Rachel Karchin

List of Publications by Year
in descending order

117	27,913	25014	20943
papers	citations	57 h-index	115 g-index

135	135	135	39399
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	An Integrated Genomic Analysis of Human Glioblastoma Multiforme. Science, 2008, 321, 1807-1812.	6.0	5,230
2	Core Signaling Pathways in Human Pancreatic Cancers Revealed by Global Genomic Analyses. Science, 2008, 321, 1801-1806.	6.0	3,755
3	The Genomic Landscapes of Human Breast and Colorectal Cancers. Science, 2007, 318, 1108-1113.	6.0	3,049
4	Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell, 2018, 173, 371-385.e18.	13.5	1,670
5	Accumulation of driver and passenger mutations during tumor progression. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 18545-18550.	3.3	742
6	Evolution of Neoantigen Landscape during Immune Checkpoint Blockade in Nonâ€“Small Cell Lung Cancer. Cancer Discovery, 2017, 7, 264-276.	7.7	706
7	The Genetic Landscape of the Childhood Cancer Medulloblastoma. Science, 2011, 331, 435-439.	6.0	652
8	Whole-exome sequencing of neoplastic cysts of the pancreas reveals recurrent mutations in components of ubiquitin-dependent pathways. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 21188-21193.	3.3	585
9	Exome sequencing identifies frequent inactivating mutations in BAP1, ARID1A and PBRM1 in intrahepatic cholangiocarcinomas. Nature Genetics, 2013, 45, 1470-1473.	9.4	564
10	High grade serous ovarian carcinomas originate in the fallopian tube. Nature Communications, 2017, 8, 1093.	5.8	515
11	Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. Nature Genetics, 2013, 45, 1160-1167.	9.4	513
12	Cancer-Specific High-Throughput Annotation of Somatic Mutations: Computational Prediction of Driver Missense Mutations. Cancer Research, 2009, 69, 6660-6667.	0.4	416
13	The genomic landscape of response to EGFR blockade in colorectal cancer. Nature, 2015, 526, 263-267.	13.7	398
14	A Combination of Molecular Markers and Clinical Features Improve the Classification of Pancreatic Cysts. Gastroenterology, 2015, 149, 1501-1510.	0.6	376
15	Identifying Mendelian disease genes with the Variant Effect Scoring Tool. BMC Genomics, 2013, 14, S3.	1.2	360
16	Evaluating the evaluation of cancer driver genes. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 14330-14335.	3.3	325
17	Whole Genome Sequencing Defines the Genetic Heterogeneity of Familial Pancreatic Cancer. Cancer Discovery, 2016, 6, 166-175.	7.7	282
18	Combining local-structure, fold-recognition, and new fold methods for protein structure prediction. Proteins: Structure, Function and Bioinformatics, 2003, 53, 491-496.	1.5	269

