Sumit Middha

List of Publications by Citations

81 papers	8,077 citations	39 h-index	86 g-index
86 ext. papers	10,511 ext. citations	8.7 avg, IF	5.14 L-index

#	Paper	IF	Citations
81	Mutational landscape of metastatic cancer revealed from prospective clinical sequencing of 10,000 patients. *Nature Medicine*, **2017**, 23, 703-713	50.5	1638
80	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. *American Journal of Human Genetics*, **2016**, 99, 877-885	11	722
79	The Genomic Landscape of Endocrine-Resistant Advanced Breast Cancers. *Cancer Cell*, **2018**, 34, 427-438.e6	24.3	339
78	Clinical Sequencing Defines the Genomic Landscape of Metastatic Colorectal Cancer. *Cancer Cell*, **2018**, 33, 125-136.e3	24.3	338
77	Whole-genome sequencing of multiple myeloma from diagnosis to plasma cell leukemia reveals genomic initiating events, evolution, and clonal tides. *Blood*, **2012**, 120, 1060-6	2.2	310
76	Mutations in DNMT1 cause hereditary sensory neuropathy with dementia and hearing loss. *Nature Genetics*, **2011**, 43, 595-600	36.3	284
75	Analysis of the Prevalence of Microsatellite Instability in Prostate Cancer and Response to Immune Checkpoint Blockade. *JAMA Oncology*, **2019**, 5, 471-478	13.4	257
74	Integrated genomic characterization reveals novel, therapeutically relevant drug targets in FGFR and EGFR pathways in sporadic intrahepatic cholangiocarcinoma. *PLoS Genetics*, **2014**, 10, e1004135	6	239
73	Genetic diversity of tumors with mismatch repair deficiency influences anti-PD-1 immunotherapy response. *Science*, **2019**, 364, 485-491	33.3	228
72	Microsatellite Instability Is Associated With the Presence of Lynch Syndrome Pan-Cancer. *Journal of Clinical Oncology*, **2019**, 37, 286-295	2.2	203
71	MAP-RSeq: Mayo Analysis Pipeline for RNA sequencing. *BMC Bioinformatics*, **2014**, 15, 224	3.6	191
70	Genetic Predictors of Response to Systemic Therapy in Esophagogastric Cancer. *Cancer Discovery*, **2018**, 8, 49-58	24.4	180
69	The human gut microbiome: current knowledge, challenges, and future directions. *Translational Research*, **2012**, 160, 246-57	11	178
68	Brain expression genome-wide association study (eGWAS) identifies human disease-associated variants. *PLoS Genetics*, **2012**, 8, e1002707	6	174
67	Reliable Detection of Mismatch Repair Deficiency in Colorectal Cancers Using Mutational Load in Next-Generation Sequencing Panels. *Journal of Clinical Oncology*, **2016**, 34, 2141-7	2.2	170
66	3Qtag digital gene expression profiling of human brain and universal reference RNA using Illumina Genome Analyzer. *BMC Genomics*, **2009**, 10, 531	4.5	132
65	Reliable Pan-Cancer Microsatellite Instability Assessment by Using Targeted Next-Generation Sequencing Data. *JCO Precision Oncology*, **2017**, 2017,	3.6	128

