	A Homozygous Mutation in KCTD7 Links Neuronal Ceroid Lipofuscinosis to the Ubiquitin-Proteasome System. American Journal of Human Genetics, 2012, 91, 202-208.	2.6	97
306	Exploring the genetics of irritable bowel syndrome: a GWA study in the general population and replication in multinational case-control cohorts. Gut, 2015, 64, 1774-1782.	6.1	97

#	Article	IF	Citations
307	Acheiropodia Is Caused by a Genomic Deletion in C7orf2, the Human Orthologue of the Lmbr1 Gene. American Journal of Human Genetics, 2001, 68, 38-45.	2.6	95
308	Genetic Variation in Myosin IXB Is Associated With Ulcerative Colitis. Gastroenterology, 2006, 131, 1768-1774.	0.6	95
309	Fine Mapping in 94 Inbred Mouse Strains Using a High-Density Haplotype Resource. Genetics, 2010, 185, 1081-1095.	1.2	95
310	Whole-exome sequencing identifies rare, functional CFH variants in families with macular degeneration. Human Molecular Genetics, 2014, 23, 5283-5293.	1.4	95
311	Medical relevance of protein-truncating variants across 337,205 individuals in the UK Biobank study. Nature Communications, 2018, 9, 1612.	5.8	95
312	C1orf106 is a colitis risk gene that regulates stability of epithelial adherens junctions. Science, 2018, 359, 1161-1166.	6.0	95
313	Genome-wide association study implicates CHRNA2 in cannabis use disorder. Nature Neuroscience, 2019, 22, 1066-1074.	7.1	94
314	Bacterial, Fungal, and Viral Disease Resistance Loci Mapped in a Recombinant Inbred Common Bean Population (`Dorado'/XAN 176). Journal of the American Society for Horticultural Science, 2000, 125, 476-481.	0.5	92
315	Ca 2+ /Calmodulin-Dependent Protein Kinase II Is a Modulator of CARMA1-Mediated NF-ÎºB Activation. Molecular and Cellular Biology, 2006, 26, 5497-5508.	1.1	91
316	A high-density association screen of 155 ion transport genes for involvement with common migraine. Human Molecular Genetics, 2008, 17, 3318-3331.	1.4	90
317	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. Nature Neuroscience, 2016, 19, 1563-1565.	7.1	90
318	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	3.7	90
319	Geographic Variation and Bias in the Polygenic Scores of Complex Diseases and Traits in Finland. American Journal of Human Genetics, 2019, 104, 1169-1181.	2.6	90
320	Interchromosomal recombination in the extremely radioresistant bacterium Deinococcus radiodurans. Journal of Bacteriology, 1995, 177, 5495-5505.	1.0	89
321	Genome-Wide Association Studies in an Isolated Founder Population from the Pacific Island of Kosrae. PLoS Genetics, 2009, 5, e1000365.	1.5	89
322	Fine-Scale Genetic Structure in Finland. G3: Genes, Genomes, Genetics, 2017, 7, 3459-3468.	0.8	86
323	GWAS of thyroid stimulating hormone highlights pleiotropic effects and inverse association with thyroid cancer. Nature Communications, 2020, 11, 3981.	5.8	86
324	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. PLoS Genetics, 2010, 6, e1001183.	1.5	85

#	Article	IF	Citations
325	Germline Mutations Affecting GÎ±₁₁ in Hypoparathyroidism. New England Journal of Medicine, 2013, 368, 2532-2534.	13.9	85
326	Evidence for linkage of familial Diamond-Blackfan anemia to chromosome 8p23.3-p22 and for non-19q non-8p disease. Blood, 2001, 97, 2145-2150.	0.6	84
327	Lack of Association Between the Trp719Arg Polymorphism in Kinesin-Like Protein-6 and Coronary Artery Disease in 19 Case-Control Studies. Journal of the American College of Cardiology, 2010, 56, 1552-1563.	1.2	84
328	Interpreting de novo Variation in Human Disease Using denovolyzeR. Current Protocols in Human Genetics, 2015, 87, 7.25.1-7.25.15.	3.5	84
329	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	5.8	84
330	A model for repair of radiation-induced DNA double-strand breaks in the extreme radiophileDeinococcus radiodurans. BioEssays, 1995, 17, 457-464.	1.2	83
331	Genetic Control of Serum IgE Levels and Asthma: Linkage and Linkage Disequilibrium Studies in an Isolated Population. Human Molecular Genetics, 1997, 6, 2069-2076.	1.4	83
332	Candidate Gene Association Study for Diabetic Retinopathy in Persons with Type 2 Diabetes: The Candidate Gene Association Resource (CARe). , 2011, 52, 7593.		82
333	Comparing strategies to fine-map the association of common SNPs at chromosome 9p21 with type 2 diabetes and myocardial infarction. Nature Genetics, 2011, 43, 801-805.	9.4	79
334	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. Nature Communications, 2018, 9, 2606.	5.8	79
335	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	15.2	79
336	Association Testing in 9,000 People Fails to Confirm the Association of the Insulin Receptor Substrate-1 G972R Polymorphism With Type 2 Diabetes. Diabetes, 2004, 53, 3313-3318.	0.3	78
337	Segmental Phylogenetic Relationships of Inbred Mouse Strains Revealed by Fine-Scale Analysis of Sequence Variation Across 4.6 Mb of Mouse Genome. Genome Research, 2004, 14, 1493-1500.	2.4	78
338	Refined genomic localization and ethnic differences observed for the IBD5 association with Crohn's disease. European Journal of Human Genetics, 2007, 15, 328-335.	1.4	78
339	Gene-centric association mapping of chromosome 3p implicates MST1 in IBD pathogenesis. Mucosal Immunology, 2008, 1, 131-138.	2.7	77
340	Common Variants in the ENPP1 Gene Are Not Reproducibly Associated With Diabetes or Obesity. Diabetes, 2006, 55, 3180-3184.	0.3	76
341	Heritability of the Weight Loss Response to Gastric Bypass Surgery. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1630-E1633.	1.8	76
342	Association Testing of Variants in the Hepatocyte Nuclear Factor 4Â Gene With Risk of Type 2 Diabetes in 7,883 People. Diabetes, 2005, 54, 886-892.	0.3	75

