

# Gregory Cooper

## List of Publications by Year in descending order

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Version: 2024-02-01

86  
papers

32,688  
citations

61857

43  
h-index

58464

82  
g-index

101  
all docs

101  
docs citations

101  
times ranked

50123  
citing authors

#	ARTICLE	IF	CITATIONS
1	Genome sequencing as a first-line diagnostic test for hospitalized infants. <i>Genetics in Medicine</i> , 2022, 24, 851-861.	1.1	22
2	Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls. <i>Genetics in Medicine</i> , 2021, 23, 280-288.	1.1	9
3	Aberrant regulation of a poison exon caused by a non-coding variant in a mouse model of Scn1a-associated epileptic encephalopathy. <i>PLoS Genetics</i> , 2021, 17, e1009195.	1.5	18
4	Long-read genome sequencing for the molecular diagnosis of neurodevelopmental disorders. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100023.	1.0	20
5	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 857-873.	2.6	19
6	Title is missing!. , 2021, 17, e1009195.		0
7	Title is missing!. , 2021, 17, e1009195.		0
8	Title is missing!. , 2021, 17, e1009195.		0
9	Title is missing!. , 2021, 17, e1009195.		0
10	Fibulin-5 mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis. <i>Neurology: Genetics</i> , 2020, 6, e476.	0.9	0
11	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. <i>European Journal of Human Genetics</i> , 2020, 28, 1422-1431.	1.4	25
12	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. <i>European Journal of Human Genetics</i> , 2020, 28, 770-782.	1.4	27
13	How secondary findings are made. , 2020, , 59-75.		0
14	Non-coding and Loss-of-Function Coding Variants in TET2 are Associated with Multiple Neurodegenerative Diseases. <i>American Journal of Human Genetics</i> , 2020, 106, 632-645.	2.6	50
15	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. <i>Brain</i> , 2019, 142, 2617-2630.	3.7	31
16	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. <i>European Journal of Human Genetics</i> , 2019, 27, 747-759.	1.4	47
17	A YWHAZ Variant Associated With Cardiofaciocutaneous Syndrome Activates the RAF-ERK Pathway. <i>Frontiers in Physiology</i> , 2019, 10, 388.	1.3	23
18	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 104, 701-708.	2.6	19

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19	Child Neurology: Spastic paraparesis and dystonia with a novel ADCY5 mutation. <i>Neurology</i> , 2019, 93, 510-514.	1.5	7
20	Evaluating the strength of genetic results: Risks and responsibilities. <i>PLoS Genetics</i> , 2019, 15, e1008437.	1.5	1
21	Genome sequencing for early-onset or atypical dementia: high diagnostic yield and frequent observation of multiple contributory alleles. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003491.	0.5	25
22	Clinical utility of genomic sequencing. <i>Current Opinion in Pediatrics</i> , 2019, 31, 732-738.	1.0	14
23	CADD: predicting the deleteriousness of variants throughout the human genome. <i>Nucleic Acids Research</i> , 2019, 47, D886-D894.	6.5	2,360
24	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 104, 319-330.	2.6	30
25	Systematic reanalysis of genomic data improves quality of variant interpretation. <i>Clinical Genetics</i> , 2018, 94, 174-178.	1.0	30
26	Genomic sequencing identifies secondary findings in a cohort of parent study participants. <i>Genetics in Medicine</i> , 2018, 20, 1635-1643.	1.1	24
27	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. <i>Neuron</i> , 2018, 100, 1354-1368.e5.	3.8	35
28	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. <i>American Journal of Human Genetics</i> , 2018, 103, 1022-1029.	2.6	76
29	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. <i>PLoS Genetics</i> , 2018, 14, e1007671.	1.5	16
30	<i>NBEA</i>: Developmental disease gene with early generalized epilepsy phenotypes. <i>Annals of Neurology</i> , 2018, 84, 788-795.	2.8	44
31	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. <i>Human Genetics</i> , 2018, 137, 375-388.	1.8	46
32	Genomic diagnosis for children with intellectual disability and/or developmental delay. <i>Genome Medicine</i> , 2017, 9, 43.	3.6	188
33	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2017, 100, 117-127.	2.6	62
34	A genome-wide interactome of DNA-associated proteins in the human liver. <i>Genome Research</i> , 2017, 27, 1950-1960.	2.4	10
35	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. <i>Genome Research</i> , 2017, 27, 38-52.	2.4	244
36	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. <i>Human Mutation</i> , 2016, 37, 653-660.	1.1	40

