Rachel Karchin

List of Publications by Year in Descending Order

115 papers	21,803 citations	54 h-index	135 g-index
135 ext. papers	25,517 ext. citations	11.9 avg, IF	5.94 L-index

#	Paper	IF	Citations
115	The Genetic Evolution of Treatment-Resistant Cutaneous, Acral, and Uveal Melanomas. *Clinical Cancer Research*, **2021**, 27, 1516-1525	12.9	2
114	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	50.5	14
113	Multiregion whole-exome sequencing of intraductal papillary mucinous neoplasms reveals frequent somatic mutations predominantly in low-grade regions. *Gut*, **2021**, 70, 928-939	19.2	14
112	High-Throughput Prediction of MHC Class I and II Neoantigens with MHCnuggets. *Cancer Immunology Research*, **2020**, 8, 396-408	12.5	38
111	Assessing aneuploidy with repetitive element sequencing. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 4858-4863	11.5	26
110	Integrated Informatics Analysis of Cancer-Related Variants. *JCO Clinical Cancer Informatics*, **2020**, 4, 310-317	5.1	17
109	Multimodal genomic features predict outcome of immune checkpoint blockade in non-small-cell lung cancer. *Nature Cancer*, **2020**, 1, 99-111	15.4	67
108	Integrative Tumor and Immune Cell Multi-omic Analyses Predict Response to Immune Checkpoint Blockade in Melanoma. *Cell Reports Medicine*, **2020**, 1, 100139	18	17
107	Genomic characterization of malignant progression in neoplastic pancreatic cysts. *Nature Communications*, **2020**, 11, 4085	17.4	27
106	Intraductal Papillary Mucinous Neoplasms Arise From Multiple Independent Clones, Each With Distinct Mutations. *Gastroenterology*, **2019**, 157, 1123-1137.e22	13.3	40
105	CHASMplus Reveals the Scope of Somatic Missense Mutations Driving Human Cancers. *Cell Systems*, **2019**, 9, 9-23.e8	10.6	47
104	Assessing computational predictions of the phenotypic effect of cystathionine-beta-synthase variants. *Human Mutation*, **2019**, 40, 1530-1545	4.7	3
103	A multimodality test to guide the management of patients with a pancreatic cyst. *Science Translational Medicine*, **2019**, 11,	17.5	71
102	Characterization of genetic subclonal evolution in pancreatic cancer mouse models. *Nature Communications*, **2019**, 10, 5435	17.4	11
101	Single-cell sequencing defines genetic heterogeneity in pancreatic cancer precursor lesions. *Journal of Pathology*, **2019**, 247, 347-356	9.4	27
100	Dynamics of Tumor and Immune Responses during Immune Checkpoint Blockade in Non-Small Cell Lung Cancer. *Cancer Research*, **2019**, 79, 1214-1225	10.1	117
99	IPMNs with co-occurring invasive cancers: neighbours but not always relatives. *Gut*, **2018**, 67, 1652-1662	19.2	58

98	Comprehensive Characterization of Cancer Driver Genes and Mutations. *Cell*, **2018**, 173, 371-385.e18	56.2	854
97	Assessment of the Clinical Relevance of BRCA2 Missense Variants by Functional and Computational Approaches. *American Journal of Human Genetics*, **2018**, 102, 233-248	11	38
96	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	11.5	30
95	Network Analysis of Protein Adaptation: Modeling the Functional Impact of Multiple Mutations. *Molecular Biology and Evolution*, **2018**, 35, 1507-1519	8.3	7
94	Evaluation of liquid from the Papanicolaou test and other liquid biopsies for the detection of endometrial and ovarian cancers. *Science Translational Medicine*, **2018**, 10,	17.5	110
93	Systematic Functional Annotation of Somatic Mutations in Cancer. *Cancer Cell*, **2018**, 33, 450-462.e10	24.3	114
92	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	5.6	6
91	A machine learning approach for somatic mutation discovery. *Science Translational Medicine*, **2018**, 10,	17.5	44
90	Minimal functional driver gene heterogeneity among untreated metastases. *Science*, **2018**, 361, 1033-1037	33.3	147
89	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	2.4	3
88	Non-invasive detection of urothelial cancer through the analysis of driver gene mutations and aneuploidy. *ELife*, **2018**, 7,	8.9	72
87	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	8.6	24
86	Non-Coding Variation: The 2016 Annual Scientific Meeting of the Human Genome Variation Society. *Human Mutation*, **2017**, 38, 460-463	4.7	1
85	Assessment of Whole-Exome Sequence Data in Attempted Suicide within a Bipolar Disorder Cohort. *Molecular Neuropsychiatry*, **2017**, 3, 1-11	4.9	11
84	Matching phenotypes to whole genomes: Lessons learned from four iterations of the personal genome project community challenges. *Human Mutation*, **2017**, 38, 1266-1276	4.7	9
83	Evolution of Neoantigen Landscape during Immune Checkpoint Blockade in Non-Small Cell Lung Cancer. *Cancer Discovery*, **2017**, 7, 264-276	24.4	491
82	High grade serous ovarian carcinomas originate in the fallopian tube. *Nature Communications*, **2017**, 8, 1093	17.4	325
81	Mapping genetic variations to three-dimensional protein structures to enhance variant interpretation: a proposed framework. *Genome Medicine*, **2017**, 9, 113	14.4	31