#	Article	IF	Citations
343	Genome-wide common and rare variant analysis provides novel insights into clozapine-associated neutropenia. Molecular Psychiatry, 2017, 22, 1502-1508.	4.1	75
344	Trans-biobank analysis with 676,000 individuals elucidates the association of polygenic risk scores of complex traits with human lifespan. Nature Medicine, 2020, 26, 542-548.	15.2	74
345	Association of Common Variation in the HNF1Â Gene Region With Risk of Type 2 Diabetes. Diabetes, 2005, 54, 2336-2342.	0.3	73
346	Heritability and Genome-Wide Association Study to Assess Genetic Differences between Advanced Age-related Macular Degeneration Subtypes. Ophthalmology, 2012, 119, 1874-1885.	2.5	73
347	ARMS2/HTRA1 Locus Can Confer Differential Susceptibility to the Advanced Subtypes of Age-Related Macular Degeneration. American Journal of Ophthalmology, 2011, 151, 345-352.e3.	1.7	71
348	Association of DLG5 R30Q variant with inflammatory bowel disease. European Journal of Human Genetics, 2005, 13, 835-839.	1.4	70
349	Annotation of loci from genome-wide association studies using tissue-specific quantitative interaction proteomics. Nature Methods, 2014, 11, 868-874.	9.0	70
350	Comprehensive characterization of amino acid positions in protein structures reveals molecular effect of missense variants. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 28201-28211.	3.3	68
351	Genetic association of Alzheimer's disease with multiple polymorphisms in alpha-2-macroglobulin. Human Molecular Genetics, 2003, 12, 2765-2776.	1.4	67
352	Genetic associations of protein-coding variants in human disease. Nature, 2022, 603, 95-102.	13.7	67
353	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	1.5	66
354	Enhancing linkage analysis of complex disorders: an evaluation of high-density genotyping. Human Molecular Genetics, 2004, 13, 1943-1949.	1.4	65
355	Weight Loss after Gastric Bypass Is Associated with a Variant at 15q26.1. American Journal of Human Genetics, 2013, 92, 827-834.	2.6	65
356	Phenotypic extremes in rare variant study designs. European Journal of Human Genetics, 2016, 24, 924-930.	1.4	65
357	Genome-wide association study of electrocardiographic conduction measures in an isolated founder population: Kosrae. Heart Rhythm, 2009, 6, 634-641.	0.3	64
358	The human genetic epidemiology of COVID-19. Nature Reviews Genetics, 2022, 23, 533-546.	7.7	64
359	Absence of linkage between inflammatory bowel disease and selected loci on chromosomes 3, 7, 12, and 16. Gastroenterology, 1998, 115, 1062-1065.	0.6	63
360	Association between polygenic risk for schizophrenia, neurocognition and social cognition across development. Translational Psychiatry, 2016, 6, e924-e924.	2.4	63

#	Article	IF	Citations
361	Clinical and Genomic Correlates of Neutrophil Reactive Oxygen Species Production in Pediatric Patients With Crohnâ€™s Disease. Gastroenterology, 2018, 154, 2097-2110.	0.6	63
362	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. Neuron, 2018, 98, 743-753.e4.	3.8	63
363	Paternal-age-related de novo mutations and risk for five disorders. Nature Communications, 2019, 10, 3043.	5.8	63
364	Association analysis of NOTCH4 loci in schizophrenia using family and population-based controls. Nature Genetics, 2001, 28, 126-128.	9.4	62
365	Evaluating potential for whole-genome studies in Kosrae, an isolated population in Micronesia. Nature Genetics, 2006, 38, 214-217.	9.4	61
366	Evidence of transmission ratio distortion of DLG5 R30Q variant in general and implication of an association with Crohn disease in men. Human Genetics, 2006, 119, 305-311.	1.8	61
367	Whole-genome view of the consequences of a population bottleneck using 2926 genome sequences from Finland and United Kingdom. European Journal of Human Genetics, 2017, 25, 477-484.	1.4	60
368	Parental Phenotypes in Family-Based Association Analysis. American Journal of Human Genetics, 2005, 76, 249-259.	2.6	59
369	Haplotype Sharing Provides Insights into Fine-Scale Population History and Disease in Finland. American Journal of Human Genetics, 2018, 102, 760-775.	2.6	57
370	The critical needs and challenges for genetic architecture studies in Africa. Current Opinion in Genetics and Development, 2018, 53, 113-120.	1.5	57
371	Using a Genome-Wide Scan and Meta-analysis to Identify a Novel IBD Locus and Confirm Previously Identified IBD Loci. Inflammatory Bowel Diseases, 2002, 8, 375-381.	0.9	56
372	Selective Modulation of Autophagy, Innate Immunity, and Adaptive Immunity by Small Molecules. ACS Chemical Biology, 2013, 8, 2724-2733.	1.6	56
373	Germline Lysine-Specific Demethylase 1 (LSD1/KDM1A) Mutations Confer Susceptibility to Multiple Myeloma. Cancer Research, 2018, 78, 2747-2759.	0.4	56
374	Identification of Novel Genes That Mediate Innate Immunity Using Inbred Mice. Genetics, 2009, 183, 1535-1544.	1.2	55
375	Associations of CFHR1â€“CFHR3 deletion and a CFH SNP to age-related macular degeneration are not independent. Nature Genetics, 2010, 42, 553-555.	9.4	55
376	An Excess of Risk-Increasing Low-Frequency Variants Can Be a Signal of Polygenic Inheritance in Complex Diseases. American Journal of Human Genetics, 2014, 94, 437-452.	2.6	55
377	Glycerol as a Correlate of Impaired Glucose Tolerance: Dissection of a Complex System by Use of a Simple Genetic Trait. American Journal of Human Genetics, 2000, 66, 1558-1568.	2.6	53
378	Diagnostic misclassification reduces the ability to detect linkage in inflammatory bowel disease genetic studies. Gut, 2001, 49, 773-776.	6.1	53