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37	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. <i>American Journal of Human Genetics</i> , 2016, 98, 1051-1066.	2.6	137
38	De novo <i>FGF12</i> mutation in 2 patients with neonatal-onset epilepsy. <i>Neurology: Genetics</i> , 2016, 2, e120.	0.9	29
39	Germline and somatic mutations in the <i>MTOR</i> gene in focal cortical dysplasia and epilepsy. <i>Neurology: Genetics</i> , 2016, 2, e118.	0.9	125
40	Parlez-vous VUS?. <i>Genome Research</i> , 2015, 25, 1423-1426.	2.4	36
41	Promoter-distal RNA polymerase II binding discriminates active from inactive CCAAT/ enhancer-binding protein beta binding sites. <i>Genome Research</i> , 2015, 25, 1791-1800.	2.4	30
42	Guidelines for investigating causality of sequence variants in human disease. <i>Nature</i> , 2014, 508, 469-476.	13.7	1,130
43	A general framework for estimating the relative pathogenicity of human genetic variants. <i>Nature Genetics</i> , 2014, 46, 310-315.	9.4	5,167
44	Accelerated exon evolution within primate segmental duplications. <i>Genome Biology</i> , 2013, 14, R9.	13.9	19
45	Distinct Properties of Cell-Type-Specific and Shared Transcription Factor Binding Sites. <i>Molecular Cell</i> , 2013, 52, 25-36.	4.5	283
46	Mapping genome-wide transcription factor binding sites in frozen tissues. <i>Epigenetics and Chromatin</i> , 2013, 6, 30.	1.8	29
47	Specifying and Sustaining Pigmentation Patterns in Domestic and Wild Cats. <i>Science</i> , 2012, 337, 1536-1541.	6.0	110
48	Massively parallel functional dissection of mammalian enhancers in vivo. <i>Nature Biotechnology</i> , 2012, 30, 265-270.	9.4	468
49	A copy number variation morbidity map of developmental delay. <i>Nature Genetics</i> , 2011, 43, 838-846.	9.4	1,141
50	Detection of Copy Number Variation Using SNP Genotyping. <i>Methods in Molecular Biology</i> , 2011, 767, 243-252.	0.4	14
51	Needles in stacks of needles: finding disease-causal variants in a wealth of genomic data. <i>Nature Reviews Genetics</i> , 2011, 12, 628-640.	7.7	531
52	Identification, Replication, and Functional Fine-Mapping of Expression Quantitative Trait Loci in Primary Human Liver Tissue. <i>PLoS Genetics</i> , 2011, 7, e1002078.	1.5	191
53	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. <i>Nature Genetics</i> , 2010, 42, 203-209.	9.4	539
54	Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. <i>Nature Genetics</i> , 2010, 42, 790-793.	9.4	1,238