80	CRAVAT 4: Cancer-Related Analysis of Variants Toolkit. *Cancer Research*, **2017**, 77, e35-e38	10.1	33
79	Autologous reconstitution of human cancer and immune system in vivo. *Oncotarget*, **2017**, 8, 2053-2068	3.3	11
78	Evaluating the evaluation of cancer driver genes. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, 14330-14335	11.5	197
77	Whole Genome Sequencing Defines the Genetic Heterogeneity of Familial Pancreatic Cancer. *Cancer Discovery*, **2016**, 6, 166-75	24.4	206
76	Assessing the Pathogenicity of Insertion and Deletion Variants with the Variant Effect Scoring Tool (VEST-Indel). *Human Mutation*, **2016**, 37, 28-35	4.7	65
75	Whole-Genome Sequencing of Salivary Gland Adenoid Cystic Carcinoma. *Cancer Prevention Research*, **2016**, 9, 265-74	3.2	59
74	Towards Increasing the Clinical Relevance of In Silico Methods to Predict Pathogenic Missense Variants. *PLoS Computational Biology*, **2016**, 12, e1004725	5	26
73	Exome-Scale Discovery of Hotspot Mutation Regions in Human Cancer Using 3D Protein Structure. *Cancer Research*, **2016**, 76, 3719-31	10.1	61
72	Exome Sequencing of Familial Bipolar Disorder. *JAMA Psychiatry*, **2016**, 73, 590-7	14.5	69
71	A combination of molecular markers and clinical features improve the classification of pancreatic cysts. *Gastroenterology*, **2015**, 149, 1501-10	13.3	286
70	Missense variants in CFTR nucleotide-binding domains predict quantitative phenotypes associated with cystic fibrosis disease severity. *Human Molecular Genetics*, **2015**, 24, 1908-17	5.6	10
69	The genomic landscape of response to EGFR blockade in colorectal cancer. *Nature*, **2015**, 526, 263-7	50.4	310
68	Proteins linked to autosomal dominant and autosomal recessive disorders harbor characteristic rare missense mutation distribution patterns. *Human Molecular Genetics*, **2015**, 24, 5995-6002	5.6	24
67	Predicting survival in head and neck squamous cell carcinoma from TP53 mutation. *Human Genetics*, **2015**, 134, 497-507	6.3	23
66	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, 287-95	3.2	35
65	SubClonal Hierarchy Inference from Somatic Mutations: Automatic Reconstruction of Cancer Evolutionary Trees from Multi-region Next Generation Sequencing. *PLoS Computational Biology*, **2015**, 11, e1004416	5	47
64	Experimental assessment of splicing variants using expression minigenes and comparison with in silico predictions. *Human Mutation*, **2014**, 35, 1249-59	4.7	41
63	A probabilistic model to predict clinical phenotypic traits from genome sequencing. *PLoS Computational Biology*, **2014**, 10, e1003825	5	8