#	Article	IF	Citations
379	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	2.6	53
380	New data and an old puzzle: the negative association between schizophrenia and rheumatoid arthritis. International Journal of Epidemiology, 2015, 44, 1706-1721.	0.9	53
381	Founder population-specific HapMap panel increases power in GWA studies through improved imputation accuracy and CNV tagging. Genome Research, 2010, 20, 1344-1351.	2.4	52
382	Shared Genetic Risk Factors Across Carbamazepineâ€Induced Hypersensitivity Reactions. Clinical Pharmacology and Therapeutics, 2019, 106, 1028-1036.	2.3	52
383	A Novel Hybrid Yeast-Human Network Analysis Reveals an Essential Role for FNBP1L in Antibacterial Autophagy. Journal of Immunology, 2009, 182, 4917-4930.	0.4	51
384	Mutations ineIF4ENIF1Are Associated With Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1534-E1539.	1.8	51
385	A Frameshift in CSF2RB Predominant Among Ashkenazi Jews Increases Risk for Crohn's Disease and Reduces Monocyte Signaling via GM-CSF. Gastroenterology, 2016, 151, 710-723.e2.	0.6	51
386	A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis. Nature Communications, 2016, 7, 12342.	5.8	50
387	Association Testing of the Protein Tyrosine Phosphatase 1B Gene (PTPN1) With Type 2 Diabetes in 7,883 People. Diabetes, 2005, 54, 1884-1891.	0.3	49
388	Burden of unique and low prevalence somatic mutations correlates with cancer survival. Scientific Reports, 2019, 9, 4848.	1.6	49
389	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. Nature Communications, 2020, 11, 3368.	5.8	49
390	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. American Journal of Human Genetics, 2021, 108, 656-668.	2.6	49
391	Investigating Shared Genetic Basis Across Tourette Syndrome and Comorbid Neurodevelopmental Disorders Along the Impulsivity-Compulsivity Spectrum. Biological Psychiatry, 2021, 90, 317-327.	0.7	49
392	Genetic architecture of tuberculosis resistance in a mouse model of infection. Genes and Immunity, 2006, 7, 201-210.	2.2	48
393	Mapping rare, deleterious mutations in Factor H: Association with early onset, drusen burden and lower antigenic levels in familial AMD. Scientific Reports, 2016, 6, 31531.	1.6	48
394	Direct or indirect association in a complex disease: the role ofSLC22A4 andSLC22A5 functional variants in Crohn disease. Human Mutation, 2006, 27, 778-785.	1.1	47
395	No Evidence for Association of Autism with Rare Heterozygous Point Mutations in Contactin-Associated Protein-Like 2 (CNTNAP2), or in Other Contactin-Associated Proteins or Contactins. PLoS Genetics, 2015, 11, e1004852.	1.5	47
396	A Low-Frequency Inactivating AKT2 Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. Diabetes, 2017, 66, 2019-2032.	0.3	47

#	Article	IF	Citations
397	Identification of pathogenic variant enriched regions across genes and gene families. Genome Research, 2020, 30, 62-71.	2.4	47
398	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
399	SMAD3 gene variant is a risk factor for recurrent surgery in patients with Crohn's disease. Journal of Crohn's and Colitis, 2014, 8, 845-851.	0.6	46
400	The genetic architecture of pediatric cognitive abilities in the Philadelphia Neurodevelopmental Cohort. Molecular Psychiatry, 2015, 20, 454-458.	4.1	46
401	A large-scale genomic investigation of susceptibility to infection and its association with mental disorders in the Danish population. Translational Psychiatry, 2019, 9, 283.	2.4	46
402	Glycobiology and schizophrenia: a biological hypothesis emerging from genomic research. Molecular Psychiatry, 2020, 25, 3129-3139.	4.1	46
403	A molecular-properties-based approach to understanding PDZ domain proteins and PDZ ligands. Genome Research, 2006, 16, 1056-1072.	2.4	45
404	Common genetic variants differentially influence the transition from clinically defined states of fasting glucose metabolism. Diabetologia, 2012, 55, 331-339.	2.9	45
405	Exome Sequencing in Suspected Monogenic Dyslipidemias. Circulation: Cardiovascular Genetics, 2015, 8, 343-350.	5.1	45
406	Integrated Genomics of Crohnâ€™s Disease Risk Variant Identifies a Role for CLEC12A in Antibacterial Autophagy. Cell Reports, 2015, 11, 1905-1918.	2.9	45
407	Small-molecule inhibitors directly target CARD9 and mimic its protective variant in inflammatory bowel disease. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 11392-11397.	3.3	45
408	Targeted gene sequencing in 6994 individuals with neurodevelopmental disorder with epilepsy. Genetics in Medicine, 2019, 21, 2496-2503.	1.1	45
409	Altered Intestinal ACE2 Levels Are Associated With Inflammation, Severe Disease, and Response to Anti-Cytokine Therapy in Inflammatory Bowel Disease. Gastroenterology, 2021, 160, 809-822.e7.	0.6	45
410	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
411	Linkage Thresholds for Two-stage Genome Scans. American Journal of Human Genetics, 1998, 62, 994-995.	2.6	43
412	Biases and Reconciliation in Estimates of Linkage Disequilibrium in the Human Genome. American Journal of Human Genetics, 2006, 78, 588-603.	2.6	43
413	The schizophrenia risk locus in SLC39A8 alters brain metal transport and plasma glycosylation. Scientific Reports, 2020, 10, 13162.	1.6	43
414	Common and Rare Variant Prediction and Penetrance of IBD in a Large, Multi-ethnic, Health System-based Biobank Cohort. Gastroenterology, 2021, 160, 1546-1557.	0.6	43

