James C Mullikin

List of Publications by Year
in descending order

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80	35,884	101543	69250
papers	citations	36 h-index	77 g-index

87	87	87	43838
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	27.8	21,074
2	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	27.8	4,137
3	Accurate whole human genome sequencing using reversible terminator chemistry. Nature, 2008, 456, 53-59.	27.8	3,118
4	The complete sequence of a human genome. Science, 2022, 376, 44-53.	12.6	1,222
5	The DNA sequence of the human X chromosome. Nature, 2005, 434, 325-337.	27.8	985
6	Telomere-to-telomere assembly of a complete human X chromosome. Nature, 2020, 585, 79-84.	27.8	549
7	Exome sequencing of serous endometrial tumors identifies recurrent somatic mutations in chromatin-remodeling and ubiquitin ligase complex genes. Nature Genetics, 2012, 44, 1310-1315.	21.4	365
8	Initial sequence and comparative analysis of the cat genome. Genome Research, 2007, 17, 1675-1689.	5.5	311
9	Exome sequencing: the sweet spot before whole genomes. Human Molecular Genetics, 2010, 19, R145-R151.	2.9	263
10	QoRTs: a comprehensive toolset for quality control and data processing of RNA-Seq experiments. BMC Bioinformatics, 2015, 16, 224.	2.6	248
11	The ClinSeq Project: Piloting large-scale genome sequencing for research in genomic medicine. Genome Research, 2009, 19, 1665-1674.	5.5	236
12	A robust benchmark for detection of germline large deletions and insertions. Nature Biotechnology, 2020, 38, 1347-1355.	17.5	233
13	Automated, high-throughput derivation, characterization and differentiation of induced pluripotent stem cells. Nature Methods, 2015, 12, 885-892.	19.0	214
14	Systematic comparison of three genomic enrichment methods for massively parallel DNA sequencing. Genome Research, 2010, 20, 1420-1431.	5.5	194
15	Secondary Variants in Individuals Undergoing Exome Sequencing: Screening of 572 Individuals Identifies High-Penetrance Mutations in Cancer-Susceptibility Genes. American Journal of Human Genetics, 2012, 91, 97-108.	6.2	190
16	De novo assembly of the goldfish (Carassius auratus) genome and the evolution of genes after whole-genome duplication. Science Advances, 2019, 5, eaav0547.	10.3	182
17	VarSifter: Visualizing and analyzing exome-scale sequence variation data on a desktop computer. Bioinformatics, 2012, 28, 599-600.	4.1	137
18	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	6.2	127