#	Article	IF	Citations
19	MODBASE: a database of annotated comparative protein structure models and associated resources. Nucleic Acids Research, 2006, 34, D291-D295.	6.5	265
20	Classifying G-protein coupled receptors with support vector machines. Bioinformatics, 2002, 18, 147-159.	1.8	261
21	Dynamics of Tumor and Immune Responses during Immune Checkpoint Blockade in Nonâ€“Small Cell Lung Cancer. Cancer Research, 2019, 79, 1214-1225.	0.4	226
22	Minimal functional driver gene heterogeneity among untreated metastases. Science, 2018, 361, 1033-1037.	6.0	223
23	Discriminative prediction of mammalian enhancers from DNA sequence. Genome Research, 2011, 21, 2167-2180.	2.4	222
24	Mutational Signature of Aristolochic Acid Exposure as Revealed by Whole-Exome Sequencing. Science Translational Medicine, 2013, 5, 197ra102.	5.8	220
25	Systematic Functional Annotation of Somatic Mutations in Cancer. Cancer Cell, 2018, 33, 450-462.e10.	7.7	213
26	Whole-exome sequencing combined with functional genomics reveals novel candidate driver cancer genes in endometrial cancer. Genome Research, 2012, 22, 2120-2129.	2.4	206
27	LS-SNP: large-scale annotation of coding non-synonymous SNPs based on multiple information sources. Bioinformatics, 2005, 21, 2814-2820.	1.8	202
28	Human bile contains MicroRNA-laden extracellular vesicles that can be used for cholangiocarcinoma diagnosis. Hepatology, 2014, 60, 896-907.	3.6	187
29	Hidden Markov models that use predicted local structure for fold recognition: Alphabets of backbone geometry. Proteins: Structure, Function and Bioinformatics, 2003, 51, 504-514.	1.5	178
30	Evaluation of liquid from the Papanicolaou test and other liquid biopsies for the detection of endometrial and ovarian cancers. Science Translational Medicine, 2018, 10, .	5.8	178
31	Computational approaches to identify functional genetic variants in cancer genomes. Nature Methods, 2013, 10, 723-729.	9.0	161
32	MODBASE, a database of annotated comparative protein structure models and associated resources. Nucleic Acids Research, 2009, 37, D347-D354.	6.5	154
33	Correlation of Somatic Mutation and Expression Identifies Genes Important in Human Glioblastoma Progression and Survival. Cancer Research, 2011, 71, 4550-4561.	0.4	148
34	Whole-genome sequencing identifies a recurrent functional synonymous mutation in melanoma. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 13481-13486.	3.3	147
35	Multimodal genomic features predict outcome of immune checkpoint blockade in non-small-cell lung cancer. Nature Cancer, 2020, 1, 99-111.	5.7	141
36	CRAVAT: cancer-related analysis of variants toolkit. Bioinformatics, 2013, 29, 647-648.	1.8	140

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37	A multimodality test to guide the management of patients with a pancreatic cyst. Science Translational Medicine, 2019, 11, .	5.8	129
38	Non-invasive detection of urothelial cancer through the analysis of driver gene mutations and aneuploidy. ELife, 2018, 7, .	2.8	118
39	CHASM and SNVBox: toolkit for detecting biologically important single nucleotide mutations in cancer. Bioinformatics, 2011, 27, 2147-2148.	1.8	116
40	Determination of Cancer Risk Associated with Germ Line BRCA1 Missense Variants by Functional Analysis. Cancer Research, 2007, 67, 1494-1501.	0.4	110
41	What is the value added by human intervention in protein structure prediction?. Proteins: Structure, Function and Bioinformatics, 2001, 45, 86-91.	1.5	104
42	IPMNs with co-occurring invasive cancers: neighbours but not always relatives. Gut, 2018, 67, 1652-1662.	6.1	104
43	Exome-Scale Discovery of Hotspot Mutation Regions in Human Cancer Using 3D Protein Structure. Cancer Research, 2016, 76, 3719-3731.	0.4	103
44	High-Throughput Prediction of MHC Class I and II Neoantigens with MHCnuggets. Cancer Immunology Research, 2020, 8, 396-408.	1.6	103
45	Next generation tools for the annotation of human SNPs. Briefings in Bioinformatics, 2009, 10, 35-52.	3.2	101
46	Assessing the Pathogenicity of Insertion and Deletion Variants with the Variant Effect Scoring Tool (VESTâ€indel). Human Mutation, 2016, 37, 28-35.	1.1	101
47	Exome Sequencing of Familial Bipolar Disorder. JAMA Psychiatry, 2016, 73, 590.	6.0	97
48	Charting the Landscape of Tandem BRCT Domainâ€“Mediated Protein Interactions. Science Signaling, 2012, 5, rs6.	1.6	88
49	CHASMplus Reveals the Scope of Somatic Missense Mutations Driving Human Cancers. Cell Systems, 2019, 9, 9-23.e8.	2.9	83
50	Intraductal Papillary Mucinous Neoplasms Arise From Multiple Independent Clones, Each With Distinct Mutations. Gastroenterology, 2019, 157, 1123-1137.e22.	0.6	82
51	Whole-Genome Sequencing of Salivary Gland Adenoid Cystic Carcinoma. Cancer Prevention Research, 2016, 9, 265-274.	0.7	80
52	A machine learning approach for somatic mutation discovery. Science Translational Medicine, 2018, 10, .	5.8	80
53	Prioritization of driver mutations in pancreatic cancer using cancer-specific high-throughput annotation of somatic mutations (CHASM). Cancer Biology and Therapy, 2010, 10, 582-587.	1.5	79
54	Using bioinformatics to predict the functional impact of SNVs. Bioinformatics, 2011, 27, 441-448.	1.8	78

