Sumit Middha

List of Publications by Year
in descending order

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81 papers	12,214 citations	61984 43 h-index	64796 79 g-index
86 all docs	86 docs citations	86 times ranked	26029 citing authors

#	Article	IF	Citations
1	Mutational landscape of metastatic cancer revealed from prospective clinical sequencing of 10,000 patients. Nature Medicine, 2017, 23, 703-713.	30.7	2,473
2	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. American Journal of Human Genetics, 2016, 99, 877-885.	6.2	1,555
3	The Genomic Landscape of Endocrine-Resistant Advanced Breast Cancers. Cancer Cell, 2018, 34, 427-438.e6.	16.8	633
4	Clinical Sequencing Defines the Genomic Landscape of Metastatic Colorectal Cancer. Cancer Cell, 2018, 33, 125-136.e3.	16.8	589
5	Analysis of the Prevalence of Microsatellite Instability in Prostate Cancer and Response to Immune Checkpoint Blockade. JAMA Oncology, 2019, 5, 471.	7.1	426
6	Microsatellite Instability Is Associated With the Presence of Lynch Syndrome Pan-Cancer. Journal of Clinical Oncology, 2019, 37, 286-295.	1.6	397
7	Genetic diversity of tumors with mismatch repair deficiency influences antiâ€“PD-1 immunotherapy response. Science, 2019, 364, 485-491.	12.6	395
8	Whole-genome sequencing of multiple myeloma from diagnosis to plasma cell leukemia reveals genomic initiating events, evolution, and clonal tides. Blood, 2012, 120, 1060-1066.	1.4	357
9	Mutations in DNMT1 cause hereditary sensory neuropathy with dementia and hearing loss. Nature Genetics, 2011, 43, 595-600.	21.4	342
10	Integrated Genomic Characterization Reveals Novel, Therapeutically Relevant Drug Targets in FGFR and EGFR Pathways in Sporadic Intrahepatic Cholangiocarcinoma. PLoS Genetics, 2014, 10, e1004135.	3.5	292
11	MAP-RSeq: Mayo Analysis Pipeline for RNA sequencing. BMC Bioinformatics, 2014, 15, 224.	2.6	284
12	Genetic Predictors of Response to Systemic Therapy in Esophagogastric Cancer. Cancer Discovery, 2018, 8, 49-58.	9.4	275
13	The human gut microbiome: current knowledge, challenges, and future directions. Translational Research, 2012, 160, 246-257.	5.0	249
14	Brain Expression Genome-Wide Association Study (eGWAS) Identifies Human Disease-Associated Variants. PLoS Genetics, 2012, 8, e1002707.	3.5	225
15	Reliable Pan-Cancer Microsatellite Instability Assessment by Using Targeted Next-Generation Sequencing Data. JCO Precision Oncology, 2017, 2017, 1-17.	3.0	209
16	Reliable Detection of Mismatch Repair Deficiency in Colorectal Cancers Using Mutational Load in Next-Generation Sequencing Panels. Journal of Clinical Oncology, 2016, 34, 2141-2147.	1.6	204
17	The genetic landscape of endometrial clear cell carcinomas. Journal of Pathology, 2017, 243, 230-241.	4.5	168
18	3' tag digital gene expression profiling of human brain and universal reference RNA using Illumina Genome Analyzer. BMC Genomics, 2009, 10, 531.	2.8	151

