# 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 | KLF3 and PAX6 are candidate driver genes in late-stage, MSI-hypermutated endometrioid endometrial carcinomas. PLoS ONE, 2022, 17, e0251286. | 2.5 | 2 |
| 2 | The complete sequence of a human genome. Science, 2022, 376, 44-53. | 12.6 | 1,222 |
| 3 | Progressive pulmonary fibrosis in a murine model of Hermansky-Pudlak syndrome. Respiratory Research, 2022, 23, 112. | 3.6 | 1 |
| 4 | Automated Digital Quantification of Pulmonary Fibrosis in Human Histopathology Specimens. Frontiers in Medicine, 2021, 8, 607720. | 2.6 | 13 |
| 5 | CB<sub>1</sub>R and iNOS are distinct players promoting pulmonary fibrosis in Hermanskyâ€“Pudlak syndrome. Clinical and Translational Medicine, 2021, 11, e471. | 4.0 | 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. | 2.9 | 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. | 2.4 | 16 |
| 8 | Comparative clinical and genomic analysis of neurofibromatosis type 2-associated cranial and spinal meningiomas. Scientific Reports, 2020, 10, 12563. | 3.3 | 16 |
| 9 | Telomere-to-telomere assembly of a complete human X chromosome. Nature, 2020, 585, 79-84. | 27.8 | 549 |
| 10 | A robust benchmark for detection of germline large deletions and insertions. Nature Biotechnology, 2020, 38, 1347-1355. | 17.5 | 233 |
| 11 | Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. Molecular Genetics and Metabolism, 2020, 130, 49-57. | 1.1 | 15 |
| 12 | Admixture mapping identifies genetic regions associated with blood pressure phenotypes in African Americans. PLoS ONE, 2020, 15, e0232048. | 2.5 | 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 ( Carassius auratus ) genome and the evolution of genes after whole-genome duplication. Science Advances, 2019, 5, eaav0547. | 10.3 | 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. | 1.1 | 9 |

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|---|---|---|---|
| 19 | DNA methylation in mice is influenced by genetics as well as sex and life experience. Nature Communications, 2019, 10, 305. | 12.8 | 40 |
| 20 | 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 |
| 21 | 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 |
| 22 | 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 |
| 23 | 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 |
| 24 | The FOXA2 transcription factor is frequently somatically mutated in uterine carcinosarcomas and carcinomas. Cancer, 2018, 124, 65-73. | 4.1 | 27 |
| 25 | 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 |
| 26 | 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 |
| 27 | 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 |
| 28 | 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 |
| 29 | Neurodegeneration as the presenting symptom in 2 adults with xeroderma pigmentosum complementation group F. Neurology: Genetics, 2018, 4, e240. | 1.9 | 9 |
| 30 | 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 |
| 31 | 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 |
| 32 | Applications and efficiencies of the first cat 63K DNA array. Scientific Reports, 2018, 8, 7024. | 3.3 | 38 |
| 33 | Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266. | 6.2 | 127 |
| 34 | Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. Human Genetics, 2017, 136, 399-408. | 3.8 | 30 |
| 35 | 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 |
| 36 | Neurologic involvement in patients with atypical Chediak-Higashi disease. Neurology, 2017, 88, e57-e65. | 1.1 | 20 |

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|---|---|---|---|
| 37 | Somatic mutation profiles of clear cell endometrial tumors revealed by whole exome and targeted gene sequencing. Cancer, 2017, 123, 3261-3268. | 4.1 | 72 |
| 38 | Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. Brain, 2017, 140, 568-581. | 7.6 | 53 |
| 39 | Phospholipase A<sub>2</sub>-activating protein is associated with a novel form of leukoencephalopathy. Brain, 2017, 140, 370-386. | 7.6 | 18 |
| 40 | 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 |
| 41 | Prospective Evaluation of Kidney Disease in Joubert Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1962-1973. | 4.5 | 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. BMC Cancer, 2017, 17, 127. | 2.6 | 13 |
| 43 | Abnormal glycosylation in Joubert syndrome type 10. Cilia, 2017, 6, 2. | 1.8 | 14 |
| 44 | Assessing the capability of massively parallel sequencing for opportunistic pharmacogenetic screening. Genetics in Medicine, 2017, 19, 357-361. | 2.4 | 15 |
| 45 | ATP6V1H Deficiency Impairs Bone Development through Activation of MMP9 and MMP13. PLoS Genetics, 2017, 13, e1006481. | 3.5 | 42 |
| 46 | 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 |
| 47 | TMEM231Gene Conversion Associated with Joubert and Meckel-Gruber Syndromes in the Same Family. Human Mutation, 2016, 37, 1144-1148. | 2.5 | 16 |
| 48 | Detection and visualization of differential splicing in RNA-Seq data with JunctionSeq. Nucleic Acids Research, 2016, 44, gkw501. | 14.5 | 126 |
| 49 | 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 |
| 50 | 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 |
| 51 | 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 |
| 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 | QoRTs: a comprehensive toolset for quality control and data processing of RNA-Seq experiments. BMC Bioinformatics, 2015, 16, 224. | 2.6 | 248 |
| 54 | 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 |

