

Ian M Campbell

List of Publications by Year in descending order

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Version: 2024-02-01

43
papers

2,448
citations

236925

25
h-index

265206

42
g-index

45
all docs

45
docs citations

45
times ranked

5320
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical exome sequencing data reveal high diagnostic yields for congenital diaphragmatic hernia plus (CDH+) and new phenotypic expansions involving CDH. <i>Journal of Medical Genetics</i> , 2022, 59, 270-278.	3.2	27
2	A Case of Prenatally Diagnosed Periventricular Nodular Heterotopia in a Surviving Male Patient with FLNA Mutation. <i>Journal of Pediatric Neurology</i> , 2022, 20, 057-059.	0.2	0
3	Distinct immune trajectories in patients with chromosome 22q11.2 deletion syndrome and immune-mediated diseases. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 445-450.	2.9	15
4	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	2.4	44
5	Clinical Effectiveness of Telemedicine-Based Pediatric Genetics Care. <i>Pediatrics</i> , 2022, 150, .	2.1	5
6	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1649-1665.	1.2	34
7	Provider Perspectives on the Impact of the COVID-19 Pandemic on Newborn Screening. <i>International Journal of Neonatal Screening</i> , 2021, 7, 38.	3.2	4
8	Proposed criteria for nevoid basal cell carcinoma syndrome in children assessed using statistical optimization. <i>Scientific Reports</i> , 2021, 11, 19791.	3.3	5
9	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of ALX4 and TWIST1. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 916-922.	1.2	1
10	A Not So Common Infection in an Extremely Low-Birth-Weight Infant. <i>Clinical Pediatrics</i> , 2020, 59, 1040-1042.	0.8	2
11	Isolation of Aggressive Behavior Mutants in <i>Drosophila</i> Using a Screen for Wing Damage. <i>Genetics</i> , 2018, 208, 273-282.	2.9	15
12	Cover Image, Volume 176A, Number 10, October 2018. , 2018, 176, i-i.		0
13	What is new with 22q? An update from the 22q and You Center at the Children's Hospital of Philadelphia. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2058-2069.	1.2	106
14	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. <i>Journal of Pediatric Genetics</i> , 2018, 07, 164-173.	0.7	15
15	Predicting human genes susceptible to genomic instability associated with Alu-mediated rearrangements. <i>Genome Research</i> , 2018, 28, 1228-1242.	5.5	74
16	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. <i>Cell</i> , 2017, 168, 830-842.e7.	28.9	66
17	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. <i>Genome Medicine</i> , 2016, 8, 13.	8.2	37
18	Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. <i>Human Mutation</i> , 2016, 37, 231-234.	2.5	18

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19	Secondary findings and carrier test frequencies in a large multiethnic sample. <i>Genome Medicine</i> , 2015, 7, 54.	8.2	47
20	Comparative Genomic Analyses of the Human NPHP1 Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. <i>PLoS Genetics</i> , 2015, 11, e1005686.	3.5	21
21	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. <i>Human Molecular Genetics</i> , 2015, 24, 4061-4077.	2.9	83
22	Genome-wide analyses of LINE-mediated nonallelic homologous recombination. <i>Nucleic Acids Research</i> , 2015, 43, 2188-2198.	14.5	79
23	Somatic mosaicism: implications for disease and transmission genetics. <i>Trends in Genetics</i> , 2015, 31, 382-392.	6.7	234
24	Variant interpretation through Bayesian fusion of frequency and genomic knowledge. <i>Genome Medicine</i> , 2015, 7, 4.	8.2	1
25	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. <i>Neuron</i> , 2015, 88, 499-513.	8.1	258
26	Mus81 and converging forks limit the mutagenicity of replication fork breakage. <i>Science</i> , 2015, 349, 742-747.	12.6	162
27	Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. <i>BMC Biology</i> , 2014, 12, 74.	3.8	60
28	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. <i>Cell</i> , 2014, 157, 636-650.	28.9	189
29	NetComm: a network analysis tool based on communicability. <i>Bioinformatics</i> , 2014, 30, 3387-3389.	4.1	2
30	Parent of Origin, Mosaicism, and Recurrence Risk: Probabilistic Modeling Explains the Broken Symmetry of Transmission Genetics. <i>American Journal of Human Genetics</i> , 2014, 95, 345-359.	6.2	103
31	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. <i>American Journal of Human Genetics</i> , 2014, 95, 173-182.	6.2	219
32	The Alu-Rich Genomic Architecture of SPAST Predisposes to Diverse and Functionally Distinct Disease-Associated CNV Alleles. <i>American Journal of Human Genetics</i> , 2014, 95, 143-161.	6.2	87
33	Recurrent HERV-H-Mediated 3q13.2-q13.31 Deletions Cause a Syndrome of Hypotonia and Motor, Language, and Cognitive Delays. <i>Human Mutation</i> , 2013, 34, 1415-1423.	2.5	40
34	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. <i>American Journal of Human Genetics</i> , 2013, 93, 197-210.	6.2	43
35	Mutations in <i>VRK1</i> Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. <i>JAMA Neurology</i> , 2013, 70, 1491-8.	9.0	54
36	Fusion of Large-Scale Genomic Knowledge and Frequency Data Computationally Prioritizes Variants in Epilepsy. <i>PLoS Genetics</i> , 2013, 9, e1003797.	3.5	22

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37	Screening and familial characterization of copy-number variations in <i>NR5A1</i> in 46,XY disorders of sex development and premature ovarian failure. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2487-2494.	1.2	12
38	Incidental copy-number variants identified by routine genome testing in a clinical population. <i>Genetics in Medicine</i> , 2013, 15, 45-54.	2.4	37
39	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. <i>Genome Research</i> , 2013, 23, 1383-1394.	5.5	62
40	Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A. <i>Genetics in Medicine</i> , 2012, 14, 868-876.	2.4	51
41	Differential Transit Peptide Recognition during Preprotein Binding and Translocation into Flowering Plant Plastids. <i>Plant Cell</i> , 2012, 24, 3040-3059.	6.6	46
42	<i>TGFBR2</i> deletion in a 20-month-old female with developmental delay and microcephaly. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1442-1447.	1.2	18
43	In Vitro Comparative Kinetic Analysis of the Chloroplast Toc GTPases. <i>Journal of Biological Chemistry</i> , 2007, 282, 11410-11426.	3.4	43