62	Human bile contains microRNA-laden extracellular vesicles that can be used for cholangiocarcinoma diagnosis. *Hepatology*, **2014**, 60, 896-907	11.2	139
61	Predicting the functional consequences of somatic missense mutations found in tumors. *Methods in Molecular Biology*, **2014**, 1101, 135-59	1.4	8
60	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	2.2	
59	Identifying Mendelian disease genes with the variant effect scoring tool. *BMC Genomics*, **2013**, 14 Suppl 3, S3	4.5	240
58	Computational approaches to identify functional genetic variants in cancer genomes. *Nature Methods*, **2013**, 10, 723-9	21.6	129
57	Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. *Nature Genetics*, **2013**, 45, 1160-7	36.3	413
56	MuPIT interactive: webserver for mapping variant positions to annotated, interactive 3D structures. *Human Genetics*, **2013**, 132, 1235-43	6.3	56
55	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, 13481-6	11.5	127
54	Exome sequencing identifies frequent inactivating mutations in BAP1, ARID1A and PBRM1 in intrahepatic cholangiocarcinomas. *Nature Genetics*, **2013**, 45, 1470-1473	36.3	464
53	Mutational signature of aristolochic acid exposure as revealed by whole-exome sequencing. *Science Translational Medicine*, **2013**, 5, 197ra102	17.5	178
52	Collections of simultaneously altered genes as biomarkers of cancer cell drug response. *Cancer Research*, **2013**, 73, 1699-708	10.1	34
51	CRAVAT: cancer-related analysis of variants toolkit. *Bioinformatics*, **2013**, 29, 647-8	7.2	98
50	A hybrid likelihood model for sequence-based disease association studies. *PLoS Genetics*, **2013**, 9, e1003224	8	16
49	Somatic mutations in the Notch, NF-KB, PIK3CA, and Hedgehog pathways in human breast cancers. *Genes Chromosomes and Cancer*, **2012**, 51, 480-9	5	50
48	Whole-exome sequencing combined with functional genomics reveals novel candidate driver cancer genes in endometrial cancer. *Genome Research*, **2012**, 22, 2120-9	9.7	178
47	Charting the landscape of tandem BRCT domain-mediated protein interactions. *Science Signaling*, **2012**, 5, rs6	8.8	74
46	Phenotype-optimized sequence ensembles substantially improve prediction of disease-causing mutation in cystic fibrosis. *Human Mutation*, **2012**, 33, 1267-74	4.7	17
45	Network models of TEM β-lactamase mutations coevolving under antibiotic selection show modular structure and anticipate evolutionary trajectories. *PLoS Computational Biology*, **2011**, 7, e1002184	5	28

44	The genetic landscape of the childhood cancer medulloblastoma. *Science*, **2011**, 331, 435-9	33.3	576
43	Evaluation of the disease liability of CFTR variants. *Methods in Molecular Biology*, **2011**, 742, 355-72	1.4	21
42	Integrating diverse genomic data using gene sets. *Genome Biology*, **2011**, 12, R105	18.3	45
41	Exonic DNA sequencing of ERBB4 in bipolar disorder. *PLoS ONE*, **2011**, 6, e20242	3.7	12
40	Response to Predictable difficulty or difficulty to predict *Protein Science*, **2011**, 20, 4-5	6.3	78
39	Yeast two-hybrid junk sequences contain selected linear motifs. *Nucleic Acids Research*, **2011**, 39, e128	20.1	9
38	Correlation of somatic mutation and expression identifies genes important in human glioblastoma progression and survival. *Cancer Research*, **2011**, 71, 4550-61	10.1	122
37	Using bioinformatics to predict the functional impact of SNVs. *Bioinformatics*, **2011**, 27, 441-8	7.2	65
36	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, 21188-93	11.5	484
35	Discriminative prediction of mammalian enhancers from DNA sequence. *Genome Research*, **2011**, 21, 2167-80	9.7	168
34	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, 7020-33	20.1	20
33	CHASM and SNVBox: toolkit for detecting biologically important single nucleotide mutations in cancer. *Bioinformatics*, **2011**, 27, 2147-8	7.2	86
32	Prioritization of driver mutations in pancreatic cancer using cancer-specific high-throughput annotation of somatic mutations (CHASM). *Cancer Biology and Therapy*, **2010**, 10, 582-7	4.6	70
31	Accumulation of driver and passenger mutations during tumor progression. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 18545-50	11.5	574
30	Functional hot spots in human ATP-binding cassette transporter nucleotide binding domains. *Protein Science*, **2010**, 19, 2110-21	6.3	15
29	LS-SNP/PDB: annotated non-synonymous SNPs mapped to Protein Data Bank structures. *Bioinformatics*, **2009**, 25, 1431-2	7.2	64
28	MODBASE, a database of annotated comparative protein structure models and associated resources. *Nucleic Acids Research*, **2009**, 37, D347-54	20.1	143
27	Detecting species-site dependencies in large multiple sequence alignments. *Nucleic Acids Research*, **2009**, 37, 5959-68	20.1	12