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|---|---|---|---|
| 55 | Automated, high-throughput derivation, characterization and differentiation of induced pluripotent stem cells. Nature Methods, 2015, 12, 885-892. | 19.0 | 214 |
| 56 | 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 |
| 57 | The transcription factors Ets1 and Sox10 interact during murine melanocyte development. Developmental Biology, 2015, 407, 300-312. | 2.0 | 14 |
| 58 | Neurotranscriptomics: The Effects of Neonatal Stimulus Deprivation on the Rat Pineal Transcriptome. PLoS ONE, 2015, 10, e0137548. | 2.5 | 29 |
| 59 | Atypical presentation of GNE myopathy with asymmetric hand weakness. Neuromuscular Disorders, 2014, 24, 1063-1067. | 0.6 | 21 |
| 60 | The evolution of comparative genomics. Molecular Genetics & Genomic Medicine, 2014, 2, 363-368. | 1.2 | 2 |
| 61 | Sialyllactose ameliorates myopathic phenotypes in symptomatic GNE myopathy model mice. Brain, 2014, 137, 2670-2679. | 7.6 | 52 |
| 62 | GNE myopathy: New name and new mutation nomenclature. Neuromuscular Disorders, 2014, 24, 387-389. | 0.6 | 61 |
| 63 | A Defined Zebrafish Line for High-Throughput Genetics and Genomics: NHGRI-1. Genetics, 2014, 198, 167-170. | 2.9 | 99 |
| 64 | Interpreting Secondary Cardiac Disease Variants in an Exome Cohort. Circulation: Cardiovascular Genetics, 2013, 6, 337-346. | 5.1 | 70 |
| 65 | Shimmer: detection of genetic alterations in tumors using next-generation sequence data. Bioinformatics, 2013, 29, 1498-1503. | 4.1 | 59 |
| 66 | Using Exome Data to Identify Malignant Hyperthermia Susceptibility Mutations. Anesthesiology, 2013, 119, 1043-1053. | 2.5 | 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. PLoS ONE, 2013, 8, e67105. | 2.5 | 32 |
| 68 | Extent of Linkage Disequilibrium in the Domestic Cat, Felis silvestris catus, and Its Breeds. PLoS ONE, 2013, 8, e53537. | 2.5 | 54 |
| 69 | VarSifter: Visualizing and analyzing exome-scale sequence variation data on a desktop computer. Bioinformatics, 2012, 28, 599-600. | 4.1 | 137 |
| 70 | 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 |
| 71 | 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 |
| 72 | Exome sequencing: the sweet spot before whole genomes. Human Molecular Genetics, 2010, 19, R145-R151. | 2.9 | 263 |

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|---|---|---|---|
| 73 | Light whole genome sequence for SNP discovery across domestic cat breeds. BMC Genomics, 2010, 11, 406. | 2.8 | 51 |
| 74 | Systematic comparison of three genomic enrichment methods for massively parallel DNA sequencing. Genome Research, 2010, 20, 1420-1431. | 5.5 | 194 |
| 75 | The ClinSeq Project: Piloting large-scale genome sequencing for research in genomic medicine. Genome Research, 2009, 19, 1665-1674. | 5.5 | 236 |
| 76 | Accurate whole human genome sequencing using reversible terminator chemistry. Nature, 2008, 456, 53-59. | 27.8 | 3,118 |
| 77 | Initial sequence and comparative analysis of the cat genome. Genome Research, 2007, 17, 1675-1689. | 5.5 | 311 |
| 78 | A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861. | 27.8 | 4,137 |
| 79 | The DNA sequence of the human X chromosome. Nature, 2005, 434, 325-337. | 27.8 | 985 |
| 80 | Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921. | 27.8 | 21,074 |