#	Article	IF	Citations
415	Genome-wide scan in Portuguese Island families implicates multiple loci in bipolar disorder: Fine mapping adds support on chromosomes 6 and 11. American Journal of Medical Genetics Part A, 2004, 127B, 30-34.	2.4	42
416	TMEM258 Is a Component of the Oligosaccharyltransferase Complex Controlling ER Stress and Intestinal Inflammation. Cell Reports, 2016, 17, 2955-2965.	2.9	42
417	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	3.6	42
418	Established genetic risk factors do not distinguish early and later onset CrohnÊ¼s disease. Inflammatory Bowel Diseases, 2009, 15, 1508-1514.	0.9	41
419	A Recurrent ERCC3 Truncating Mutation Confers Moderate Risk for Breast Cancer. Cancer Discovery, 2016, 6, 1267-1275.	7.7	41
420	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. Genome Research, 2018, 28, 968-974.	2.4	41
421	A role for genetic susceptibility in sporadic focal segmental glomerulosclerosis. Journal of Clinical Investigation, 2016, 126, 1067-1078.	3.9	41
422	Consistently Replicating Locus Linked to Migraine on 10q22-q23. American Journal of Human Genetics, 2008, 82, 1051-1063.	2.6	40
423	A Genomewide Linkage-Disequilibrium Scan Localizes the Saguenayâ€“Lac-Saint-Jean Cytochrome Oxidase Deficiency to 2p16. American Journal of Human Genetics, 2001, 68, 397-409.	2.6	39
424	Population Histories of the United States Revealed through Fine-Scale Migration and Haplotype Analysis. American Journal of Human Genetics, 2020, 106, 371-388.	2.6	39
425	Mutations in ATP13A2 (PARK9) are associated with an amyotrophic lateral sclerosis-like phenotype, implicating this locus in further phenotypic expansion. Human Genomics, 2019, 13, 19.	1.4	38
426	Immunity and mental illness: findings from a Danish population-based immunogenetic study of seven psychiatric and neurodevelopmental disorders. European Journal of Human Genetics, 2019, 27, 1445-1455.	1.4	38
427	High-Quality Exome Sequencing of Whole-Genome Amplified Neonatal Dried Blood Spot DNA. PLoS ONE, 2016, 11, e0153253.	1.1	38
428	Protective Low-Frequency Variants for Preeclampsia in the Fms Related Tyrosine Kinase 1 Gene in the Finnish Population. Hypertension, 2017, 70, 365-371.	1.3	37
429	Principles and methods of in-silico prioritization of non-coding regulatory variants. Human Genetics, 2018, 137, 15-30.	1.8	37
430	MAST3: a novel IBD risk factor that modulates TLR4 signaling. Genes and Immunity, 2008, 9, 602-612.	2.2	35
431	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. Nature Communications, 2016, 7, 13293.	5.8	35
432	Two Quantitative Trait Loci for Prepulse Inhibition of Startle Identified on Mouse Chromosome 16 Using Chromosome Substitution Strains. Genetics, 2005, 171, 1895-1904.	1.2	34

#	Article	IF	Citations
433	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. International Journal of Epidemiology, 2020, 49, 1022-1031.	0.9	34
434	Allele-Specific Methylation Occurs at Genetic Variants Associated with Complex Disease. PLoS ONE, 2014, 9, e98464.	1.1	33
435	A missense variant in SLC39A8 confers risk for Crohnâ€™s disease by disrupting manganese homeostasis and intestinal barrier integrity. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 28930-28938.	3.3	33
436	Identification of EFHC2 as a quantitative trait locus for fear recognition in Turner syndrome. Human Molecular Genetics, 2007, 16, 107-113.	1.4	32
437	Rare Variant, Gene-Based Association Study of Hereditary Melanoma Using Whole-Exome Sequencing. Journal of the National Cancer Institute, 2017, 109, .	3.0	32
438	Contribution of rare and common variants to intellectual disability in a sub-isolate of Northern Finland. Nature Communications, 2019, 10, 410.	5.8	32
439	Molecular genetic overlap between posttraumatic stress disorder and sleep phenotypes. Sleep, 2020, 43, .	0.6	32
440	Genetic resistance to diet-induced obesity in chromosome substitution strains of mice. Mammalian Genome, 2010, 21, 115-129.	1.0	31
441	Genome-wide association study of serious blistering skin rash caused by drugs. Pharmacogenomics Journal, 2012, 12, 96-104.	0.9	31
442	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179.	2.4	31
443	Rare protein-altering variants in ANGPTL7 lower intraocular pressure and protect against glaucoma. PLoS Genetics, 2020, 16, e1008682.	1.5	31
444	Genomewide search and association studies in a Finnish celiac disease population: Identification of a novel locus and replication of the HLA and CTLA4 loci. , 2004, 130A, 345-350.		30
445	Enhanced epigenetic profiling of classical human monocytes reveals a specific signature of healthy aging in the DNA methylome. Nature Aging, 2021, 1, 124-141.	5.3	30
446	Characterization of candidate genes in inflammatory bowel diseaseâ€“associated risk loci. JCI Insight, 2016, 1, e87899.	2.3	30
447	The female protective effect against autism spectrum disorder. Cell Genomics, 2022, 2, 100134.	3.0	30
448	Multiple genetic loci modify susceptibility to plasmacytoma-related morbidity in EÂ-v-abl transgenic mice. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 11299-11304.	3.3	29
449	A Novel Test for Recessive Contributions to Complex Diseases Implicates Bardet-Biedl Syndrome Gene BBS10 in Idiopathic Type 2 Diabetes and Obesity. American Journal of Human Genetics, 2014, 95, 509-520.	2.6	29
450	A framework for the detection of de novo mutations in family-based sequencing data. European Journal of Human Genetics, 2017, 25, 227-233.	1.4	29