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55	Genomic characterization of malignant progression in neoplastic pancreatic cysts. Nature Communications, 2020, 11, 4085.	5.8	77
56	LS-SNP/PDB: annotated non-synonymous SNPs mapped to Protein Data Bank structures. Bioinformatics, 2009, 25, 1431-1432.	1.8	68
57	MuPIT interactive: webserver for mapping variant positions to annotated, interactive 3D structures. Human Genetics, 2013, 132, 1235-1243.	1.8	68
58	Assessment of the Clinical Relevance of BRCA2 Missense Variants by Functional and Computational Approaches. American Journal of Human Genetics, 2018, 102, 233-248.	2.6	64
59	Durvalumab with platinum-pemetrexed for unresectable pleural mesothelioma: survival, genomic and immunologic analyses from the phase 2 PrE0505 trial. Nature Medicine, 2021, 27, 1910-1920.	15.2	62
60	SubClonal Hierarchy Inference from Somatic Mutations: Automatic Reconstruction of Cancer Evolutionary Trees from Multi-region Next Generation Sequencing. PLoS Computational Biology, 2015, 11, e1004416.	1.5	61
61	Evaluation of local structure alphabets based on residue burial. Proteins: Structure, Function and Bioinformatics, 2004, 55, 508-518.	1.5	58
62	Somatic mutations in the notch, NFâ€KB, PIK3CA, and hedgehog pathways in human breast cancers. Genes Chromosomes and Cancer, 2012, 51, 480-489.	1.5	58
63	Functional Impact of Missense Variants in BRCA1 Predicted by Supervised Learning. PLoS Computational Biology, 2007, 3, e26.	1.5	57
64	Integrated Informatics Analysis of Cancer-Related Variants. JCO Clinical Cancer Informatics, 2020, 4, 310-317.	1.0	57
65	Experimental Assessment of Splicing Variants Using Expression Minigenes and Comparison with In Silico Predictions. Human Mutation, 2014, 35, 1249-1259.	1.1	56
66	Integrating diverse genomic data using gene sets. Genome Biology, 2011, 12, R105.	13.9	52
67	Singleâ€cell sequencing defines genetic heterogeneity in pancreatic cancer precursor lesions. Journal of Pathology, 2019, 247, 347-356.	2.1	52
68	CRAVAT 4: Cancer-Related Analysis of Variants Toolkit. Cancer Research, 2017, 77, e35-e38.	0.4	51
69	Assessing aneuploidy with repetitive element sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 4858-4863.	3.3	50
70	Detection of aneuploidy in patients with cancer through amplification of long interspersed nucleotide elements (LINEs). Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 1871-1876.	3.3	48
71	Multiregion whole-exome sequencing of intraductal papillary mucinous neoplasms reveals frequent somatic KLF4 mutations predominantly in low-grade regions. Gut, 2021, 70, 928-939.	6.1	48
72	Mapping genetic variations to three-dimensional protein structures to enhance variant interpretation: a proposed framework. Genome Medicine, 2017, 9, 113.	3.6	47