64	Novel late-onset Alzheimer disease loci variants associate with brain gene expression. *Neurology*, **2012**, 79, 221-8	6.5	124
63	CAP-miRSeq: a comprehensive analysis pipeline for microRNA sequencing data. *BMC Genomics*, **2014**, 15, 423	4.5	118
62	The genetic landscape of endometrial clear cell carcinomas. *Journal of Pathology*, **2017**, 243, 230-241	9.4	116
61	New DNA Methylation Markers for Pancreatic Cancer: Discovery, Tissue Validation, and Pilot Testing in Pancreatic Juice. *Clinical Cancer Research*, **2015**, 21, 4473-81	12.9	82
60	A novel bioinformatics pipeline for identification and characterization of fusion transcripts in breast cancer and normal cell lines. *Nucleic Acids Research*, **2011**, 39, e100	20.1	82
59	Exome sequencing and systems biology converge to identify novel mutations in the L-type calcium channel, CACNA1C, linked to autosomal dominant long QT syndrome. *Circulation: Cardiovascular Genetics*, **2013**, 6, 279-89		80
58	Novel High-grade Endometrial Stromal Sarcoma: A Morphologic Mimicker of Myxoid Leiomyosarcoma. *American Journal of Surgical Pathology*, **2017**, 41, 12-24	6.7	79
57	Multi-platform analysis of microRNA expression measurements in RNA from fresh frozen and FFPE tissues. *PLoS ONE*, **2013**, 8, e52517	3.7	79
56	Homozygous/Compound Heterozygous Triadin Mutations Associated With Autosomal-Recessive Long-QT Syndrome and Pediatric Sudden Cardiac Arrest: Elucidation of the Triadin Knockout Syndrome. *Circulation*, **2015**, 131, 2051-60	16.7	74
55	Colorectal Carcinomas Containing Hypermethylated MLH1 Promoter and Wild-Type BRAF/KRAS Are Enriched for Targetable Kinase Fusions. *Cancer Research*, **2019**, 79, 1047-1053	10.1	73
54	Histone deacetylase inhibition promotes osteoblast maturation by altering the histone H4 epigenome and reduces Akt phosphorylation. *Journal of Biological Chemistry*, **2013**, 288, 28783-91	5.4	65
53	Genomic Alterations in Fatal Forms of Non-Anaplastic Thyroid Cancer: Identification of and as Novel Thyroid Cancer Genes Associated with Tumor Virulence. *Clinical Cancer Research*, **2017**, 23, 5970-5980	12.9	64
52	Clinical Utility of Prospective Molecular Characterization in Advanced Endometrial Cancer. *Clinical Cancer Research*, **2018**, 24, 5939-5947	12.9	60
51	Glutathione S-transferase omega genes in Alzheimer and Parkinson disease risk, age-at-diagnosis and brain gene expression: an association study with mechanistic implications. *Molecular Neurodegeneration*, **2012**, 7, 13	19	57
50	Optimizing Workflows and Processing of Cytologic Samples for Comprehensive Analysis by Next-Generation Sequencing: Memorial Sloan Kettering Cancer Center Experience. *Archives of Pathology and Laboratory Medicine*, **2016**, 140, 1200-1205	5	55
49	TREAT: a bioinformatics tool for variant annotations and visualizations in targeted and exome sequencing data. *Bioinformatics*, **2012**, 28, 277-8	7.2	50
48	Identification of candidate genes for prostate cancer-risk SNPs utilizing a normal prostate tissue eQTL data set. *Nature Communications*, **2015**, 6, 8653	17.4	49
47	Novel TRAF1-ALK fusion identified by deep RNA sequencing of anaplastic large cell lymphoma. *Genes Chromosomes and Cancer*, **2013**, 52, 1097-102	5	45