#	Article	IF	Citations
451	High heritability of ascending aortic diameter and trans-ancestry prediction of thoracic aortic disease. Nature Genetics, 2022, 54, 772-782.	9.4	29
452	Haplotype Structures and Large-Scale Association Testing of the 5' AMP-Activated Protein Kinase Genes PRKAA2, PRKAB1, and PRKAB2 With Type 2 Diabetes. Diabetes, 2006, 55, 849-855.	0.3	28
453	What have we learned from six years of GWAS in autoimmune diseases, and what is next?. Current Opinion in Immunology, 2012, 24, 571-575.	2.4	28
454	ANGPTL8 protein-truncating variant associated with lower serum triglycerides and risk of coronary disease. PLoS Genetics, 2021, 17, e1009501.	1.5	28
455	Genetic modifiers of hypertension in soluble guanylate cyclase Î±1â€“deficient mice. Journal of Clinical Investigation, 2012, 122, 2316-2325.	3.9	28
456	Genetic profile for five common variants associated with age-related macular degeneration in densely affected families: a novel analytic approach. European Journal of Human Genetics, 2010, 18, 496-501.	1.4	27
457	Rare deleterious mutations of the gene EFR3A in autism spectrum disorders. Molecular Autism, 2014, 5, 31.	2.6	27
458	Androgen-sensitive hypertension associated with soluble guanylate cyclase-Î±₁ deficiency is mediated by 20-HETE. American Journal of Physiology - Heart and Circulatory Physiology, 2016, 310, H1790-H1800.	1.5	27
459	Applicability of the Mutationâ€“Selection Balance Model to Population Genetics of Heterozygous Protein-Truncating Variants in Humans. Molecular Biology and Evolution, 2019, 36, 1701-1710.	3.5	27
460	A survey of allelic imbalance in F1 mice. Genome Research, 2008, 18, 555-563.	2.4	26
461	Whole exome sequencing analyses reveal geneâ€“microbiota interactions in the context of IBD. Gut, 2021, 70, gutjnl-2019-319706.	6.1	26
462	Multigenic control of tuberculosis resistance: analysis of a QTL on mouse chromosome 7 and its synergism with sst1. Genes and Immunity, 2009, 10, 37-46.	2.2	25
463	Reply to "Statistical concerns about the GSEA procedure". Nature Genetics, 2004, 36, 663-663.	9.4	24
464	Haplotype-based association analysis of 56 functional candidate genes in the IBD6 locus on chromosome 19. European Journal of Human Genetics, 2006, 14, 780-790.	1.4	24
465	Increased power of mixed models facilitates association mapping of 10 loci for metabolic traits in an isolated population. Human Molecular Genetics, 2011, 20, 827-839.	1.4	24
466	Network Analysis of Genome-Wide Selective Constraint Reveals a Gene Network Active in Early Fetal Brain Intolerant of Mutation. PLoS Genetics, 2016, 12, e1006121.	1.5	24
467	Haplotype structure of TNFRSF5-TNFSF5 (CD40â€“CD40L) and association analysis in systemic lupus erythematosus. European Journal of Human Genetics, 2005, 13, 669-676.	1.4	23
468	Systematic haplotype analysis resolves a complex plasma plant sterol locus on the Micronesian Island of Kosrae. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 13886-13891.	3.3	23

#	Article	IF	Citations
469	Use of a genetic isolate to identify rare disease variants: C7 on 5p associated with MS. Human Molecular Genetics, 2009, 18, 1670-1683.	1.4	22
470	Genetic variants and pathways implicated in a pediatric inflammatory bowel disease cohort. Genes and Immunity, 2019, 20, 131-142.	2.2	22
471	The 22q11.2 region regulates presynaptic gene-products linked to schizophrenia. Nature Communications, 2022, 13, .	5.8	22
472	Partners in crime. Nature Genetics, 2005, 37, 337-338.	9.4	21
473	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. American Journal of Human Genetics, 2016, 98, 857-868.	2.6	21
474	Whole-genome sequencing of African Americans implicates differential genetic architecture in inflammatory bowel disease. American Journal of Human Genetics, 2021, 108, 431-445.	2.6	21
475	Enhancing Discovery of Genetic Variants for Posttraumatic Stress Disorder Through Integration of Quantitative Phenotypes and Trauma Exposure Information. Biological Psychiatry, 2022, 91, 626-636.	0.7	21
476	Genetic analysis of multiple sclerosis. Journal of Autoimmunity, 2003, 21, 111-116.	3.0	20
477	The importance of being independent: sib pair analysis in diabetes. Nature Genetics, 1996, 14, 131-132.	9.4	19
478	New Approaches to Gene Hunting in IBD. Inflammatory Bowel Diseases, 2004, 10, 312-317.	0.9	19
479	An expanded analysis framework for multivariate GWAS connects inflammatory biomarkers to functional variants and disease. European Journal of Human Genetics, 2021, 29, 309-324.	1.4	19
480	Prioritization of disease genes from GWAS using ensemble-based positive-unlabeled learning. European Journal of Human Genetics, 2021, 29, 1527-1535.	1.4	19
481	Estimating the Human Gene Count. Cell, 2002, 109, 283-284.	13.5	18
482	The value of gene-based selection of tag SNPs in genome-wide association studies. European Journal of Human Genetics, 2006, 14, 1209-1214.	1.4	18
483	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. Human Genetics, 2011, 130, 685-699.	1.8	18
484	Habitual sleep disturbances and migraine: a Mendelian randomization study. Annals of Clinical and Translational Neurology, 2020, 7, 2370-2380.	1.7	18
485	Chromosome Xq23 is associated with lower atherogenic lipid concentrations and favorable cardiometabolic indices. Nature Communications, 2021, 12, 2182.	5.8	17
486	The Kruppel-Like Factor 11 (KLF11) Q62R Polymorphism Is Not Associated With Type 2 Diabetes in 8,676 People. Diabetes, 2006, 55, 3620-3624.	0.3	16

