Gregory Cooper

List of Publications by Year
in descending order

86 papers	32,688 citations	61984 43 h-index	58581 82 g-index
101 all docs	101 docs citations	101 times ranked	50123 citing authors

#	Article	IF	Citations
1	A general framework for estimating the relative pathogenicity of human genetic variants. Nature Genetics, 2014, 46, 310-315.	21.4	5,167
2	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature, 2007, 447, 799-816.	27.8	4,709
3	CADD: predicting the deleteriousness of variants throughout the human genome. Nucleic Acids Research, 2019, 47, D886-D894.	14.5	2,360
4	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	27.8	1,943
5	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. Science, 2008, 320, 539-543.	12.6	1,654
6	Identifying a High Fraction of the Human Genome to be under Selective Constraint Using GERP++. PLoS Computational Biology, 2010, 6, e1001025.	3.2	1,443
7	Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol or triglycerides in humans. Nature Genetics, 2008, 40, 189-197.	21.4	1,286
8	Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. Nature Genetics, 2010, 42, 790-793.	21.4	1,238
9	Distribution and intensity of constraint in mammalian genomic sequence. Genome Research, 2005, 15, 901-913.	5.5	1,230
10	A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846.	21.4	1,141
11	Guidelines for investigating causality of sequence variants in human disease. Nature, 2014, 508, 469-476.	27.8	1,130
12	Mapping and sequencing of structural variation from eight human genomes. Nature, 2008, 453, 56-64.	27.8	983
13	LAGAN and Multi-LAGAN: Efficient Tools for Large-Scale Multiple Alignment of Genomic DNA. Genome Research, 2003, 13, 721-731.	5.5	960
14	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	21.4	539
15	Needles in stacks of needles: finding disease-causal variants in a wealth of genomic data. Nature Reviews Genetics, 2011, 12, 628-640.	16.3	531
16	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. American Journal of Human Genetics, 2009, 84, 148-161.	6.2	530
17	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
18	Massively parallel functional dissection of mammalian enhancers in vivo. Nature Biotechnology, 2012, 30, 265-270.	17.5	468

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19	A genome-wide scan for common genetic variants with a large influence on warfarin maintenance dose. Blood, 2008, 112, 1022-1027.	1.4	410
20	Distinct Properties of Cell-Type-Specific and Shared Transcription Factor Binding Sites. Molecular Cell, 2013, 52, 25-36.	9.7	283
21	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. Genome Research, 2017, 27, 38-52.	5.5	244
22	Recurrent reciprocal deletions and duplications of 16p13.11: the deletion is a risk factor for MR/MCA while the duplication may be a rare benign variant. Journal of Medical Genetics, 2009, 46, 223-232.	3.2	241
23	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. American Journal of Human Genetics, 2007, 81, 1057-1069.	6.2	222
24	Mutational and selective effects on copy-number variants in the human genome. Nature Genetics, 2007, 39, S22-S29.	21.4	221
25	Systematic assessment of copy number variant detection via genome-wide SNP genotyping. Nature Genetics, 2008, 40, 1199-1203.	21.4	198
26	Identification, Replication, and Functional Fine-Mapping of Expression Quantitative Trait Loci in Primary Human Liver Tissue. PLoS Genetics, 2011, 7, e1002078.	3.5	191
27	Genomic diagnosis for children with intellectual disability and/or developmental delay. Genome Medicine, 2017, 9, 43.	8.2	188
28	Analyses of deep mammalian sequence alignments and constraint predictions for 1% of the human genome. Genome Research, 2007, 17, 760-774.	5.5	184
29	Polymorphisms of the HNF1A Gene Encoding Hepatocyte Nuclear Factor-1Î± are Associated with C-Reactive Protein. American Journal of Human Genetics, 2008, 82, 1193-1201.	6.2	170
30	Single-nucleotide evolutionary constraint scores highlight disease-causing mutations. Nature Methods, 2010, 7, 250-251.	19.0	162
31	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
32	Characterization of Evolutionary Rates and Constraints in Three Mammalian Genomes. Genome Research, 2004, 14, 539-548.	5.5	125
33	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	1.9	125
34	A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. Genome Research, 2009, 19, 1579-1585.	5.5	118
35	Specifying and Sustaining Pigmentation Patterns in Domestic and Wild Cats. Science, 2012, 337, 1536-1541.	12.6	110
36	Quantitative Estimates of Sequence Divergence for Comparative Analyses of Mammalian Genomes. Genome Research, 2003, 13, 813-820.	5.5	106

