Ian M Carr

List of Publications by Year
in descending order

48 papers	1,436 citations	394421 19 h-index	361022 35 g-index
51 all docs	51 docs citations	51 times ranked	3193 citing authors

#	Article	IF	Citations
1	CCDC151 Mutations Cause Primary Ciliary Dyskinesia by Disruption of the Outer Dynein Arm Docking Complex Formation. American Journal of Human Genetics, 2014, 95, 257-274.	6.2	149
2	Interactive visual analysis of SNP data for rapid autozygosity mapping in consanguineous families. Human Mutation, 2006, 27, 1041-1046.	2.5	140
3	An analysis of IL-36 signature genes and individuals with IL1RL2 knockout mutations validates IL-36 as a psoriasis therapeutic target. Science Translational Medicine, 2017, 9, .	12.4	124
4	The Tudor SND1 protein is an m6A RNA reader essential for replication of Kaposiâ€™s sarcoma-associated herpesvirus. ELife, 2019, 8, .	6.0	107
5	HEATR2 Plays a Conserved Role in Assembly of the Ciliary Motile Apparatus. PLoS Genetics, 2014, 10, e1004577.	3.5	67
6	Enhanced diagnostic yield in Meckel-Gruber and Joubert syndrome through exome sequencing supplemented with split-read mapping. BMC Medical Genetics, 2016, 17, 1.	2.1	67
7	Biallelic Mutations in PDE10A Lead to Loss of Striatal PDE10A and a Hyperkinetic Movement Disorder with Onset in Infancy. American Journal of Human Genetics, 2016, 98, 735-743.	6.2	65
8	IL-36 Promotes Systemic IFN-I Responses in Severe Forms of Psoriasis. Journal of Investigative Dermatology, 2020, 140, 816-826.e3.	0.7	64
9	Sequence analysis and editing for bisulphite genomic sequencing projects. Nucleic Acids Research, 2007, 35, e79.	14.5	50
10	Autozygosity Mapping with Exome Sequence Data. Human Mutation, 2013, 34, 50-56.	2.5	49
11	Biallelic Mutations in LRRC56, Encoding a Protein Associated with Intraflagellar Transport, Cause Mucociliary Clearance and Laterality Defects. American Journal of Human Genetics, 2018, 103, 727-739.	6.2	49
12	HACE1deficiency causes an autosomal recessive neurodevelopmental syndrome. Journal of Medical Genetics, 2015, 52, 797-803.	3.2	40
13	m6aViewer: software for the detection, analysis, and visualization of N⁶-methyladenosine peaks from m⁶A-seq/ME-RIP sequencing data. Rna, 2017, 23, 1493-1501.	3.5	34
14	Cas9-based enrichment and single-molecule sequencing for precise characterization of genomic duplications. Laboratory Investigation, 2020, 100, 135-146.	3.7	33
15	Novel universal primers for metabarcoding environmental DNA surveys of marine mammals and other marine vertebrates. Environmental DNA, 2020, 2, 460-476.	5.8	26
16	IBDfinderandSNPsetter: Tools for pedigree-independent identification of autozygous regions in individuals with recessive inherited disease. Human Mutation, 2009, 30, 960-967.	2.5	25
17	GeneTIER: prioritization of candidate disease genes using tissue-specific gene expression profiles. Bioinformatics, 2015, 31, 2728-2735.	4.1	25
18	OVA: integrating molecular and physical phenotype data from multiple biomedical domain ontologies with variant filtering for enhanced variant prioritization. Bioinformatics, 2015, 31, 3822-3829.	4.1	24