#	Article	IF	Citations
487	Meta-Analysis of Genome-Wide Association Studies. Cold Spring Harbor Protocols, 2010, 2010, pdb.top81.	0.2	16
488	Lack of association of rare functional variants in TSC1/TSC2 genes with autism spectrum disorder. Molecular Autism, 2013, 4, 5.	2.6	16
489	Response to â€˜Predicting the diagnosis of autism spectrum disorder using gene pathway analysisâ€™. Molecular Psychiatry, 2014, 19, 860-861.	4.1	16
490	Protective coding variants inCFHandPELI3and a variant nearCTRB1are associated with age-related macular degeneration^{â€}. Human Molecular Genetics, 2016, 25, ddw336.	1.4	16
491	Inflammation status modulates the effect of host genetic variation on intestinal gene expression in inflammatory bowel disease. Nature Communications, 2021, 12, 1122.	5.8	16
492	Haplotype analysis of tumour necrosis factor receptor genes in 1p36: no evidence for association with systemic lupus erythematosus. European Journal of Human Genetics, 2006, 14, 69-78.	1.4	15
493	Ascertainment Through Family History of Disease Often Decreases the Power of Family-based Association Studies. Behavior Genetics, 2007, 37, 631-636.	1.4	15
494	Genome wide association study identifies variants in NBEA associated with migraine in bipolar disorder. Journal of Affective Disorders, 2015, 172, 453-461.	2.0	15
495	Genome-wide Analysis of Immune System Genes by Expressed Sequence Tag Profiling. Journal of Immunology, 2013, 190, 5578-5587.	0.4	14
496	(Epi)Genetic Analyses of Age-Related Macular Degeneration: Case-Control and Discordant Twin Studies. Human Heredity, 2014, 78, 59-72.	0.4	14
497	Late-Onset Crohnâ€™s Disease Is A Subgroup Distinct in Genetic and Behavioral Risk Factors With UC-Like Characteristics. Inflammatory Bowel Diseases, 2018, 24, 2413-2422.	0.9	14
498	MISCAST: MIssense variant to protein StruCture Analysis web SuiTe. Nucleic Acids Research, 2020, 48, W132-W139.	6.5	14
499	A large population-based investigation into the genetics of susceptibility to gastrointestinal infections and the link between gastrointestinal infections and mental illness. Human Genetics, 2020, 139, 593-604.	1.8	14
500	Functional screen of inflammatory bowel disease genes reveals key epithelial functions. Genome Medicine, 2021, 13, 181.	3.6	14
501	Whole exome sequencing reveals minimal differences between cell line and whole blood derived DNA. Genomics, 2013, 102, 270-277.	1.3	13
502	Association testing of common variants in the insulin receptor substrate-1 gene (IRS1) with type 2 diabetes. Diabetologia, 2007, 50, 1209-1217.	2.9	12
503	Assessment of genetic variant burden in epilepsy-associated brain lesions. European Journal of Human Genetics, 2019, 27, 1738-1744.	1.4	12
504	Neurological disorder-associated genetic variants in individuals with psychogenic nonepileptic seizures. Scientific Reports, 2020, 10, 15205.	1.6	12

#	Article	IF	Citations
505	Not All Autism Genes Are Created Equal: A Response to Myers etÂal.. American Journal of Human Genetics, 2020, 107, 1000-1003.	2.6	11
506	On the level: IRGM gene function is all about expression. Autophagy, 2009, 5, 96-99.	4.3	10
507	Wains: A Pattern-Seeking Artificial Life Species. Artificial Life, 2012, 18, 399-423.	1.0	10
508	Mosaic mutations in blood DNA sequence are associated with solid tumor cancers. Npj Genomic Medicine, 2017, 2, 22.	1.7	10
509	SLC39A8 missense variant is associated with Crohn's disease but does not have a major impact on gut microbiome composition in healthy subjects. PLoS ONE, 2019, 14, e0211328.	1.1	10
510	Serum Analyte Profiles Associated With Crohnâ€™s Disease and Disease Location. Inflammatory Bowel Diseases, 2022, 28, 9-20.	0.9	10
511	Comprehensive genome-wide association study of different forms of hernia identifies more than 80 associated loci. Nature Communications, 2022, 13, .	5.8	9
512	Diabetes, dependence, asymptotics, selection and significance. Nature Genetics, 1997, 17, 148-148.	9.4	8
513	Linkage disequilibrium and haplotype structure of five GABAA receptor subunit genes investigated for association with schizophrenia. Molecular Psychiatry, 2005, 10, 1057-1057.	4.1	8
514	Reply to Elson et al.. American Journal of Human Genetics, 2007, 80, 382-383.	2.6	8
515	Association of Exome Sequences With Cardiovascular Traits Among Blacks in the Jackson Heart Study. Circulation: Cardiovascular Genetics, 2016, 9, 368-374.	5.1	8
516	Guidelineâ€based and bioinformatic reassessment of lesionâ€associated gene and variant pathogenicity in focal human epilepsies. Epilepsia, 2018, 59, 2145-2152.	2.6	8
517	Genetic variants in cellular transport do not affect mesalamine response in ulcerative colitis. PLoS ONE, 2018, 13, e0192806.	1.1	8
518	Genetic and Transcriptomic Variation Linked to Neutrophil Granulocyteâ€“Macrophage Colony-Stimulating Factor Signaling in Pediatric Crohnâ€™s Disease. Inflammatory Bowel Diseases, 2019, 25, 547-560.	0.9	8
519	Polygenic burden has broader impact on health, cognition, and socioeconomic outcomes than most rare and high-risk copy number variants. Molecular Psychiatry, 2021, 26, 4884-4895.	4.1	8
520	Changes in the fine-scale genetic structure of Finland through the 20th century. PLoS Genetics, 2021, 17, e1009347.	1.5	8
521	Untangling the Genetics of a Complex Disease. JAMA - Journal of the American Medical Association, 1998, 280, 652.	3.8	7
522	Localization of an acromesomelic dysplasia on chromosome 9 by homozygosity mapping. Clinical Genetics, 2000, 57, 278-283.	1.0	7

