

Sumit Middha

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Clinical Experience of Cerebrospinal Fluid-Based Liquid Biopsy Demonstrates Superiority of Cell-Free DNA over Cell Pellet Genomic DNA for Molecular Profiling. <i>Journal of Molecular Diagnostics</i> , 2021 , 23, 742-752	5.1	1
80	A Pan-Cancer Study of Somatic TERT Promoter Mutations and Amplification in 30,773 Tumors Profiled by Clinical Genomic Sequencing. <i>Journal of Molecular Diagnostics</i> , 2021 , 23, 253-263	5.1	4
79	Role of Bioinformatics in Molecular Medicine 2020 , 55-68		
78	Retained mismatch repair protein expression occurs in approximately 6% of microsatellite instability-high cancers and is associated with missense mutations in mismatch repair genes. <i>Modern Pathology</i> , 2020 , 33, 871-879	9.8	23
77	Outcome and molecular characteristics of non-invasive encapsulated follicular variant of papillary thyroid carcinoma with oncocytic features. <i>Endocrine</i> , 2019 , 64, 97-108	4	20
76	Clinical Genomic Sequencing of Pediatric and Adult Osteosarcoma Reveals Distinct Molecular Subsets with Potentially Targetable Alterations. <i>Clinical Cancer Research</i> , 2019 , 25, 6346-6356	12.9	39
75	Identification of prognostic molecular biomarkers in 157 HPV-positive and HPV-negative squamous cell carcinomas of the oropharynx. <i>International Journal of Cancer</i> , 2019 , 145, 3152-3162	7.5	24
74	Genetic diversity of tumors with mismatch repair deficiency influences anti-PD-1 immunotherapy response. <i>Science</i> , 2019 , 364, 485-491	33.3	228
73	Majority of -Mutant and -Deficient Colorectal Carcinomas Achieve Clinical Benefit From Immune Checkpoint Inhibitor Therapy and Are Microsatellite Instability-High. <i>JCO Precision Oncology</i> , 2019 , 3,	3.6	32
72	Microsatellite Instability Is Associated With the Presence of Lynch Syndrome Pan-Cancer. <i>Journal of Clinical Oncology</i> , 2019 , 37, 286-295	2.2	203
71	Somatic HNF1A mutations in the malignant transformation of hepatocellular adenomas: a retrospective analysis of data from MSK-IMPACT and TCGA. <i>Human Pathology</i> , 2019 , 83, 1-6	3.7	7
70	Analysis of the Prevalence of Microsatellite Instability in Prostate Cancer and Response to Immune Checkpoint Blockade. <i>JAMA Oncology</i> , 2019 , 5, 471-478	13.4	257
69	Colorectal Carcinomas Containing Hypermethylated MLH1 Promoter and Wild-Type BRAF/KRAS Are Enriched for Targetable Kinase Fusions. <i>Cancer Research</i> , 2019 , 79, 1047-1053	10.1	73
68	Gene Expression Signatures Characterized by Longitudinal Stability and Interindividual Variability Delineate Baseline Phenotypic Groups with Distinct Responses to Immune Stimulation. <i>Journal of Immunology</i> , 2018 , 200, 1917-1928	5.3	4
67	Clinical Sequencing Defines the Genomic Landscape of Metastatic Colorectal Cancer. <i>Cancer Cell</i> , 2018 , 33, 125-136.e3	24.3	338
66	A FISH assay efficiently screens for BRAF gene rearrangements in pancreatic acinar-type neoplasms. <i>Modern Pathology</i> , 2018 , 31, 132-140	9.8	9
65	Clinical Utility of Prospective Molecular Characterization in Advanced Endometrial Cancer. <i>Clinical Cancer Research</i> , 2018 , 24, 5939-5947	12.9	60

64	amplification is a mechanism of acquired resistance to EGFR inhibitors identified by transposon mutagenesis and clinical genomics. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018 , 115, E6030-E6038	11.5	24
63	Genetic Predictors of Response to Systemic Therapy in Esophagogastric Cancer. <i>Cancer Discovery</i> , 2018 , 8, 49-58	24.4	180
