

James C Mullikin

List of Publications by Year in descending order

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80
papers

35,884
citations

101384

36
h-index

69108

77
g-index

87
all docs

87
docs citations

87
times ranked

43838
citing authors

#	ARTICLE	IF	CITATIONS
1	Initial sequencing and analysis of the human genome. <i>Nature</i> , 2001, 409, 860-921.	13.7	21,074
2	A second generation human haplotype map of over 3.1 million SNPs. <i>Nature</i> , 2007, 449, 851-861.	13.7	4,137
3	Accurate whole human genome sequencing using reversible terminator chemistry. <i>Nature</i> , 2008, 456, 53-59.	13.7	3,118
4	The complete sequence of a human genome. <i>Science</i> , 2022, 376, 44-53.	6.0	1,222
5	The DNA sequence of the human X chromosome. <i>Nature</i> , 2005, 434, 325-337.	13.7	985
6	Telomere-to-telomere assembly of a complete human X chromosome. <i>Nature</i> , 2020, 585, 79-84.	13.7	549
7	Exome sequencing of serous endometrial tumors identifies recurrent somatic mutations in chromatin-remodeling and ubiquitin ligase complex genes. <i>Nature Genetics</i> , 2012, 44, 1310-1315.	9.4	365
8	Initial sequence and comparative analysis of the cat genome. <i>Genome Research</i> , 2007, 17, 1675-1689.	2.4	311
9	Exome sequencing: the sweet spot before whole genomes. <i>Human Molecular Genetics</i> , 2010, 19, R145-R151.	1.4	263
10	QoRTs: a comprehensive toolset for quality control and data processing of RNA-Seq experiments. <i>BMC Bioinformatics</i> , 2015, 16, 224.	1.2	248
11	The ClinSeq Project: Piloting large-scale genome sequencing for research in genomic medicine. <i>Genome Research</i> , 2009, 19, 1665-1674.	2.4	236
12	A robust benchmark for detection of germline large deletions and insertions. <i>Nature Biotechnology</i> , 2020, 38, 1347-1355.	9.4	233
13	Automated, high-throughput derivation, characterization and differentiation of induced pluripotent stem cells. <i>Nature Methods</i> , 2015, 12, 885-892.	9.0	214
14	Systematic comparison of three genomic enrichment methods for massively parallel DNA sequencing. <i>Genome Research</i> , 2010, 20, 1420-1431.	2.4	194
15	Secondary Variants in Individuals Undergoing Exome Sequencing: Screening of 572 Individuals Identifies High-Penetrance Mutations in Cancer-Susceptibility Genes. <i>American Journal of Human Genetics</i> , 2012, 91, 97-108.	2.6	190
16	De novo assembly of the goldfish (<i>Carassius auratus</i>) genome and the evolution of genes after whole-genome duplication. <i>Science Advances</i> , 2019, 5, eaav0547.	4.7	182
17	VarSifter: Visualizing and analyzing exome-scale sequence variation data on a desktop computer. <i>Bioinformatics</i> , 2012, 28, 599-600.	1.8	137
18	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 100, 257-266.	2.6	127