#	Article	IF	Citations
523	IBD5 is associated with an extensive complicated Crohn's disease feature: implications from genotype-phenotype analysis. Gut, 2007, 56, 149-150.	6.1	7
524	Association of the MYOC p.(Gln368Ter) Variant With Glaucoma in a Finnish Population. JAMA Ophthalmology, 2021, 139, 762.	1.4	7
525	Elucidating the relationship between migraine risk and brain structure using genetic data. Brain, 2022, 145, 3214-3224.	3.7	7
526	Integrating Autoimmune Risk Loci with Gene-Expression Data Identifies Specific Pathogenic Immune Cell Subsets. American Journal of Human Genetics, 2011, 89, 682.	2.6	6
527	Fine-mapping of genetic loci driving spontaneous clearance of hepatitis C virus infection. Scientific Reports, 2017, 7, 15843.	1.6	6
528	Reply to â€˜Selective effects of heterozygous protein-truncating variantsâ€™. Nature Genetics, 2019, 51, 3-4.	9.4	6
529	Exome sequencing in patientâ€parent trios suggests new candidate genes for earlyâ€onset primary sclerosing cholangitis. Liver International, 2021, 41, 1044-1057.	1.9	6
530	The intersection of genetics and COVID-19 in 2021: preview of the 2021 Rodney Howell Symposium. Genetics in Medicine, 2021, 23, 1001-1003.	1.1	6
531	A novel variant in SMG9 causes intellectual disability, confirming a role for nonsense-mediated decay components in neurocognitive development. European Journal of Human Genetics, 2022, 30, 619-627.	1.4	6
532	Implementation of CYP2D6 copy-number imputation panel and frequency of key pharmacogenetic variants in Finnish individuals with a psychotic disorder. Pharmacogenomics Journal, 2022, 22, 166-172.	0.9	6
533	Variant Score Rankerâ€”a web application for intuitive missense variant prioritization. Bioinformatics, 2019, 35, 4478-4479.	1.8	5
534	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	2.6	5
535	No Bias in Linkage Analysis. American Journal of Human Genetics, 2004, 75, 722-723.	2.6	4
536	O-002â€ƒGenes in IBD-Associated Risk Loci Demonstrate Genotype-, Tissue-, and Inflammation-Specific Patterns of Expression in Terminal Ileum and Colon Mucosal Tissue. Inflammatory Bowel Diseases, 2016, 22, S1.	0.9	4
537	Host Genetic Variant in CXCL16 May Be Associated With Hepatitis B Virusâ€“Related Acute LiverÂFailure. Cellular and Molecular Gastroenterology and Hepatology, 2019, 7, 477-479.e4.	2.3	4
538	Caseâ€“control analysis identifies shared properties of rare germline variation in cancer predisposing genes. European Journal of Human Genetics, 2019, 27, 824-828.	1.4	4
539	Meta-Analysis of IBD Gut Samples Gene Expression Identifies Specific Markers of Ileal and Colonic Diseases. Inflammatory Bowel Diseases, 2022, 28, 775-782.	0.9	4
540	Common body mass index-associated variants confer risk of extreme obesity. Human Molecular Genetics, 2010, 19, 3690-3691.	1.4	3

#	Article	IF	Citations
541	Polygenic risk heterogeneity among focal epilepsies. Epilepsia, 2020, 61, e179-e185.	2.6	3
542	A data-driven medication score predicts 10-year mortality among aging adults. Scientific Reports, 2020, 10, 15760.	1.6	3
543	Genotype imputation and polygenic score estimation in northwestern Russian population. PLoS ONE, 2022, 17, e0269434.	1.1	3
544	Subpopulation difference scanning: a strategy for exclusion mapping of susceptibility genes. Journal of Medical Genetics, 2005, 43, 590-597.	1.5	2
545	Reply to Tenesa et al â€˜Association of DLG5 and inflammatory bowel disease across populationsâ€™. European Journal of Human Genetics, 2006, 14, 260-261.	1.4	2
546	P164 USING EXOME SEQUENCING TO EXPAND THE GENETIC ARCHITECTURE OF INFLAMMATORY BOWEL DISEASE. Gastroenterology, 2018, 154, S88.	0.6	2
547	2017 William Allan Award Introduction: KÃ¡ri Stefansson. American Journal of Human Genetics, 2018, 102, 350.	2.6	2
548	Reply to: On powerful GWAS in admixed populations. Nature Genetics, 2021, 53, 1634-1635.	9.4	2
549	Bayesian model comparison for rare-variant association studies. American Journal of Human Genetics, 2021, 108, 2354-2367.	2.6	2
550	MYO9B polymorphisms in multiple sclerosis. European Journal of Human Genetics, 2009, 17, 840-843.	1.4	1
551	LRRK2 is an Interferon-gamma Target Involved in Immune Responses. Clinical Immunology, 2010, 135, S56.	1.4	1
552	2014 Curt Stern Award: A Tryst with Genetics1. American Journal of Human Genetics, 2015, 96, 369-371.	2.6	1
553	Rare mutations in factor H predispose to severe preeclampsia. Molecular Immunology, 2018, 102, 184-185.	1.0	1
554	LARGE META-ANALYSIS OF SCANDINAVIAN EXOME SEQUENCING STUDIES OF SCHIZOPHRENIA. European Neuropsychopharmacology, 2019, 29, S813.	0.3	1
555	54EXOME SEQUENCING OF 23,851 CASES IMPLICATES NOVEL RISK GENES AND PROVIDES INSIGHTS INTO THE GENETIC ARCHITECTURE OF SCHIZOPHRENIA. European Neuropsychopharmacology, 2019, 29, S1098.	0.3	1
556	LIMITED CONTRIBUTION OF RARE, NONCODING VARIATION TO AUTISM SPECTRUM DISORDER FROM SEQUENCING OF 2,076 GENOMES IN QUARTET FAMILIES. European Neuropsychopharmacology, 2019, 29, S784-S785.	0.3	1
557	Genetic modifiers of hypertension in soluble guanylate cyclase Î±1â€“deficient mice. Journal of Clinical Investigation, 2012, 122, 3024-3024.	3.9	1
558	A genome-wide scan in a canadian inflammatory bowel disease (IBD) population reveals two novel susceptibility loci. Gastroenterology, 2000, 118, A708.	0.6	0