46	Morphological characterization of colorectal cancers in The Cancer Genome Atlas reveals distinct morphology-molecular associations: clinical and biological implications. *Modern Pathology*, **2017**, 30, 599-609	9.8	43
45	SAAP-RRBS: streamlined analysis and annotation pipeline for reduced representation bisulfite sequencing. *Bioinformatics*, **2012**, 28, 2180-1	7.2	40
44	Clinical Genomic Sequencing of Pediatric and Adult Osteosarcoma Reveals Distinct Molecular Subsets with Potentially Targetable Alterations. *Clinical Cancer Research*, **2019**, 25, 6346-6356	12.9	39
43	HiChIP: a high-throughput pipeline for integrative analysis of ChIP-Seq data. *BMC Bioinformatics*, **2014**, 15, 280	3.6	38
42	Application of whole exome sequencing in undiagnosed inherited polyneuropathies. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, 1265-72	5.5	38
41	Gene networks in Drosophila melanogaster: integrating experimental data to predict gene function. *Genome Biology*, **2009**, 10, R97	18.3	38
40	Mitochondrial Aging and Physical Decline: Insights From Three Generations of Women. *Journals of Gerontology - Series A Biological Sciences and Medical Sciences*, **2015**, 70, 1409-17	6.4	36
39	Identification of novel variants in colorectal cancer families by high-throughput exome sequencing. *Cancer Epidemiology Biomarkers and Prevention*, **2013**, 22, 1239-51	4	36
38	Evaluation of oligonucleotide sequence capture arrays and comparison of next-generation sequencing platforms for use in molecular diagnostics. *Clinical Chemistry*, **2010**, 56, 1297-306	5.5	36
37	Identification and characterization of a novel promoter for the human ANO1 gene regulated by the transcription factor signal transducer and activator of transcription 6 (STAT6). *FASEB Journal*, **2015**, 29, 152-63	0.9	35
36	Identification of Targetable Kinase Alterations in Patients with Colorectal Carcinoma That are Preferentially Associated with Wild-Type RAS/RAF. *Molecular Cancer Research*, **2016**, 14, 296-301	6.6	34
35	Research resource: whole transcriptome RNA sequencing detects multiple 1,25-dihydroxyvitamin D(3)-sensitive metabolic pathways in developing zebrafish. *Molecular Endocrinology*, **2012**, 26, 1630-42		33
34	Majority of -Mutant and -Deficient Colorectal Carcinomas Achieve Clinical Benefit From Immune Checkpoint Inhibitor Therapy and Are Microsatellite Instability-High. *JCO Precision Oncology*, **2019**, 3,	3.6	32
33	Evolution and potential function of fibrinogen-like domains across twelve Drosophila species. *BMC Genomics*, **2008**, 9, 260	4.5	30
32	Characterizing genetic variation of adrenergic signalling pathways in Takotsubo (stress) cardiomyopathy exomes. *European Journal of Heart Failure*, **2014**, 16, 942-9	12.3	28
31	Comprehensively evaluating cis-regulatory variation in the human prostate transcriptome by using gene-level allele-specific expression. *American Journal of Human Genetics*, **2015**, 96, 869-82	11	27
30	Genome-wide characterization of transcriptional patterns in high and low antibody responders to rubella vaccination. *PLoS ONE*, **2013**, 8, e62149	3.7	26
29	TNNI3K mutation in familial syndrome of conduction system disease, atrial tachyarrhythmia and dilated cardiomyopathy. *Human Molecular Genetics*, **2014**, 23, 5793-804	5.6	25