62	The Genomic Landscape of Endocrine-Resistant Advanced Breast Cancers. <i>Cancer Cell</i> , 2018 , 34, 427-438	24.6	339
61	Chromosome 20q Amplification Defines a Subtype of Microsatellite Stable, Left-Sided Colon Cancers with Wild-type RAS/RAF and Better Overall Survival. <i>Molecular Cancer Research</i> , 2017 , 15, 708-713	6.6	11
60	Universal screening for microsatellite instability in colorectal cancer in the clinical genomics era: new recommendations, methods, and considerations. <i>Familial Cancer</i> , 2017 , 16, 525-529	3	14
59	Mutational landscape of metastatic cancer revealed from prospective clinical sequencing of 10,000 patients. <i>Nature Medicine</i> , 2017 , 23, 703-713	50.5	1638
58	Genomic Alterations in Fatal Forms of Non-Anaplastic Thyroid Cancer: Identification of and as Novel Thyroid Cancer Genes Associated with Tumor Virulence. <i>Clinical Cancer Research</i> , 2017 , 23, 5970-5980	12.9	64
57	Morphological characterization of colorectal cancers in The Cancer Genome Atlas reveals distinct morphology-molecular associations: clinical and biological implications. <i>Modern Pathology</i> , 2017 , 30, 599-609	9.8	43
56	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. <i>Oncotarget</i> , 2017 , 8, 1495-1507	3.3	8
55	Reliable Pan-Cancer Microsatellite Instability Assessment by Using Targeted Next-Generation Sequencing Data. <i>JCO Precision Oncology</i> , 2017 , 2017,	3.6	128
54	Targeted sequencing of 36 known or putative colorectal cancer susceptibility genes. <i>Molecular Genetics & Genomic Medicine</i> , 2017 , 5, 553-569	2.3	20
53	The genetic landscape of endometrial clear cell carcinomas. <i>Journal of Pathology</i> , 2017 , 243, 230-241	9.4	116
52	Novel High-grade Endometrial Stromal Sarcoma: A Morphologic Mimicker of Myxoid Leiomyosarcoma. <i>American Journal of Surgical Pathology</i> , 2017 , 41, 12-24	6.7	79
51	Optimizing Workflows and Processing of Cytologic Samples for Comprehensive Analysis by Next-Generation Sequencing: Memorial Sloan Kettering Cancer Center Experience. <i>Archives of Pathology and Laboratory Medicine</i> , 2016 , 140, 1200-1205	5	55
50	Identification of Targetable Kinase Alterations in Patients with Colorectal Carcinoma That are Preferentially Associated with Wild-Type RAS/RAF. <i>Molecular Cancer Research</i> , 2016 , 14, 296-301	6.6	34
49	Recurrent, truncating SOX9 mutations are associated with SOX9 overexpression, KRAS mutation, and TP53 wild type status in colorectal carcinoma. <i>Oncotarget</i> , 2016 , 7, 50875-50882	3.3	15
48	Reliable Detection of Mismatch Repair Deficiency in Colorectal Cancers Using Mutational Load in Next-Generation Sequencing Panels. <i>Journal of Clinical Oncology</i> , 2016 , 34, 2141-7	2.2	170
47	REVEL: An Ensemble Method for Predicting the Pathogenicity of Rare Missense Variants. <i>American Journal of Human Genetics</i> , 2016 , 99, 877-885	11	722

46	Homozygous/Compound Heterozygous Triadin Mutations Associated With Autosomal-Recessive Long-QT Syndrome and Pediatric Sudden Cardiac Arrest: Elucidation of the Triadin Knockout Syndrome. <i>Circulation</i> , 2015 , 131, 2051-60	16.7	74
45	Comparison of the Life Cycles of Genetically Distant Species C and Species D Human Adenoviruses Ad6 and Ad26 in Human Cells. <i>Journal of Virology</i> , 2015 , 89, 12401-17	6.6	5
44	Mitochondrial Aging and Physical Decline: Insights From Three Generations of Women. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , 2015 , 70, 1409-17	6.4	36