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55	Single-nucleotide evolutionary constraint scores highlight disease-causing mutations. <i>Nature Methods</i> , 2010, 7, 250-251.	9.0	162
56	Targeted interrogation of copy number variation using SCIMMkit. <i>Bioinformatics</i> , 2010, 26, 120-122.	1.8	7
57	Evolutionary constraint facilitates interpretation of genetic variation in resequenced human genomes. <i>Genome Research</i> , 2010, 20, 301-310.	2.4	77
58	Identifying a High Fraction of the Human Genome to be under Selective Constraint Using GERP++. <i>PLoS Computational Biology</i> , 2010, 6, e1001025.	1.5	1,443
59	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. <i>Journal of Medical Genetics</i> , 2009, 46, 223-232.	1.5	241
60	A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. <i>Genome Research</i> , 2009, 19, 1579-1585.	2.4	118
61	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. <i>American Journal of Human Genetics</i> , 2009, 84, 148-161.	2.6	530
62	Mapping and sequencing of structural variation from eight human genomes. <i>Nature</i> , 2008, 453, 56-64.	13.7	983
63	Closing gaps in the human genome with fosmid resources generated from multiple individuals. <i>Nature Genetics</i> , 2008, 40, 96-101.	9.4	50
64	Systematic assessment of copy number variant detection via genome-wide SNP genotyping. <i>Nature Genetics</i> , 2008, 40, 1199-1203.	9.4	198
65	Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol or triglycerides in humans. <i>Nature Genetics</i> , 2008, 40, 189-197.	9.4	1,286
66	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. <i>Nature Genetics</i> , 2008, 40, 322-328.	9.4	509
67	Polymorphisms of the HNF1A Gene Encoding Hepatocyte Nuclear Factor-1 $\alpha$ are Associated with C-Reactive Protein. <i>American Journal of Human Genetics</i> , 2008, 82, 1193-1201.	2.6	170
68	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. <i>Science</i> , 2008, 320, 539-543.	6.0	1,654
69	Qualifying the relationship between sequence conservation and molecular function. <i>Genome Research</i> , 2008, 18, 201-205.	2.4	91
70	A genome-wide scan for common genetic variants with a large influence on warfarin maintenance dose. <i>Blood</i> , 2008, 112, 1022-1027.	0.6	410
71	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. <i>American Journal of Human Genetics</i> , 2007, 81, 1057-1069.	2.6	222
72	Functional constraint and small insertions and deletions in the ENCODE regions of the human genome. <i>Genome Biology</i> , 2007, 8, R180.	13.9	32

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73	Analyses of deep mammalian sequence alignments and constraint predictions for 1% of the human genome. <i>Genome Research</i> , 2007, 17, 760-774.	2.4	184
74	Mammalian Comparative Sequence Analysis of the Agrp Locus. <i>PLoS ONE</i> , 2007, 2, e702.	1.1	5
75	Mutational and selective effects on copy-number variants in the human genome. <i>Nature Genetics</i> , 2007, 39, S22-S29.	9.4	221
76	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. <i>Nature</i> , 2007, 447, 799-816.	13.7	4,709
77	Distribution and intensity of constraint in mammalian genomic sequence. <i>Genome Research</i> , 2005, 15, 901-913.	2.4	1,230
78	TRADE-OFFS IN DETECTING EVOLUTIONARILY CONSTRAINED SEQUENCE BY COMPARATIVE GENOMICS. <i>Annual Review of Genomics and Human Genetics</i> , 2005, 6, 143-164.	2.5	41
79	Automated Whole-Genome Multiple Alignment of Rat, Mouse, and Human. <i>Genome Research</i> , 2004, 14, 685-692.	2.4	79
80	Characterization of Evolutionary Rates and Constraints in Three Mammalian Genomes. <i>Genome Research</i> , 2004, 14, 539-548.	2.4	125
81	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. <i>Nature</i> , 2004, 428, 493-521.	13.7	1,943
82	ABC: software for interactive browsing of genomic multiple sequence alignment data. <i>BMC Bioinformatics</i> , 2004, 5, 192.	1.2	12
83	Trends in occupational lead exposure since the 1978 OSHA lead standard. <i>American Journal of Industrial Medicine</i> , 2004, 45, 558-572.	1.0	22
84	Genomic regulatory regions: insights from comparative sequence analysis. <i>Current Opinion in Genetics and Development</i> , 2003, 13, 604-610.	1.5	57
85	Quantitative Estimates of Sequence Divergence for Comparative Analyses of Mammalian Genomes. <i>Genome Research</i> , 2003, 13, 813-820.	2.4	106
86	LAGAN and Multi-LAGAN: Efficient Tools for Large-Scale Multiple Alignment of Genomic DNA. <i>Genome Research</i> , 2003, 13, 721-731.	2.4	960