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19	Primary Hypertrophic Osteoarthropathy: An Update on Patient Features and Treatment. Journal of Rheumatology, 2015, 42, 2211.2-2214.	2.0	24
20	A tubulin alpha 8 mouse knockout model indicates a likely role in spermatogenesis but not in brain development. PLoS ONE, 2017, 12, e0174264.	2.5	23
21	Targeting human plasmacytoid dendritic cells through BDCA2 prevents skin inflammation and fibrosis in a novel xenotransplant mouse model of scleroderma. Annals of the Rheumatic Diseases, 2021, 80, 920-929.	0.9	23
22	GeneScreen: a program for high-throughput mutation detection in DNA sequence electropherograms. Journal of Medical Genetics, 2011, 48, 123-130.	3.2	20
23	Longâ€read nanopore sequencing resolves a TMEM231 gene conversion event causing Meckelâ€“Gruber syndrome. Human Mutation, 2020, 41, 525-531.	2.5	18
24	Paracrine cyclooxygenase-2 activity by macrophages drives colorectal adenoma progression in the Apc Min/+ mouse model of intestinal tumorigenesis. Scientific Reports, 2017, 7, 6074.	3.3	17
25	Rapid Detection of Rare Deleterious Variants by Next Generation Sequencing with Optional Microarray SNP Genotype Data. Human Mutation, 2015, 36, 823-830.	2.5	15
26	Deficiency of the myogenic factor MyoD causes a perinatally lethal fetal akinesia. Journal of Medical Genetics, 2016, 53, 264-269.	3.2	15
27	Induction of epigenetic variation in Arabidopsis by over-expression of DNA METHYLTRANSFERASE1 (MET1). PLoS ONE, 2018, 13, e0192170.	2.5	15
28	CantÃº syndrome with coexisting familial pituitary adenoma. Endocrine, 2018, 59, 677-684.	2.3	13
29	Novel avian paramyxovirus isolated from gulls in Caspian seashore in Kazakhstan. PLoS ONE, 2017, 12, e0190339.	2.5	12
30	Identification and Characterization of a Sequence Related to Human Sorbitol Dehydrogenase. FEBS Journal, 1997, 245, 760-767.	0.2	10
31	Ferries and Environmental DNA: Underway Sampling From Commercial Vessels Provides New Opportunities for Systematic Genetic Surveys of Marine Biodiversity. Frontiers in Marine Science, 2021, 8, .	2.5	10
32	Estimating the degree of identity by descent in consanguineous couples. Human Mutation, 2011, 32, 1350-1358.	2.5	9
33	Assessing the utility of long-read nanopore sequencing for rapid and efficient characterization of mobile element insertions. Laboratory Investigation, 2021, 101, 442-449.	3.7	9
34	Transcriptome profiles of stem-like cells from primary breast cancers allow identification of ITGA7 as a predictive marker of chemotherapy response. British Journal of Cancer, 2021, 125, 983-993.	6.4	9
35	DominantMapper: Rule-based analysis of SNP data for rapid mapping of dominant diseases in related nuclear families. Human Mutation, 2011, 32, 1359-1366.	2.5	6
36	Shadow autozygosity mapping by linkage exclusion (SAMPLE): a simple strategy to identify the genetic basis of lethal autosomal recessive disorders. Human Mutation, 2009, 30, 1642-1649.	2.5	5

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37	Illuminator, a desktop program for mutation detection using short-read clonal sequencing. Genomics, 2011, 98, 302-309.	2.9	5
38	Effects of EGFR Inhibitor on Helicobacter pylori Induced Gastric Epithelial Pathology in Vivo. Pathogens, 2013, 2, 571-590.	2.8	5
39	Characterization and Genomic Localization of a SMAD4 Processed Pseudogene. Journal of Molecular Diagnostics, 2017, 19, 933-940.	2.8	5
40	A Chromosome 7 Pericentric Inversion Defined at Single-Nucleotide Resolution Using Diagnostic Whole Genome Sequencing in a Patient with Hand-Foot-Genital Syndrome. PLoS ONE, 2016, 11, e0157075.	2.5	5
41	Structural and evolutionary characterization of the human sorbitol dehydrogenase gene duplication. Mammalian Genome, 1998, 9, 1042-1048.	2.2	4
42	Identification of autosomal recessive disease loci using out-bred nuclear families. Human Mutation, 2012, 33, 338-342.	2.5	4
43	Simple and Efficient Identification of Rare Recessive Pathologically Important Sequence Variants from Next Generation Exome Sequence Data. Human Mutation, 2013, 34, 945-952.	2.5	4
44	Increased Sensitivity of Diagnostic Mutation Detection by Re-analysis Incorporating Local Reassembly of Sequence Reads. Molecular Diagnosis and Therapy, 2017, 21, 685-692.	3.8	4
45	Translating Biomarkers of Cholangiocarcinoma for Theranosis: A Systematic Review. Cancers, 2020, 12, 2817.	3.7	4
46	Detection of somatic mutations in tumors using unaligned clonal sequencing data. Laboratory Investigation, 2014, 94, 1173-1183.	3.7	2
47	Rapid Visualisation of Microarray Copy Number Data for the Detection of Structural Variations Linked to a Disease Phenotype. PLoS ONE, 2012, 7, e43466.	2.5	1
48	RNAseq Supports the Molecular Genetic Diagnosis of Late-Onset ADA Deficiency. Journal of Clinical Immunology, 2019, 39, 270-273.	3.8	0