43	Gene expression patterns in CD4+ peripheral blood cells in healthy subjects and stage IV melanoma patients. <i>Cancer Immunology, Immunotherapy</i> , 2015 , 64, 1437-47	7.4	4
42	Identification of candidate genes for prostate cancer-risk SNPs utilizing a normal prostate tissue eQTL data set. <i>Nature Communications</i> , 2015 , 6, 8653	17.4	49
41	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). <i>FASEB Journal</i> , 2015 , 29, 152-63	0.9	35
40	How well do whole exome sequencing results correlate with medical findings? A study of 89 Mayo Clinic Biobank samples. <i>Frontiers in Genetics</i> , 2015 , 6, 244	4.5	8
39	Comprehensively evaluating cis-regulatory variation in the human prostate transcriptome by using gene-level allele-specific expression. <i>American Journal of Human Genetics</i> , 2015 , 96, 869-82	11	27
38	New DNA Methylation Markers for Pancreatic Cancer: Discovery, Tissue Validation, and Pilot Testing in Pancreatic Juice. <i>Clinical Cancer Research</i> , 2015 , 21, 4473-81	12.9	82
37	Mutational landscape of candidate genes in familial prostate cancer. <i>Prostate</i> , 2014 , 74, 1371-8	4.2	13
36	MAP-RSeq: Mayo Analysis Pipeline for RNA sequencing. <i>BMC Bioinformatics</i> , 2014 , 15, 224	3.6	191
35	HiChIP: a high-throughput pipeline for integrative analysis of ChIP-Seq data. <i>BMC Bioinformatics</i> , 2014 , 15, 280	3.6	38
34	CAP-miRSeq: a comprehensive analysis pipeline for microRNA sequencing data. <i>BMC Genomics</i> , 2014 , 15, 423	4.5	118
33	TNNI3K mutation in familial syndrome of conduction system disease, atrial tachyarrhythmia and dilated cardiomyopathy. <i>Human Molecular Genetics</i> , 2014 , 23, 5793-804	5.6	25
32	Whole genome analyses of a well-differentiated liposarcoma reveals novel SYT1 and DDR2 rearrangements. <i>PLoS ONE</i> , 2014 , 9, e87113	3.7	13
31	Application of whole exome sequencing in undiagnosed inherited polyneuropathies. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014 , 85, 1265-72	5.5	38
30	Integrated genomic characterization reveals novel, therapeutically relevant drug targets in FGFR and EGFR pathways in sporadic intrahepatic cholangiocarcinoma. <i>PLoS Genetics</i> , 2014 , 10, e1004135	6	239
29	Ubiquitin ligase defect by DCAF8 mutation causes HMSN2 with giant axons. <i>Neurology</i> , 2014 , 82, 873-8	6.5	20

28	Comprehensive assessment of genetic variants within TCF4 in Fuchs' endothelial corneal dystrophy. <i>PLoS ONE</i> , 2014 , 55, 6101-7		24
27	Characterizing genetic variation of adrenergic signalling pathways in Takotsubo (stress) cardiomyopathy exomes. <i>European Journal of Heart Failure</i> , 2014 , 16, 942-9	12.3	28
26	From days to hours: reporting clinically actionable variants from whole genome sequencing. <i>PLoS ONE</i> , 2014 , 9, e86803	3.7	3
25	Identification of novel variants in colorectal cancer families by high-throughput exome sequencing. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2013 , 22, 1239-51	4	36
24	Histone deacetylase inhibition promotes osteoblast maturation by altering the histone H4 epigenome and reduces Akt phosphorylation. <i>Journal of Biological Chemistry</i> , 2013 , 288, 28783-91	5.4	65
23	Novel TRAF1-ALK fusion identified by deep RNA sequencing of anaplastic large cell lymphoma. <i>Genes Chromosomes and Cancer</i> , 2013 , 52, 1097-102	5	45
22	769 Novel Methylated DNA Markers Predict Site of Gastrointestinal Cancer. <i>Gastroenterology</i> , 2013 , 144, S-84	13.3	3