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73	Collections of Simultaneously Altered Genes as Biomarkers of Cancer Cell Drug Response. Cancer Research, 2013, 73, 1699-1708.	0.4	46
74	Classifying Variants of Undetermined Significance in BRCA2 with Protein Likelihood Ratios. Cancer Informatics, 2008, 6, CIN.S618.	0.9	45
75	Integrative Tumor and Immune Cell Multi-omic Analyses Predict Response to Immune Checkpoint Blockade in Melanoma. Cell Reports Medicine, 2020, 1, 100139.	3.3	45
76	Cleaved NOTCH1 Expression Pattern in Head and Neck Squamous Cell Carcinoma Is Associated with NOTCH1 Mutation, HPV Status, and High-Risk Features. Cancer Prevention Research, 2015, 8, 287-295.	0.7	43
77	Proteins linked to autosomal dominant and autosomal recessive disorders harbor characteristic rare missense mutation distribution patterns. Human Molecular Genetics, 2015, 24, 5995-6002.	1.4	40
78	Calibrating E-values for hidden Markov models using reverse-sequence null models. Bioinformatics, 2005, 21, 4107-4115.	1.8	38
79	Analysis of a set of missense, frameshift, and in-frame deletion variants of BRCA1. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2009, 660, 1-11.	0.4	38
80	Network Models of TEM Î²-Lactamase Mutations Coevolving under Antibiotic Selection Show Modular Structure and Anticipate Evolutionary Trajectories. PLoS Computational Biology, 2011, 7, e1002184.	1.5	36
81	P<scp>redict</scp>-2<scp>nd</scp>: a tool for generalized protein local structure prediction. Bioinformatics, 2008, 24, 2453-2459.	1.8	34
82	A novel approach for selecting combination clinical markers of pathology applied to a large retrospective cohort of surgically resected pancreatic cysts. Journal of the American Medical Informatics Association: JAMIA, 2017, 24, 145-152.	2.2	34
83	Towards Increasing the Clinical Relevance of In Silico Methods to Predict Pathogenic Missense Variants. PLoS Computational Biology, 2016, 12, e1004725.	1.5	34
84	Predicting survival in head and neck squamous cell carcinoma from TP53 mutation. Human Genetics, 2015, 134, 497-507.	1.8	31
85	Evaluation of the Disease Liability of CFTR Variants. Methods in Molecular Biology, 2011, 742, 355-372.	0.4	28
86	Roles of DNA polymerase I in leading and lagging-strand replication defined by a high-resolution mutation footprint of ColE1 plasmid replication. Nucleic Acids Research, 2011, 39, 7020-7033.	6.5	25
87	Phenotype-optimized sequence ensembles substantially improve prediction of disease-causing mutation in cystic fibrosis. Human Mutation, 2012, 33, 1267-1274.	1.1	23
88	PIK3CA somatic mutations in breast cancer: Mechanistic insights from Langevin dynamics simulations. Proteins: Structure, Function and Bioinformatics, 2009, 75, 499-508.	1.5	21
89	Autologous reconstitution of human cancer and immune system in vivo. Oncotarget, 2017, 8, 2053-2068.	0.8	20
90	Functional hot spots in human ATPâ€binding cassette transporter nucleotide binding domains. Protein Science, 2010, 19, 2110-2121.	3.1	19

