James Knight

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7739110/publications.pdf

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23 papers	2,304 citations	567281 15 h-index	610901 24 g-index
28 all docs	28 docs citations	28 times ranked	5587 citing authors

#	Article	IF	Citations
1	Integrative molecular and clinical profiling of acral melanoma links focal amplification of 22q11.21 to metastasis. Nature Communications, 2022, 13, 898.	12.8	19
2	Determining the serotype composition of mixed samples of pneumococcus using whole-genome sequencing. Microbial Genomics, 2021, 7, .	2.0	10
3	Genetics of agenesis/hypoplasia of the uterus and vagina: narrowing down the number of candidate genes for Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser Syndrome. Human Genetics, 2021, 140, 667-680.	3.8	16
4	DIAPH1 Variants in Nonâ€“East Asian Patients With Sporadic Moyamoya Disease. JAMA Neurology, 2021, 78, 993.	9.0	33
5	Dâ€bifunctional protein deficiency caused by splicing variants in a neonate with severe peroxisomal dysfunction and persistent hypoglycemia. American Journal of Medical Genetics, Part A, 2021, , .	1.2	2
6	Long-read single molecule real-time (SMRT) sequencing of GBA1 locus in Gaucher disease national cohort from Argentina reveals high frequency of complex allele underlying severe skeletal phenotypes: Collaborative study from the Argentine Group for Diagnosis and Treatment of Gaucher Disease. Molecular Genetics and Metabolism Reports, 2021, 29, 100820.	1.1	3
7	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	30.7	84
8	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	21.4	96
9	Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia. IScience, 2020, 23, 101552.	4.1	32
10	The omentum of obese girls harbors small adipocytes and browning transcripts. JCI Insight, 2020, 5, .	5.0	8
11	Genomic sites hypersensitive to ultraviolet radiation. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 24196-24205.	7.1	66
12	The Use of Whole Exome Sequencing in a Cohort of Transgender Individuals to Identify Rare Genetic Variants. Scientific Reports, 2019, 9, 20099.	3.3	18
13	Insights into the evolution and drug susceptibility of Babesia duncani from the sequence of its mitochondrial and apicoplast genomes. International Journal for Parasitology, 2019, 49, 105-113.	3.1	13
14	Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. Neuron, 2019, 101, 429-443.e4.	8.1	56
15	Hypokalemia Associated With a Claudin 10 Mutation: A Case Report. American Journal of Kidney Diseases, 2019, 73, 425-428.	1.9	24
16	Translocation of a gut pathobiont drives autoimmunity in mice and humans. Science, 2018, 359, 1156-1161.	12.6	608
17	Distinct adaptive mechanisms drive recovery from aneuploidy caused by loss of the Ulp2 SUMO protease. Nature Communications, 2018, 9, 5417.	12.8	21
18	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	8.1	112

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19	Inflammasome Priming Mediated via Toll-Like Receptors 2 and 4, Induces Th1-Like Regulatory T Cells in De Novo Autoimmune Hepatitis. Frontiers in Immunology, 2018, 9, 1612.	4.8	16
20	Clonal evolution analysis of paired anaplastic and wellâ€differentiated thyroid carcinomas reveals shared common ancestor. Genes Chromosomes and Cancer, 2018, 57, 645-652.	2.8	31
21	An improved genome assembly uncovers prolific tandem repeats in Atlantic cod. BMC Genomics, 2017, 18, 95.	2.8	153
22	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	21.4	624
23	Integrated genomic characterization of IDH1-mutant glioma malignant progression. Nature Genetics, 2016, 48, 59-66.	21.4	253