#	Article	IF	Citations
559	Effect of diagnostic misclassification on the ability to detect linkage in inflammatory bowel disease (IBD). Gastroenterology, 2000, 118, A337.	0.6	0
560	F.33. Genetic Variants that Control the Expression of MHC Genes Do Not Affect Susceptibility to Multiple Sclerosis. Clinical Immunology, 2008, 127, S53-S54.	1.4	0
561	The Use of Protein-protein Interaction in Loci Associated to Crohn's and Rheumatoid Arthritis Reveals Evidence of Risk Spread Across Functional Networks. Clinical Immunology, 2010, 135, S119.	1.4	0
562	The Utility of Genetics in Classifying Ulcerative Colitis and Crohn's Disease Patients. Gastroenterology, 2011, 140, S-268.	0.6	0
563	An accurate and reproducible discrimination tool for inflammatory bowel disease subtype using genetics, serologies, and smoking status. Inflammatory Bowel Diseases, 2011, 17, S1.	0.9	0
564	Reply to Cipriani et al. European Journal of Human Genetics, 2012, 20, 3-3.	1.4	0
565	Identification of renin signaling as a blood pressure modifying mechanism in soluble guanylate cyclase Î±1-deficient mice. Nitric Oxide - Biology and Chemistry, 2012, 27, S25-S26.	1.2	0
566	P-175â€ƒPleiotropic Effects of Novel Functional LRRK2 Variation on CrohnÊ¼s Disease and ParkinsonÊ¼s Disease Risk. Inflammatory Bowel Diseases, 2016, 22, S62-S63.	0.9	0
567	Su1855 Genetic Burden Analysis Identified Late-Onset Crohn's Disease(CD) As a Distinct Subgroup Characterized by Low Genetic Burden, UC-Like Serological Markers, Mild Clinical Phenotypes and High Proportion of Smoking Cessation at Diagnosis. Gastroenterology, 2016, 150, S571.	0.6	0
568	Natural Selection and Neuropsychiatric Disease. , 2016, , 51-61.		0
569	Insights into the Genetic Epidemiology of Crohn's and Rare Diseases in the Ashkenazi Jewish Population. Gastroenterology, 2017, 152, S985.	0.6	0
570	PGC3 AIM 6 - Large Scale Wgs of Multiply Affected Pedigrees. European Neuropsychopharmacology, 2017, 27, S414-S415.	0.3	0
571	85. SNP-Based Dissection of PTSD from Large-Scale Genome-Wide Association Studies (GWAS) across Military and Civilian Cohorts. Biological Psychiatry, 2017, 81, S35-S36.	0.7	0
572	Identification and Characterization of Variant Intolerant Sites across Human Protein 3-Dimensional Structures. Biophysical Journal, 2018, 114, 664a.	0.2	0
573	Differences in the commonly used genotype imputation algorithms and their imputation accuracy estimates. , 2018, , .		0
574	Pamela Sklar (1959â€“2017). Nature, 2018, 554, 32-32.	13.7	0
575	27 CLINICAL AND GENOMIC CORRELATES OF NEUTROPHIL GRANULOCYTE-MACROPHAGE COLONY STIMULATING FACTOR SIGNALING IN PEDIATRIC CROHN DISEASE. Gastroenterology, 2018, 154, S10-S11.	0.6	0
576	13. The Role of Common and Rare Variants in ADHD Risk and Genetic Overlap With Other Phenotypes. Biological Psychiatry, 2019, 85, S5-S6.	0.7	0

#	Article	IF	Citations
577	153. A Novel Framework for Well-Calibrated Analysis of Complex Traits in Admixed Individuals. Biological Psychiatry, 2019, 85, S63-S64.	0.7	0
578	Large-Scale Genetic Characterization of PTSD Across Ancestry, Gender and Trauma-Type. European Neuropsychopharmacology, 2019, 29, S749-S750.	0.3	0
579	2018 William Allan Award Introduction: Eric S. Lander. American Journal of Human Genetics, 2019, 104, 373-374.	2.6	0
580	Functional Interpretation of Single Amino Acid Substitutions in 1,330 Disease-Associated Genes. Biophysical Journal, 2019, 116, 420a-421a.	0.2	0
581	68THE ROLE OF DELETERIOUS ULTRA-RARE VARIANTS IN ADHD RISK. European Neuropsychopharmacology, 2019, 29, S1106.	0.3	0
582	HAPLOTYPE STRUCTURE AND ASSOCIATION OF COMMON VARIANTS IN THE GENE ENCODING THE ISLET ATP-SENSITIVE POTASSIUM CHANNEL WITH TYPE 2 DIABETES.. Journal of Investigative Medicine, 2004, 52, S384.	0.7	0
583	Abstract 3282: Determination of cancer susceptibility in probands with breast and ovarian cancer. , 2014, , .		0
584	An Architecture for Pattern Recognition and Decision-Making. Lecture Notes in Computer Science, 2016, , 22-33.	1.0	0
585	Rare protein-altering variants in ANGPTL7 lower intraocular pressure and protect against glaucoma. , 2020, 16, e1008682.		0
586	Rare protein-altering variants in ANGPTL7 lower intraocular pressure and protect against glaucoma. , 2020, 16, e1008682.		0
587	Rare protein-altering variants in ANGPTL7 lower intraocular pressure and protect against glaucoma. , 2020, 16, e1008682.		0
588	Rare protein-altering variants in ANGPTL7 lower intraocular pressure and protect against glaucoma. , 2020, 16, e1008682.		0
589	Case Report: Supernormal Vascular Aging in Leningrad Siege Survivors. Frontiers in Cardiovascular Medicine, 2022, 9, .	1.1	0