

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
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87
all docs

87
docs citations

87
times ranked

43838
citing authors

#	ARTICLE	IF	CITATIONS
1	KLF3 and PAX6 are candidate driver genes in late-stage, MSI-hypermethylated endometrioid endometrial carcinomas. PLoS ONE, 2022, 17, e0251286.	1.1	2
2	The complete sequence of a human genome. Science, 2022, 376, 44-53.	6.0	1,222
3	Progressive pulmonary fibrosis in a murine model of Hermansky-Pudlak syndrome. Respiratory Research, 2022, 23, 112.	1.4	1
4	Automated Digital Quantification of Pulmonary Fibrosis in Human Histopathology Specimens. Frontiers in Medicine, 2021, 8, 607720.	1.2	13
5	CB ₁ R and iNOS are distinct players promoting pulmonary fibrosis in Hermansky-Pudlak syndrome. Clinical and Translational Medicine, 2021, 11, e471.	1.7	16
6	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.	1.4	16
7	NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology. Journal of Clinical Medicine, 2020, 9, 12.	1.0	16
8	Comparative clinical and genomic analysis of neurofibromatosis type 2-associated cranial and spinal meningiomas. Scientific Reports, 2020, 10, 12563.	1.6	16
9	Telomere-to-telomere assembly of a complete human X chromosome. Nature, 2020, 585, 79-84.	13.7	549
10	A robust benchmark for detection of germline large deletions and insertions. Nature Biotechnology, 2020, 38, 1347-1355.	9.4	233
11	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. Molecular Genetics and Metabolism, 2020, 130, 49-57.	0.5	15
12	Admixture mapping identifies genetic regions associated with blood pressure phenotypes in African Americans. PLoS ONE, 2020, 15, e0232048.	1.1	12
13	Title is missing!. , 2020, 15, e0232048.		0
14	Title is missing!. , 2020, 15, e0232048.		0
15	Title is missing!. , 2020, 15, e0232048.		0
16	Title is missing!. , 2020, 15, e0232048.		0
17	De novo assembly of the goldfish (<i>Carassius auratus</i>) genome and the evolution of genes after whole-genome duplication. Science Advances, 2019, 5, eaav0547.	4.7	182
18	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.	0.5	9

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19	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
20	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
21	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
22	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
23	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
24	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
25	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
26	Novel truncating mutation in <i>TENM3</i> in siblings with motor developmental delay, ocular coloboma, oval cornea, without microphthalmia. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2930-2933.	0.7	3
27	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
28	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
29	Neurodegeneration as the presenting symptom in 2 adults with xeroderma pigmentosum complementation group F. <i>Neurology: Genetics</i> , 2018, 4, e240.	0.9	9
30	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
31	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
32	Applications and efficiencies of the first cat 63K DNA array. <i>Scientific Reports</i> , 2018, 8, 7024.	1.6	38
33	Biallelic Mutations in DNJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 100, 257-266.	2.6	127
34	Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. <i>Human Genetics</i> , 2017, 136, 399-408.	1.8	30
35	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
36	Neurologic involvement in patients with atypical Chediak-Higashi disease. <i>Neurology</i> , 2017, 88, e57-e65.	1.5	20

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37	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
38	Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. <i>Brain</i> , 2017, 140, 568-581.	3.7	53
39	Phospholipase A ₂ -activating protein is associated with a novel form of leukoencephalopathy. <i>Brain</i> , 2017, 140, 370-386.	3.7	18
40	<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
41	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
42	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
43	Abnormal glycosylation in Joubert syndrome type 10. <i>Cilia</i> , 2017, 6, 2.	1.8	14
44	Assessing the capability of massively parallel sequencing for opportunistic pharmacogenetic screening. <i>Genetics in Medicine</i> , 2017, 19, 357-361.	1.1	15
45	ATP6V1H Deficiency Impairs Bone Development through Activation of MMP9 and MMP13. <i>PLoS Genetics</i> , 2017, 13, e1006481.	1.5	42
46	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
47	<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
48	Detection and visualization of differential splicing in RNA-Seq data with JunctionSeq. <i>Nucleic Acids Research</i> , 2016, 44, gkw501.	6.5	126
49	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
50	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
51	Alternative Isoform Analysis of <i>Ttc8</i> 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
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	QoRTs: a comprehensive toolset for quality control and data processing of RNA-Seq experiments. <i>BMC Bioinformatics</i> , 2015, 16, 224.	1.2	248
54	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

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55	Automated, high-throughput derivation, characterization and differentiation of induced pluripotent stem cells. <i>Nature Methods</i> , 2015, 12, 885-892.	9.0	214
56	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
57	The transcription factors Ets1 and Sox10 interact during murine melanocyte development. <i>Developmental Biology</i> , 2015, 407, 300-312.	0.9	14
58	Neurotranscriptomics: The Effects of Neonatal Stimulus Deprivation on the Rat Pineal Transcriptome. <i>PLoS ONE</i> , 2015, 10, e0137548.	1.1	29
59	Atypical presentation of GNE myopathy with asymmetric hand weakness. <i>Neuromuscular Disorders</i> , 2014, 24, 1063-1067.	0.3	21
60	The evolution of comparative genomics. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 363-368.	0.6	2
61	Sialyllactose ameliorates myopathic phenotypes in symptomatic GNE myopathy model mice. <i>Brain</i> , 2014, 137, 2670-2679.	3.7	52
62	GNE myopathy: New name and new mutation nomenclature. <i>Neuromuscular Disorders</i> , 2014, 24, 387-389.	0.3	61
63	A Defined Zebrafish Line for High-Throughput Genetics and Genomics: NHGRI-1. <i>Genetics</i> , 2014, 198, 167-170.	1.2	99
64	Interpreting Secondary Cardiac Disease Variants in an Exome Cohort. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 337-346.	5.1	70
65	Shimmer: detection of genetic alterations in tumors using next-generation sequence data. <i>Bioinformatics</i> , 2013, 29, 1498-1503.	1.8	59
66	Using Exome Data to Identify Malignant Hyperthermia Susceptibility Mutations. <i>Anesthesiology</i> , 2013, 119, 1043-1053.	1.3	69
67	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
68	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
69	VarSifter: Visualizing and analyzing exome-scale sequence variation data on a desktop computer. <i>Bioinformatics</i> , 2012, 28, 599-600.	1.8	137
70	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
71	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
72	Exome sequencing: the sweet spot before whole genomes. <i>Human Molecular Genetics</i> , 2010, 19, R145-R151.	1.4	263

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73	Light whole genome sequence for SNP discovery across domestic cat breeds. BMC Genomics, 2010, 11, 406.	1.2	51
74	Systematic comparison of three genomic enrichment methods for massively parallel DNA sequencing. Genome Research, 2010, 20, 1420-1431.	2.4	194
75	The ClinSeq Project: Piloting large-scale genome sequencing for research in genomic medicine. Genome Research, 2009, 19, 1665-1674.	2.4	236
76	Accurate whole human genome sequencing using reversible terminator chemistry. Nature, 2008, 456, 53-59.	13.7	3,118
77	Initial sequence and comparative analysis of the cat genome. Genome Research, 2007, 17, 1675-1689.	2.4	311
78	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	13.7	4,137
79	The DNA sequence of the human X chromosome. Nature, 2005, 434, 325-337.	13.7	985
80	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	13.7	21,074