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19	Detection and visualization of differential splicing in RNA-Seq data with JunctionSeq. <i>Nucleic Acids Research</i> , 2016, 44, gkw501.	6.5	126
20	Molecular genetic findings and clinical correlations in 100 patients with Joubert syndrome and related disorders prospectively evaluated at a single center. <i>Genetics in Medicine</i> , 2017, 19, 875-882.	1.1	100
21	A Defined Zebrafish Line for High-Throughput Genetics and Genomics: NHGRI-1. <i>Genetics</i> , 2014, 198, 167-170.	1.2	99
22	Somatic mutation profiles of clear cell endometrial tumors revealed by whole exome and targeted gene sequencing. <i>Cancer</i> , 2017, 123, 3261-3268.	2.0	72
23	Interpreting Secondary Cardiac Disease Variants in an Exome Cohort. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 337-346.	5.1	70
24	Using Exome Data to Identify Malignant Hyperthermia Susceptibility Mutations. <i>Anesthesiology</i> , 2013, 119, 1043-1053.	1.3	69
25	Low mutation burden and frequent loss of CDKN2A/B and SMARCA2, but not PRC2, define premalignant neurofibromatosis type 1-associated atypical neurofibromas. <i>Neuro-Oncology</i> , 2019, 21, 981-992.	0.6	69
26	GNE myopathy: New name and new mutation nomenclature. <i>Neuromuscular Disorders</i> , 2014, 24, 387-389.	0.3	61
27	Shimmer: detection of genetic alterations in tumors using next-generation sequence data. <i>Bioinformatics</i> , 2013, 29, 1498-1503.	1.8	59
28	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. <i>American Journal of Human Genetics</i> , 2019, 104, 1127-1138.	2.6	59
29	Prospective Evaluation of Kidney Disease in Joubert Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2017, 12, 1962-1973.	2.2	56
30	Extent of Linkage Disequilibrium in the Domestic Cat, <i>Felis silvestris catus</i> , and Its Breeds. <i>PLoS ONE</i> , 2013, 8, e53537.	1.1	54
31	Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. <i>Brain</i> , 2017, 140, 568-581.	3.7	53
32	Sialyllactose ameliorates myopathic phenotypes in symptomatic GNE myopathy model mice. <i>Brain</i> , 2014, 137, 2670-2679.	3.7	52
33	Light whole genome sequence for SNP discovery across domestic cat breeds. <i>BMC Genomics</i> , 2010, 11, 406.	1.2	51
34	Mutations in human homologue of chicken <i>talpid3</i> gene (<i>KIAA0586</i>) cause a hybrid ciliopathy with overlapping features of Jeune and Joubert syndromes. <i>Journal of Medical Genetics</i> , 2015, 52, 830-839.	1.5	47
35	Expanding the clinical and molecular characteristics of PIGT-CDG, a disorder of glycosylphosphatidylinositol anchors. <i>Molecular Genetics and Metabolism</i> , 2015, 115, 128-140.	0.5	44
36	Joubert Syndrome: Ophthalmological Findings in Correlation with Genotype and Hepatorenal Disease in 99 Patients Prospectively Evaluated at a Single Center. <i>Ophthalmology</i> , 2018, 125, 1937-1952.	2.5	43

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37	ATP6V1H Deficiency Impairs Bone Development through Activation of MMP9 and MMP13. <i>PLoS Genetics</i> , 2017, 13, e1006481.	1.5	42
38	DNA methylation in mice is influenced by genetics as well as sex and life experience. <i>Nature Communications</i> , 2019, 10, 305.	5.8	40
39	Applications and efficiencies of the first cat 63K DNA array. <i>Scientific Reports</i> , 2018, 8, 7024.	1.6	38
40	Congenital protein losing enteropathy: an inborn error of lipid metabolism due to DGAT1 mutations. <i>European Journal of Human Genetics</i> , 2016, 24, 1268-1273.	1.4	37
41	To the Root of the Curl: A Signature of a Recent Selective Sweep Identifies a Mutation That Defines the Cornish Rex Cat Breed. <i>PLoS ONE</i> , 2013, 8, e67105.	1.1	32
42	Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. <i>Human Genetics</i> , 2017, 136, 399-408.	1.8	30
43	Neurotranscriptomics: The Effects of Neonatal Stimulus Deprivation on the Rat Pineal Transcriptome. <i>PLoS ONE</i> , 2015, 10, e0137548.	1.1	29
44	The <i>FOXA2</i> transcription factor is frequently somatically mutated in uterine carcinosarcomas and carcinomas. <i>Cancer</i> , 2018, 124, 65-73.	2.0	27
45	Cystic cerebellar dysplasia and biallelic <i>LAMA1</i> mutations: a lamininopathy associated with tics, obsessive compulsive traits and myopia due to cell adhesion and migration defects. <i>Journal of Medical Genetics</i> , 2016, 53, 318-329.	1.5	25
46	<i>CELSR2</i> , encoding a planar cell polarity protein, is a putative gene in Joubert syndrome with cortical heterotopia, microphthalmia, and growth hormone deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 661-666.	0.7	25
47	Atypical presentation of GNE myopathy with asymmetric hand weakness. <i>Neuromuscular Disorders</i> , 2014, 24, 1063-1067.	0.3	21
48	Neurologic involvement in patients with atypical Chediak-Higashi disease. <i>Neurology</i> , 2017, 88, e57-e65.	1.5	20
49	Phospholipase A ₂ -activating protein is associated with a novel form of leukoencephalopathy. <i>Brain</i> , 2017, 140, 370-386.	3.7	18
50	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2018, 103, 948-967.	2.6	18
51	Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. <i>American Journal of Human Genetics</i> , 2018, 103, 794-807.	2.6	18
52	Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in <i>IQ Motif</i> containing <i>K</i> (<i>IQCK</i>)?. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 424-432.	0.6	17
53	<i>TMEM231</i> Gene Conversion Associated with Joubert and Meckel-Gruber Syndromes in the Same Family. <i>Human Mutation</i> , 2016, 37, 1144-1148.	1.1	16
54	Bleomycin Induces Drug Efflux in Lungs. A Pitfall for Pharmacological Studies of Pulmonary Fibrosis. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2020, 62, 178-190.	1.4	16