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19	Detection and visualization of differential splicing in RNA-Seq data with JunctionSeq. Nucleic Acids Research, 2016, 44, gkw501.	14.5	126
20	Molecular genetic findings and clinical correlations in 100 patients with Joubert syndrome and related disorders prospectively evaluated at a single center. Genetics in Medicine, 2017, 19, 875-882.	2.4	100
21	A Defined Zebrafish Line for High-Throughput Genetics and Genomics: NHGRI-1. Genetics, 2014, 198, 167-170.	2.9	99
22	Somatic mutation profiles of clear cell endometrial tumors revealed by whole exome and targeted gene sequencing. Cancer, 2017, 123, 3261-3268.	4.1	72
23	Interpreting Secondary Cardiac Disease Variants in an Exome Cohort. Circulation: Cardiovascular Genetics, 2013, 6, 337-346.	5.1	70
24	Using Exome Data to Identify Malignant Hyperthermia Susceptibility Mutations. Anesthesiology, 2013, 119, 1043-1053.	2.5	69
25	Low mutation burden and frequent loss of CDKN2A/B and SMARCA2, but not PRC2, define premalignant neurofibromatosis type 1â€“associated atypical neurofibromas. Neuro-Oncology, 2019, 21, 981-992.	1.2	69
26	GNE myopathy: New name and new mutation nomenclature. Neuromuscular Disorders, 2014, 24, 387-389.	0.6	61
27	Shimmer: detection of genetic alterations in tumors using next-generation sequence data. Bioinformatics, 2013, 29, 1498-1503.	4.1	59
28	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
29	Prospective Evaluation of Kidney Disease in Joubert Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1962-1973.	4.5	56
30	Extent of Linkage Disequilibrium in the Domestic Cat, Felis silvestris catus, and Its Breeds. PLoS ONE, 2013, 8, e53537.	2.5	54
31	Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. Brain, 2017, 140, 568-581.	7.6	53
32	Sialyllactose ameliorates myopathic phenotypes in symptomatic GNE myopathy model mice. Brain, 2014, 137, 2670-2679.	7.6	52
33	Light whole genome sequence for SNP discovery across domestic cat breeds. BMC Genomics, 2010, 11, 406.	2.8	51
34	Mutations in human homologue of chickentalpid3gene (KIAA0586) cause a hybrid ciliopathy with overlapping features of Jeune and Joubert syndromes. Journal of Medical Genetics, 2015, 52, 830-839.	3.2	47
35	Expanding the clinical and molecular characteristics of PIGT-CDG, a disorder of glycosylphosphatidylinositol anchors. Molecular Genetics and Metabolism, 2015, 115, 128-140.	1.1	44
36	Joubert Syndrome: Ophthalmological Findings in Correlation with Genotype and Hepatorenal Disease in 99 Patients Prospectively Evaluated at a Single Center. Ophthalmology, 2018, 125, 1937-1952.	5.2	43

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37	ATP6V1H Deficiency Impairs Bone Development through Activation of MMP9 and MMP13. PLoS Genetics, 2017, 13, e1006481.	3.5	42
38	DNA methylation in mice is influenced by genetics as well as sex and life experience. Nature Communications, 2019, 10, 305.	12.8	40
39	Applications and efficiencies of the first cat 63K DNA array. Scientific Reports, 2018, 8, 7024.	3.3	38
40	Congenital protein losing enteropathy: an inborn error of lipid metabolism due to DGAT1 mutations. European Journal of Human Genetics, 2016, 24, 1268-1273.	2.8	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. PLoS ONE, 2013, 8, e67105.	2.5	32
42	Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. Human Genetics, 2017, 136, 399-408.	3.8	30
43	Neurotranscriptomics: The Effects of Neonatal Stimulus Deprivation on the Rat Pineal Transcriptome. PLoS ONE, 2015, 10, e0137548.	2.5	29
44	The FOXA2 transcription factor is frequently somatically mutated in uterine carcinosarcomas and carcinomas. Cancer, 2018, 124, 65-73.	4.1	27
45	Cystic cerebellar dysplasia and biallelicLAMA1mutations: a lamininopathy associated with tics, obsessive compulsive traits and myopia due to cell adhesion and migration defects. Journal of Medical Genetics, 2016, 53, 318-329.	3.2	25
46	CELSR2, encoding a planar cell polarity protein, is a putative gene in Joubert syndrome with cortical heterotopia, microophthalmia, and growth hormone deficiency. American Journal of Medical Genetics, Part A, 2017, 173, 661-666.	1.2	25
47	Atypical presentation of GNE myopathy with asymmetric hand weakness. Neuromuscular Disorders, 2014, 24, 1063-1067.	0.6	21
48	Neurologic involvement in patients with atypical Chediak-Higashi disease. Neurology, 2017, 88, e57-e65.	1.1	20
49	Phospholipase A₂-activating protein is associated with a novel form of leukoencephalopathy. Brain, 2017, 140, 370-386.	7.6	18
50	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	6.2	18
51	Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. American Journal of Human Genetics, 2018, 103, 794-807.	6.2	18
52	Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in IQ Motif containing K (IQCK)?. Molecular Genetics & Genomic Medicine, 2015, 3, 424-432.	1.2	17
53	TMEM231Gene Conversion Associated with Joubert and Meckel-Gruber Syndromes in the Same Family. Human Mutation, 2016, 37, 1144-1148.	2.5	16
54	Bleomycin Induces Drug Efflux in Lungs. A Pitfall for Pharmacological Studies of Pulmonary Fibrosis. American Journal of Respiratory Cell and Molecular Biology, 2020, 62, 178-190.	2.9	16

