Ian M Campbell

List of Publications by Year
in descending order

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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	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
2	Somatic mosaicism: implications for disease and transmission genetics. Trends in Genetics, 2015, 31, 382-392.	6.7	234
3	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
4	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	28.9	189
5	Mus81 and converging forks limit the mutagenicity of replication fork breakage. Science, 2015, 349, 742-747.	12.6	162
6	What is new with 22q? An update from the 22q and You Center at the Children's Hospital of Philadelphia. American Journal of Medical Genetics, Part A, 2018, 176, 2058-2069.	1.2	106
7	Parent of Origin, Mosaicism, and Recurrence Risk: Probabilistic Modeling Explains the Broken Symmetry of Transmission Genetics. American Journal of Human Genetics, 2014, 95, 345-359.	6.2	103
8	The Alu-Rich Genomic Architecture of SPAST Predisposes to Diverse and Functionally Distinct Disease-Associated CNV Alleles. American Journal of Human Genetics, 2014, 95, 143-161.	6.2	87
9	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. Human Molecular Genetics, 2015, 24, 4061-4077.	2.9	83
10	Genome-wide analyses of LINEâ€“LINE-mediated nonallelic homologous recombination. Nucleic Acids Research, 2015, 43, 2188-2198.	14.5	79
11	Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Research, 2018, 28, 1228-1242.	5.5	74
12	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	28.9	66
13	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394.	5.5	62
14	Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. BMC Biology, 2014, 12, 74.	3.8	60
15	Mutations inVRK1Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. JAMA Neurology, 2013, 70, 1491-8.	9.0	54
16	Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A. Genetics in Medicine, 2012, 14, 868-876.	2.4	51
17	Secondary findings and carrier test frequencies in a large multiethnic sample. Genome Medicine, 2015, 7, 54.	8.2	47
18	Differential Transit Peptide Recognition during Preprotein Binding and Translocation into Flowering Plant Plastids. Plant Cell, 2012, 24, 3040-3059.	6.6	46

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19	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
20	In Vitro Comparative Kinetic Analysis of the Chloroplast Toc GTPases. Journal of Biological Chemistry, 2007, 282, 11410-11426.	3.4	43
21	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
22	Recurrent HERV-H-Mediated 3q13.2-q13.31 Deletions Cause a Syndrome of Hypotonia and Motor, Language, and Cognitive Delays. Human Mutation, 2013, 34, 1415-1423.	2.5	40
23	Incidental copy-number variants identified by routine genome testing in a clinical population. Genetics in Medicine, 2013, 15, 45-54.	2.4	37
24	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. Genome Medicine, 2016, 8, 13.	8.2	37
25	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
26	Clinical exome sequencing data reveal high diagnostic yields for congenital diaphragmatic hernia plus (CDH+) and new phenotypic expansions involving CDH. Journal of Medical Genetics, 2022, 59, 270-278.	3.2	27
27	Fusion of Large-Scale Genomic Knowledge and Frequency Data Computationally Prioritizes Variants in Epilepsy. PLoS Genetics, 2013, 9, e1003797.	3.5	22
28	Comparative Genomic Analyses of the Human NPHP1 Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. PLoS Genetics, 2015, 11, e1005686.	3.5	21
29	TGFBR2 deletion in a 20â€monthâ€old female with developmental delay and microcephaly. American Journal of Medical Genetics, Part A, 2011, 155, 1442-1447.	1.2	18
30	Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. Human Mutation, 2016, 37, 231-234.	2.5	18
31	Isolation of Aggressive Behavior Mutants in Drosophila Using a Screen for Wing Damage. Genetics, 2018, 208, 273-282.	2.9	15
32	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. Journal of Pediatric Genetics, 2018, 07, 164-173.	0.7	15
33	Distinct immune trajectories in patients with chromosome 22q11.2 deletion syndrome and immune-mediated diseases. Journal of Allergy and Clinical Immunology, 2022, 149, 445-450.	2.9	15
34	Screening and familial characterization of copyâ€number variations in NR5A1 in 46,XY disorders of sex development and premature ovarian failure. American Journal of Medical Genetics, Part A, 2013, 161, 2487-2494.	1.2	12
35	Proposed criteria for nevoid basal cell carcinoma syndrome in children assessed using statistical optimization. Scientific Reports, 2021, 11, 19791.	3.3	5
36	Clinical Effectiveness of Telemedicine-Based Pediatric Genetics Care. Pediatrics, 2022, 150, .	2.1	5

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37	Provider Perspectives on the Impact of the COVID-19 Pandemic on Newborn Screening. International Journal of Neonatal Screening, 2021, 7, 38.	3.2	4
38	NetComm: a network analysis tool based on communicability. Bioinformatics, 2014, 30, 3387-3389.	4.1	2
39	A Not So Common Infection in an Extremely Low-Birth-Weight Infant. Clinical Pediatrics, 2020, 59, 1040-1042.	0.8	2
40	Variant interpretation through Bayesian fusion of frequency and genomic knowledge. Genome Medicine, 2015, 7, 4.	8.2	1
41	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of ALX4 and TWIST1. American Journal of Medical Genetics, Part A, 2021, 185, 916-922.	1.2	1
42	Cover Image, Volume 176A, Number 10, October 2018. , 2018, 176, i-i.		0
43	A Case of Prenatally Diagnosed Periventricular Nodular Heterotopia in a Surviving Male Patient with FLNA Mutation. Journal of Pediatric Neurology, 2022, 20, 057-059.	0.2	0