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19	Novel late-onset Alzheimer disease loci variants associate with brain gene expression. Neurology, 2012, 79, 221-228.	1.1	144
20	CAP-miRSeq: a comprehensive analysis pipeline for microRNA sequencing data. BMC Genomics, 2014, 15, 423.	2.8	138
21	Novel High-grade Endometrial Stromal Sarcoma. American Journal of Surgical Pathology, 2017, 41, 12-24.	3.7	115
22	Colorectal Carcinomas Containing Hypermethylated MLH1 Promoter and Wild-Type BRAF/KRAS Are Enriched for Targetable Kinase Fusions. Cancer Research, 2019, 79, 1047-1053.	0.9	112
23	New DNA Methylation Markers for Pancreatic Cancer: Discovery, Tissue Validation, and Pilot Testing in Pancreatic Juice. Clinical Cancer Research, 2015, 21, 4473-4481.	7.0	108
24	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, 279-289.	5.1	102
25	Clinical Utility of Prospective Molecular Characterization in Advanced Endometrial Cancer. Clinical Cancer Research, 2018, 24, 5939-5947.	7.0	100
26	Multi-Platform Analysis of MicroRNA Expression Measurements in RNA from Fresh Frozen and FFPE Tissues. PLoS ONE, 2013, 8, e52517.	2.5	99
27	A novel bioinformatics pipeline for identification and characterization of fusion transcripts in breast cancer and normal cell lines. Nucleic Acids Research, 2011, 39, e100-e100.	14.5	94
28	Homozygous/Compound Heterozygous Triadin Mutations Associated With Autosomal-Recessive Long-QT Syndrome and Pediatric Sudden Cardiac Arrest. Circulation, 2015, 131, 2051-2060.	1.6	92
29	Genomic Alterations in Fatal Forms of Non-Anaplastic Thyroid Cancer: Identification of MED12 and RBM10 as Novel Thyroid Cancer Genes Associated with Tumor Virulence. Clinical Cancer Research, 2017, 23, 5970-5980.	7.0	89
30	Histone Deacetylase Inhibition Promotes Osteoblast Maturation by Altering the Histone H4 Epigenome and Reduces Akt Phosphorylation. Journal of Biological Chemistry, 2013, 288, 28783-28791.	3.4	78
31	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.	10.8	75
32	Clinical Genomic Sequencing of Pediatric and Adult Osteosarcoma Reveals Distinct Molecular Subsets with Potentially Targetable Alterations. Clinical Cancer Research, 2019, 25, 6346-6356.	7.0	75
33	Morphological characterization of colorectal cancers in The Cancer Genome Atlas reveals distinct morphologyâ€“molecular associations: clinical and biological implications. Modern Pathology, 2017, 30, 599-609.	5.5	74
34	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.	2.5	72
35	Identification of candidate genes for prostate cancer-risk SNPs utilizing a normal prostate tissue eQTL data set. Nature Communications, 2015, 6, 8653.	12.8	65
36	Majority of B2M-Mutant and -Deficient Colorectal Carcinomas Achieve Clinical Benefit From Immune Checkpoint Inhibitor Therapy and Are Microsatellite Instability-High. JCO Precision Oncology, 2019, 3, 1-14.	3.0	61

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37	TREAT: a bioinformatics tool for variant annotations and visualizations in targeted and exome sequencing data. Bioinformatics, 2012, 28, 277-278.	4.1	59
38	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.	5.5	58
39	HiChIP: a high-throughput pipeline for integrative analysis of ChIP-Seq data. BMC Bioinformatics, 2014, 15, 280.	2.6	55
40	Novel TRAF1â€ALK fusion identified by deep RNA sequencing of anaplastic large cell lymphoma. Genes Chromosomes and Cancer, 2013, 52, 1097-1102.	2.8	51
41	SAAP-RRBS: streamlined analysis and annotation pipeline for reduced representation bisulfite sequencing. Bioinformatics, 2012, 28, 2180-2181.	4.1	49
42	Identification of prognostic molecular biomarkers in 157 HPVâ€positive and HPVâ€negative squamous cell carcinomas of the oropharynx. International Journal of Cancer, 2019, 145, 3152-3162.	5.1	48
43	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.	3.4	46
44	Gene networks in Drosophila melanogaster: integrating experimental data to predict gene function. Genome Biology, 2009, 10, R97.	9.6	44
45	YES1 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.	7.1	44
46	Application of whole exome sequencing in undiagnosed inherited polyneuropathies. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1265-1272.	1.9	43
47	Mitochondrial Aging and Physical Decline: Insights From Three Generations of Women. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2015, 70, 1409-1417.	3.6	43
48	Evaluation of Oligonucleotide Sequence Capture Arrays and Comparison of Next-Generation Sequencing Platforms for Use in Molecular Diagnostics. Clinical Chemistry, 2010, 56, 1297-1306.	3.2	41
49	Research Resource: Whole Transcriptome RNA Sequencing Detects Multiple 1Î±,25-Dihydroxyvitamin D3-Sensitive Metabolic Pathways in Developing Zebrafish. Molecular Endocrinology, 2012, 26, 1630-1642.	3.7	41
50	TNNI3K mutation in familial syndrome of conduction system disease, atrial tachyarrhythmia and dilated cardiomyopathy. Human Molecular Genetics, 2014, 23, 5793-5804.	2.9	41
51	Identification of Novel Variants in Colorectal Cancer Families by High-Throughput Exome Sequencing. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 1239-1251.	2.5	37
52	Characterizing genetic variation of adrenergic signalling pathways in Takotsubo (stress) cardiomyopathy exomes. European Journal of Heart Failure, 2014, 16, 942-949.	7.1	37
53	Comprehensively Evaluating cis -Regulatory Variation in the Human Prostate Transcriptome by Using Gene-Level Allele-Specific Expression. American Journal of Human Genetics, 2015, 96, 869-882.	6.2	37
54	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, 152-163.	0.5	37