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55	NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology. <i>Journal of Clinical Medicine</i> , 2020, 9, 12.	1.0	16
56	Comparative clinical and genomic analysis of neurofibromatosis type 2-associated cranial and spinal meningiomas. <i>Scientific Reports</i> , 2020, 10, 12563.	1.6	16
57	CB ₁ R and iNOS are distinct players promoting pulmonary fibrosis in Hermansky-Pudlak syndrome. <i>Clinical and Translational Medicine</i> , 2021, 11, e471.	1.7	16
58	Assessing the capability of massively parallel sequencing for opportunistic pharmacogenetic screening. <i>Genetics in Medicine</i> , 2017, 19, 357-361.	1.1	15
59	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 49-57.	0.5	15
60	The transcription factors Ets1 and Sox10 interact during murine melanocyte development. <i>Developmental Biology</i> , 2015, 407, 300-312.	0.9	14
61	Abnormal glycosylation in Joubert syndrome type 10. <i>Cilia</i> , 2017, 6, 2.	1.8	14
62	First insight into the somatic mutation burden of neurofibromatosis type 2-associated grade I and grade II meningiomas: a case report comprehensive genomic study of two cranial meningiomas with vastly different clinical presentation. <i>BMC Cancer</i> , 2017, 17, 127.	1.1	13
63	Hermansky-Pudlak syndrome and oculocutaneous albinism in Chinese children with pigmentation defects and easy bruising. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 52.	1.2	13
64	Automated Digital Quantification of Pulmonary Fibrosis in Human Histopathology Specimens. <i>Frontiers in Medicine</i> , 2021, 8, 607720.	1.2	13
65	Loss of function mutations in VARS encoding cytoplasmic valyl-tRNA synthetase cause microcephaly, seizures, and progressive cerebral atrophy. <i>Human Genetics</i> , 2018, 137, 293-303.	1.8	12
66	Admixture mapping identifies genetic regions associated with blood pressure phenotypes in African Americans. <i>PLoS ONE</i> , 2020, 15, e0232048.	1.1	12
67	Identification of a novel mutation in HPS6 in a patient with hemophilia B and oculocutaneous albinism. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 284-287.	0.5	9
68	Neurodegeneration as the presenting symptom in 2 adults with xeroderma pigmentosum complementation group F. <i>Neurology: Genetics</i> , 2018, 4, e240.	0.9	9
69	Cerebral and portal vein thrombosis, macrocephaly and atypical absence seizures in Glycosylphosphatidyl inositol deficiency due to a PIGM promoter mutation. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 151-161.	0.5	9
70	Alternative Isoform Analysis of Ttc8 Expression in the Rat Pineal Gland Using a Multi-Platform Sequencing Approach Reveals Neural Regulation. <i>PLoS ONE</i> , 2016, 11, e0163590.	1.1	8
71	Severe bleeding with subclinical oculocutaneous albinism in a patient with a novel HPS6 missense variant. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2819-2823.	0.7	7
72	Cortical atrophy and hypofibrinogenemia due to FGG and TBCD mutations in a single family: a case report. <i>BMC Medical Genetics</i> , 2018, 19, 80.	2.1	7

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73	Novel truncating mutation in <i>TENM3</i> in siblings with motor developmental delay, ocular coloboma, oval cornea, without microphthalmia. American Journal of Medical Genetics, Part A, 2018, 176, 2930-2933.	0.7	3
74	The evolution of comparative genomics. Molecular Genetics & Genomic Medicine, 2014, 2, 363-368.	0.6	2
75	KLF3 and PAX6 are candidate driver genes in late-stage, MSI-hypermethylated endometrioid endometrial carcinomas. PLoS ONE, 2022, 17, e0251286.	1.1	2
76	Progressive pulmonary fibrosis in a murine model of Hermansky-Pudlak syndrome. Respiratory Research, 2022, 23, 112.	1.4	1
77	Title is missing!. , 2020, 15, e0232048.		0
78	Title is missing!. , 2020, 15, e0232048.		0
79	Title is missing!. , 2020, 15, e0232048.		0
80	Title is missing!. , 2020, 15, e0232048.		0