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55	NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology. Journal of Clinical Medicine, 2020, 9, 12.	2.4	16
56	Comparative clinical and genomic analysis of neurofibromatosis type 2-associated cranial and spinal meningiomas. Scientific Reports, 2020, 10, 12563.	3.3	16
57	CB₁R and iNOS are distinct players promoting pulmonary fibrosis in Hermanskyâ€“Pudlak syndrome. Clinical and Translational Medicine, 2021, 11, e471.	4.0	16
58	Assessing the capability of massively parallel sequencing for opportunistic pharmacogenetic screening. Genetics in Medicine, 2017, 19, 357-361.	2.4	15
59	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. Molecular Genetics and Metabolism, 2020, 130, 49-57.	1.1	15
60	The transcription factors Ets1 and Sox10 interact during murine melanocyte development. Developmental Biology, 2015, 407, 300-312.	2.0	14
61	Abnormal glycosylation in Joubert syndrome type 10. Cilia, 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. BMC Cancer, 2017, 17, 127.	2.6	13
63	Hermansky-Pudlak syndrome and oculocutaneous albinism in Chinese children with pigmentation defects and easy bruising. Orphanet Journal of Rare Diseases, 2019, 14, 52.	2.7	13
64	Automated Digital Quantification of Pulmonary Fibrosis in Human Histopathology Specimens. Frontiers in Medicine, 2021, 8, 607720.	2.6	13
65	Loss of function mutations in VARS encoding cytoplasmic valyl-tRNA synthetase cause microcephaly, seizures, and progressive cerebral atrophy. Human Genetics, 2018, 137, 293-303.	3.8	12
66	Admixture mapping identifies genetic regions associated with blood pressure phenotypes in African Americans. PLoS ONE, 2020, 15, e0232048.	2.5	12
67	Identification of a novel mutation in HPS6 in a patient with hemophilia B and oculocutaneous albinism. Molecular Genetics and Metabolism, 2016, 119, 284-287.	1.1	9
68	Neurodegeneration as the presenting symptom in 2 adults with xeroderma pigmentosum complementation group F. Neurology: Genetics, 2018, 4, e240.	1.9	9
69	Cerebral and portal vein thrombosis, macrocephaly and atypical absence seizures in Glycosylphosphatidyl inositol deficiency due to a PIGM promoter mutation. Molecular Genetics and Metabolism, 2019, 128, 151-161.	1.1	9
70	Alternative Isoform Analysis of Ttc8 Expression in the Rat Pineal Gland Using a Multi-Platform Sequencing Approach Reveals Neural Regulation. PLoS ONE, 2016, 11, e0163590.	2.5	8
71	Severe bleeding with subclinical oculocutaneous albinism in a patient with a novel HPS6 missense variant. American Journal of Medical Genetics, Part A, 2018, 176, 2819-2823.	1.2	7
72	Cortical atrophy and hypofibrinogenemia due to FGG and TBCD mutations in a single family: a case report. BMC Medical Genetics, 2018, 19, 80.	2.1	7

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73	Novel truncating mutation in TENM3 in siblings with motor developmental delay, ocular coloboma, oval cornea, without microphthalmia. American Journal of Medical Genetics, Part A, 2018, 176, 2930-2933.	1.2	3
74	The evolution of comparative genomics. Molecular Genetics & Genomic Medicine, 2014, 2, 363-368.	1.2	2
75	KLF3 and PAX6 are candidate driver genes in late-stage, MSI-hypermutated endometrioid endometrial carcinomas. PLoS ONE, 2022, 17, e0251286.	2.5	2
76	Progressive pulmonary fibrosis in a murine model of Hermansky-Pudlak syndrome. Respiratory Research, 2022, 23, 112.	3.6	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