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55	Evolution and potential function of fibrinogen-like domains across twelve Drosophila species. BMC Genomics, 2008, 9, 260.	2.8	36
56	Outcome and molecular characteristics of non-invasive encapsulated follicular variant of papillary thyroid carcinoma with oncocytic features. Endocrine, 2019, 64, 97-108.	2.3	35
57	Genome-Wide Characterization of Transcriptional Patterns in High and Low Antibody Responders to Rubella Vaccination. PLoS ONE, 2013, 8, e62149.	2.5	33
58	Targeted sequencing of 36 known or putative colorectal cancer susceptibility genes. Molecular Genetics & Genomic Medicine, 2017, 5, 553-569.	1.2	32
59	Comprehensive Assessment of Genetic Variants WithinTCF4in Fuchs' Endothelial Corneal Dystrophy. , 2014, 55, 6101.		31
60	Ubiquitin ligase defect by DCAF8 mutation causes HMSN2 with giant axons. Neurology, 2014, 82, 873-878.	1.1	28
61	Recurrent, truncating SOX9 mutations are associated with SOX9 overexpression, KRAS mutation, and TP53 wild type status in colorectal carcinoma. Oncotarget, 2016, 7, 50875-50882.	1.8	26
62	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.	3.4	24
63	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.	2.8	20
64	Universal screening for microsatellite instability in colorectal cancer in the clinical genomics era: new recommendations, methods, and considerations. Familial Cancer, 2017, 16, 525-529.	1.9	18
65	A FISH assay efficiently screens for BRAF gene rearrangements in pancreatic acinar-type neoplasms. Modern Pathology, 2018, 31, 132-140.	5.5	17
66	Clinical Experience of Cerebrospinal Fluidâ€“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.	2.8	17
67	Mutational landscape of candidate genes in familial prostate cancer. Prostate, 2014, 74, 1371-1378.	2.3	16
68	Whole Genome Analyses of a Well-Differentiated Liposarcoma Reveals Novel SYT1 and DDR2 Rearrangements. PLoS ONE, 2014, 9, e87113.	2.5	14
69	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.	2.0	14
70	Defining Novel DNA Virus-Tumor Associations and Genomic Correlates Using Prospective Clinical Tumor/Normal Matched Sequencing Data. Journal of Molecular Diagnostics, 2022, 24, 515-528.	2.8	12
71	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.	1.8	11
72	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.	2.3	9

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73	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, 12401-12417.	3.4	8
74	Novel splice site mutations in the gamma glutamyl carboxylase gene in a child with congenital combined deficiency of the vitamin Kâ€dependent coagulation factors (VKCFD). Pediatric Blood and Cancer, 2009, 53, 92-95.	1.5	6
75	Novel SOD1 mutation discovered in atypical ALS by whole exome sequencing. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 943-944.	1.9	5
76	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, ji1701099.	0.8	5
77	769 Novel Methylated DNA Markers Predict Site of Gastrointestinal Cancer. Gastroenterology, 2013, 144, S-84.	1.3	4
78	Gene expression patterns in CD4+ peripheral blood cells in healthy subjects and stage IV melanoma patients. Cancer Immunology, Immunotherapy, 2015, 64, 1437-1447.	4.2	4
79	From Days to Hours: Reporting Clinically Actionable Variants from Whole Genome Sequencing. PLoS ONE, 2014, 9, e86803.	2.5	4
80	Identification of Venous Thromboembolism (VTE)-Associated Novel Variants in the ABO Gene Using Targeted Deep Sequencing. Blood, 2011, 118, 709-709.	1.4	0
81	Role of Bioinformatics in Molecular Medicine. , 2020, , 55-68.		0