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37	Qualifying the relationship between sequence conservation and molecular function. Genome Research, 2008, 18, 201-205.	5.5	91
38	Automated Whole-Genome Multiple Alignment of Rat, Mouse, and Human. Genome Research, 2004, 14, 685-692.	5.5	79
39	Evolutionary constraint facilitates interpretation of genetic variation in resequenced human genomes. Genome Research, 2010, 20, 301-310.	5.5	77
40	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	6.2	76
41	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	6.2	62
42	Genomic regulatory regions: insights from comparative sequence analysis. Current Opinion in Genetics and Development, 2003, 13, 604-610.	3.3	57
43	Closing gaps in the human genome with fosmid resources generated from multiple individuals. Nature Genetics, 2008, 40, 96-101.	21.4	50
44	Non-coding and Loss-of-Function Coding Variants in TET2 are Associated with Multiple Neurodegenerative Diseases. American Journal of Human Genetics, 2020, 106, 632-645.	6.2	50
45	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759.	2.8	47
46	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	3.8	46
47	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
48	TRADE-OFFS IN DETECTING EVOLUTIONARILY CONSTRAINED SEQUENCE BY COMPARATIVE GENOMICS. Annual Review of Genomics and Human Genetics, 2005, 6, 143-164.	6.2	41
49	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
50	Parlez-vous VUS?. Genome Research, 2015, 25, 1423-1426.	5.5	36
51	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. Neuron, 2018, 100, 1354-1368.e5.	8.1	35
52	Functional constraint and small insertions and deletions in the ENCODE regions of the human genome. Genome Biology, 2007, 8, R180.	9.6	32
53	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	7.6	31
54	Promoter-distal RNA polymerase II binding discriminates active from inactive CCAAT/ enhancer-binding protein beta binding sites. Genome Research, 2015, 25, 1791-1800.	5.5	30

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55	Systematic reanalysis of genomic data improves quality of variant interpretation. Clinical Genetics, 2018, 94, 174-178.	2.0	30
56	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	6.2	30
57	Mapping genome-wide transcription factor binding sites in frozen tissues. Epigenetics and Chromatin, 2013, 6, 30.	3.9	29
58	De novo FGF12 mutation in 2 patients with neonatal-onset epilepsy. Neurology: Genetics, 2016, 2, e120.	1.9	29
59	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	2.8	27
60	Genome sequencing for early-onset or atypical dementia: high diagnostic yield and frequent observation of multiple contributory alleles. Journal of Physical Education and Sports Management, 2019, 5, a003491.	1.2	25
61	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25
62	Genomic sequencing identifies secondary findings in a cohort of parent study participants. Genetics in Medicine, 2018, 20, 1635-1643.	2.4	24
63	A YWHAZ Variant Associated With Cardiofaciocutaneous Syndrome Activates the RAF-ERK Pathway. Frontiers in Physiology, 2019, 10, 388.	2.8	23
64	Trends in occupational lead exposure since the 1978 OSHA lead standard. American Journal of Industrial Medicine, 2004, 45, 558-572.	2.1	22
65	Genome sequencing as a first-line diagnostic test for hospitalized infants. Genetics in Medicine, 2022, 24, 851-861.	2.4	22
66	Long-read genome sequencing for the molecular diagnosis of neurodevelopmental disorders. Human Genetics and Genomics Advances, 2021, 2, 100023.	1.7	20
67	Accelerated exon evolution within primate segmental duplications. Genome Biology, 2013, 14, R9.	9.6	19
68	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 701-708.	6.2	19
69	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19
70	Aberrant regulation of a poison exon caused by a non-coding variant in a mouse model of Scn1a-associated epileptic encephalopathy. PLoS Genetics, 2021, 17, e1009195.	3.5	18
71	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. PLoS Genetics, 2018, 14, e1007671.	3.5	16
72	Detection of Copy Number Variation Using SNP Genotyping. Methods in Molecular Biology, 2011, 767, 243-252.	0.9	14

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73	Clinical utility of genomic sequencing. Current Opinion in Pediatrics, 2019, 31, 732-738.	2.0	14
74	ABC: software for interactive browsing of genomic multiple sequence alignment data. BMC Bioinformatics, 2004, 5, 192.	2.6	12
75	A genome-wide interactome of DNA-associated proteins in the human liver. Genome Research, 2017, 27, 1950-1960.	5.5	10
76	Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls. Genetics in Medicine, 2021, 23, 280-288.	2.4	9
77	Targeted interrogation of copy number variation using SCIMMkit. Bioinformatics, 2010, 26, 120-122.	4.1	7
78	Child Neurology: Spastic paraparesis and dystonia with a novel ADCY5 mutation. Neurology, 2019, 93, 510-514.	1.1	7
79	Mammalian Comparative Sequence Analysis of the Agrp Locus. PLoS ONE, 2007, 2, e702.	2.5	5
80	Evaluating the strength of genetic results: Risks and responsibilities. PLoS Genetics, 2019, 15, e1008437.	3.5	1
81	Fibulin-5 mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis. Neurology: Genetics, 2020, 6, e476.	1.9	0
82	How secondary findings are made. , 2020, , 59-75.		0
83	Title is missing!. , 2021, 17, e1009195.		0
84	Title is missing!. , 2021, 17, e1009195.		0
85	Title is missing!. , 2021, 17, e1009195.		0
86	Title is missing!. , 2021, 17, e1009195.		0