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91	A Hybrid Likelihood Model for Sequence-Based Disease Association Studies. PLoS Genetics, 2013, 9, e1003224.	1.5	19
92	Comparative Protein Structure Modeling. , 2005, , 831-860.		15
93	Matching phenotypes to whole genomes: Lessons learned from four iterations of the personal genome project community challenges. Human Mutation, 2017, 38, 1266-1276.	1.1	14
94	Characterization of genetic subclonal evolution in pancreatic cancer mouse models. Nature Communications, 2019, 10, 5435.	5.8	14
95	Exonic DNA Sequencing of ERBB4 in Bipolar Disorder. PLoS ONE, 2011, 6, e20242.	1.1	13
96	Assessment of Whole-Exome Sequence Data in Attempted Suicide within a Bipolar Disorder Cohort. Molecular Neuropsychiatry, 2017, 3, 1-11.	3.0	13
97	Detecting species-site dependencies in large multiple sequence alignments. Nucleic Acids Research, 2009, 37, 5959-5968.	6.5	12
98	Yeast two-hybrid junk sequences contain selected linear motifs. Nucleic Acids Research, 2011, 39, e128-e128.	6.5	12
99	Missense variants in CFTR nucleotide-binding domains predict quantitative phenotypes associated with cystic fibrosis disease severity. Human Molecular Genetics, 2015, 24, 1908-1917.	1.4	11
100	A Probabilistic Model to Predict Clinical Phenotypic Traits from Genome Sequencing. PLoS Computational Biology, 2014, 10, e1003825.	1.5	10
101	Bridging the Chromosome-centric and Biology/Disease-driven Human Proteome Projects: Accessible and Automated Tools for Interpreting the Biological and Pathological Impact of Protein Sequence Variants Detected via Proteogenomics. Journal of Proteome Research, 2018, 17, 4329-4336.	1.8	9
102	Pan-Cancer HLA Gene-Mediated Tumor Immunogenicity and Immune Evasion. Molecular Cancer Research, 2022, 20, 1272-1283.	1.5	9
103	Network Analysis of Protein Adaptation: Modeling the Functional Impact of Multiple Mutations. Molecular Biology and Evolution, 2018, 35, 1507-1519.	3.5	8
104	Predicting the Functional Consequences of Somatic Missense Mutations Found in Tumors. Methods in Molecular Biology, 2014, 1101, 135-159.	0.4	8
105	The Genetic Evolution of Treatment-Resistant Cutaneous, Acral, and Uveal Melanomas. Clinical Cancer Research, 2021, 27, 1516-1525.	3.2	6
106	Genome Landscapes of Disease: Strategies to Predict the Phenotypic Consequences of Human Germline and Somatic Variation. PLoS Computational Biology, 2016, 12, e1005043.	1.5	6
107	Assessing computational predictions of the phenotypic effect of cystathionineâ€betaâ€synthase variants. Human Mutation, 2019, 40, 1530-1545.	1.1	5
108	Voices in methods development. Nature Methods, 2019, 16, 945-951.	9.0	5

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109	Evaluation of Liquid From the Papanicolaou Test and Other Liquid Biopsies for the Detection of Endometrial and Ovarian Cancers. Obstetrical and Gynecological Survey, 2018, 73, 463-464.	0.2	3
110	Non-Coding Variation: The 2016 Annual Scientific Meeting of the Human Genome Variation Society. Human Mutation, 2017, 38, 460-463.	1.1	1
111	Estimation of cancer cell fractions and clone trees from multi-region sequencing of tumors. Bioinformatics, 2022, 38, 3677-3683.	1.8	1
112	Response to â€œPredictable difficulty or difficulty to predictâ€• Protein Science, 2011, 20, 4-5.	3.1	0
113	IDENTIFICATION OF ABERRANT PATHWAY AND NETWORK ACTIVITY FROM HIGH-THROUGHPUT DATA. , 2012, , .		0
114	Human genetics special issue on computational molecular medicine. Human Genetics, 2015, 134, 455-457.	1.8	0
115	Abstract 1617: Sex-specific genomic determinants of response to immunotherapy. , 2021, , .		0
116	Abstract 1662: Immunogenomic mechanisms of response and resistance to combined radiation and immunotherapy in lung cancer. , 2021, , .		0
117	Evaluation of computational tools to determine prognostic significance of TP53 mutation in head and neck squamous cell carcinoma (HNSCC).. Journal of Clinical Oncology, 2014, 32, 6035-6035.	0.8	0