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	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	21.4	624
2	Translocation of a gut pathobiont drives autoimmunity in mice and humans. Science, 2018, 359, 1156-1161.	12.6	608
3	Integrated genomic characterization of IDH1-mutant glioma malignant progression. Nature Genetics, 2016, 48, 59-66.	21.4	253
4	An improved genome assembly uncovers prolific tandem repeats in Atlantic cod. BMC Genomics, 2017, 18, 95.	2.8	153
5	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	8.1	112
6	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	21.4	96
7	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	30.7	84
8	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
9	Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. Neuron, 2019, 101, 429-443.e4.	8.1	56
10	DIAPH1 Variants in Nonâ€“East Asian Patients With Sporadic Moyamoya Disease. JAMA Neurology, 2021, 78, 993.	9.0	33
11	Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia. IScience, 2020, 23, 101552.	4.1	32
12	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
13	Hypokalemia Associated With a Claudin 10 Mutation: A Case Report. American Journal of Kidney Diseases, 2019, 73, 425-428.	1.9	24
14	Distinct adaptive mechanisms drive recovery from aneuploidy caused by loss of the Ulp2 SUMO protease. Nature Communications, 2018, 9, 5417.	12.8	21
15	Integrative molecular and clinical profiling of acral melanoma links focal amplification of 22q11.21 to metastasis. Nature Communications, 2022, 13, 898.	12.8	19
16	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
17	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
18	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

#	Article	IF	Citations
19	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
20	Determining the serotype composition of mixed samples of pneumococcus using whole-genome sequencing. Microbial Genomics, 2021, 7, .	2.0	10
21	The omentum of obese girls harbors small adipocytes and browning transcripts. JCI Insight, 2020, 5, .	5.0	8
22	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
23	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