28	Identification of prognostic molecular biomarkers in 157 HPV-positive and HPV-negative squamous cell carcinomas of the oropharynx. *International Journal of Cancer*, **2019**, 145, 3152-3162	7.5	24
27	amplification is a mechanism of acquired resistance to EGFR inhibitors identified by transposon mutagenesis and clinical genomics. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, E6030-E6038	11.5	24
26	Comprehensive assessment of genetic variants within TCF4 in FuchsQendothelial corneal dystrophy **2014**, 55, 6101-7		24
25	Retained mismatch repair protein expression occurs in approximately 6% of microsatellite instability-high cancers and is associated with missense mutations in mismatch repair genes. *Modern Pathology*, **2020**, 33, 871-879	9.8	23
24	Outcome and molecular characteristics of non-invasive encapsulated follicular variant of papillary thyroid carcinoma with oncocytic features. *Endocrine*, **2019**, 64, 97-108	4	20
23	Targeted sequencing of 36 known or putative colorectal cancer susceptibility genes. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 553-569	2.3	20
22	Ubiquitin ligase defect by DCAF8 mutation causes HMSN2 with giant axons. *Neurology*, **2014**, 82, 873-8	6.5	20
21	Recurrent, truncating SOX9 mutations are associated with SOX9 overexpression, KRAS mutation, and TP53 wild type status in colorectal carcinoma. *Oncotarget*, **2016**, 7, 50875-50882	3.3	15
20	Universal screening for microsatellite instability in colorectal cancer in the clinical genomics era: new recommendations, methods, and considerations. *Familial Cancer*, **2017**, 16, 525-529	3	14
19	Mutational landscape of candidate genes in familial prostate cancer. *Prostate*, **2014**, 74, 1371-8	4.2	13
18	Whole genome analyses of a well-differentiated liposarcoma reveals novel SYT1 and DDR2 rearrangements. *PLoS ONE*, **2014**, 9, e87113	3.7	13
17	Chromosome 20q Amplification Defines a Subtype of Microsatellite Stable, Left-Sided Colon Cancers with Wild-type RAS/RAF and Better Overall Survival. *Molecular Cancer Research*, **2017**, 15, 708-713	6.6	11
16	A FISH assay efficiently screens for BRAF gene rearrangements in pancreatic acinar-type neoplasms. *Modern Pathology*, **2018**, 31, 132-140	9.8	9
15	Whole exome sequencing in 75 high-risk families with validation and replication in independent case-control studies identifies TANGO2, OR5H14, and CHAD as new prostate cancer susceptibility genes. *Oncotarget*, **2017**, 8, 1495-1507	3.3	8
14	How well do whole exome sequencing results correlate with medical findings? A study of 89 Mayo Clinic Biobank samples. *Frontiers in Genetics*, **2015**, 6, 244	4.5	8
13	Somatic HNF1A mutations in the malignant transformation of hepatocellular adenomas: a retrospective analysis of data from MSK-IMPACT and TCGA. *Human Pathology*, **2019**, 83, 1-6	3.7	7
12	Novel splice site mutations in the gamma glutamyl carboxylase gene in a child with congenital combined deficiency of the vitamin K-dependent coagulation factors (VKCFD). *Pediatric Blood and Cancer*, **2009**, 53, 92-5	3	6
11	Comparison of the Life Cycles of Genetically Distant Species C and Species D Human Adenoviruses Ad6 and Ad26 in Human Cells. *Journal of Virology*, **2015**, 89, 12401-17	6.6	5

10	Gene expression patterns in CD4+ peripheral blood cells in healthy subjects and stage IV melanoma patients. *Cancer Immunology, Immunotherapy*, **2015**, 64, 1437-47	7.4	4
9	Gene Expression Signatures Characterized by Longitudinal Stability and Interindividual Variability Delineate Baseline Phenotypic Groups with Distinct Responses to Immune Stimulation. *Journal of Immunology*, **2018**, 200, 1917-1928	5.3	4
8	A Pan-Cancer Study of Somatic TERT Promoter Mutations and Amplification in 30,773 Tumors Profiled by Clinical Genomic Sequencing. *Journal of Molecular Diagnostics*, **2021**, 23, 253-263	5.1	4
7	769 Novel Methylated DNA Markers Predict Site of Gastrointestinal Cancer. *Gastroenterology*, **2013** , 144, S-84	13.3	3
6	Novel SOD1 mutation discovered in atypical ALS by whole exome sequencing. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, 943-4	5.5	3
5	From days to hours: reporting clinically actionable variants from whole genome sequencing. *PLoS ONE*, **2014**, 9, e86803	3.7	3
4	YES1amplification: a mechanism of acquired resistance to EGFR inhibitors identified by transposon mutagenesis and clinical genomics		1
3	Clinical Experience of Cerebrospinal Fluid-Based Liquid Biopsy Demonstrates Superiority of Cell-Free DNA over Cell Pellet Genomic DNA for Molecular Profiling. *Journal of Molecular Diagnostics*, **2021**, 23, 742-752	5.1	1
2	Role of Bioinformatics in Molecular Medicine **2020**, 55-68		
1	Identification of Venous Thromboembolism (VTE)-Associated Novel Variants in the ABO Gene Using Targeted Deep Sequencing. *Blood*, **2011**, 118, 709-709	2.2	