26	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	3.3	31
25	PIK3CA somatic mutations in breast cancer: Mechanistic insights from Langevin dynamics simulations. *Proteins: Structure, Function and Bioinformatics*, **2009**, 75, 499-508	4.2	16
24	Next generation tools for the annotation of human SNPs. *Briefings in Bioinformatics*, **2009**, 10, 35-52	13.4	91
23	Cancer-specific high-throughput annotation of somatic mutations: computational prediction of driver missense mutations. *Cancer Research*, **2009**, 69, 6660-7	10.1	344
22	Core signaling pathways in human pancreatic cancers revealed by global genomic analyses. *Science*, **2008**, 321, 1801-6	33.3	3223
21	PREDICT-2ND: a tool for generalized protein local structure prediction. *Bioinformatics*, **2008**, 24, 2453-9	7.2	31
20	Classifying Variants of Undetermined Significance in BRCA2 with protein likelihood ratios. *Cancer Informatics*, **2008**, 6, 203-16	2.4	40
19	An integrated genomic analysis of human glioblastoma multiforme. *Science*, **2008**, 321, 1807-12	33.3	4419
18	The genomic landscapes of human breast and colorectal cancers. *Science*, **2007**, 318, 1108-13	33.3	2717
17	Functional impact of missense variants in BRCA1 predicted by supervised learning. *PLoS Computational Biology*, **2007**, 3, e26	5	47
16	Determination of cancer risk associated with germ line BRCA1 missense variants by functional analysis. *Cancer Research*, **2007**, 67, 1494-501	10.1	98
15	MODBASE: a database of annotated comparative protein structure models and associated resources. *Nucleic Acids Research*, **2006**, 34, D291-5	20.1	237
14	Comparative Protein Structure Modeling **2005**, 831-860		11
13	LS-SNP: large-scale annotation of coding non-synonymous SNPs based on multiple information sources. *Bioinformatics*, **2005**, 21, 2814-20	7.2	193
12	Calibrating E-values for hidden Markov models using reverse-sequence null models. *Bioinformatics*, **2005**, 21, 4107-15	7.2	36
11	Evaluation of local structure alphabets based on residue burial. *Proteins: Structure, Function and Bioinformatics*, **2004**, 55, 508-18	4.2	51
10	Hidden Markov models that use predicted local structure for fold recognition: alphabets of backbone geometry. *Proteins: Structure, Function and Bioinformatics*, **2003**, 51, 504-14	4.2	148
9	Combining local-structure, fold-recognition, and new fold methods for protein structure prediction. *Proteins: Structure, Function and Bioinformatics*, **2003**, 53 Suppl 6, 491-6	4.2	246

8	Classifying G-protein coupled receptors with support vector machines. *Bioinformatics*, **2002**, 18, 147-59	7.2	232
7	What is the value added by human intervention in protein structure prediction?. *Proteins: Structure, Function and Bioinformatics*, **2001**, Suppl 5, 86-91	4.2	95
6	Evaluation of machine learning methods to predict peptide binding to MHC Class I proteins		18
5	Non-invasive detection of bladder cancer through the analysis of driver gene mutations and aneuploidy		4
4	CHASMplus reveals the scope of somatic missense mutations driving human cancers		3
3	High-throughput prediction of MHC Class I and Class II neoantigens with MHCnuggets		2
2	OpenCRAVAT, an open source collaborative platform for the annotation of human genetic variation		1
1	Evaluating the Evaluation of Cancer Driver Genes		1