21	Novel SOD1 mutation discovered in atypical ALS by whole exome sequencing. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013 , 84, 943-4	5.5	3
20	Exome sequencing and systems biology converge to identify novel mutations in the L-type calcium channel, CACNA1C, linked to autosomal dominant long QT syndrome. <i>Circulation: Cardiovascular Genetics</i> , 2013 , 6, 279-89		80
19	Genome-wide characterization of transcriptional patterns in high and low antibody responders to rubella vaccination. <i>PLoS ONE</i> , 2013 , 8, e62149	3.7	26
18	Multi-platform analysis of microRNA expression measurements in RNA from fresh frozen and FFPE tissues. <i>PLoS ONE</i> , 2013 , 8, e52517	3.7	79
17	Whole-genome sequencing of multiple myeloma from diagnosis to plasma cell leukemia reveals genomic initiating events, evolution, and clonal tides. <i>Blood</i> , 2012 , 120, 1060-6	2.2	310
16	The human gut microbiome: current knowledge, challenges, and future directions. <i>Translational Research</i> , 2012 , 160, 246-57	11	178
15	Novel late-onset Alzheimer disease loci variants associate with brain gene expression. <i>Neurology</i> , 2012 , 79, 221-8	6.5	124
14	Glutathione S-transferase omega genes in Alzheimer and Parkinson disease risk, age-at-diagnosis and brain gene expression: an association study with mechanistic implications. <i>Molecular Neurodegeneration</i> , 2012 , 7, 13	19	57
13	Brain expression genome-wide association study (eGWAS) identifies human disease-associated variants. <i>PLoS Genetics</i> , 2012 , 8, e1002707	6	174
12	Research resource: whole transcriptome RNA sequencing detects multiple 1 α ,25-dihydroxyvitamin D(3)-sensitive metabolic pathways in developing zebrafish. <i>Molecular Endocrinology</i> , 2012 , 26, 1630-42		33
11	TREAT: a bioinformatics tool for variant annotations and visualizations in targeted and exome sequencing data. <i>Bioinformatics</i> , 2012 , 28, 277-8	7.2	50

10	SAAP-RRBS: streamlined analysis and annotation pipeline for reduced representation bisulfite sequencing. <i>Bioinformatics</i> , 2012 , 28, 2180-1	7.2	40
9	Mutations in DNMT1 cause hereditary sensory neuropathy with dementia and hearing loss. <i>Nature Genetics</i> , 2011 , 43, 595-600	36.3	284
8	A novel bioinformatics pipeline for identification and characterization of fusion transcripts in breast cancer and normal cell lines. <i>Nucleic Acids Research</i> , 2011 , 39, e100	20.1	82
7	Identification of Venous Thromboembolism (VTE)-Associated Novel Variants in the ABO Gene Using Targeted Deep Sequencing. <i>Blood</i> , 2011 , 118, 709-709	2.2	
6	Evaluation of oligonucleotide sequence capture arrays and comparison of next-generation sequencing platforms for use in molecular diagnostics. <i>Clinical Chemistry</i> , 2010 , 56, 1297-306	5.5	36
5	3Qtag digital gene expression profiling of human brain and universal reference RNA using Illumina Genome Analyzer. <i>BMC Genomics</i> , 2009 , 10, 531	4.5	132
4	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). <i>Pediatric Blood and Cancer</i> , 2009 , 53, 92-5	3	6
3	Gene networks in <i>Drosophila melanogaster</i> : integrating experimental data to predict gene function. <i>Genome Biology</i> , 2009 , 10, R97	18.3	38
2	Evolution and potential function of fibrinogen-like domains across twelve <i>Drosophila</i> species. <i>BMC Genomics</i> , 2008 , 9, 260	4.5	30
1	YES1 amplification: a mechanism of acquired resistance to EGFR inhibitors identified by transposon mutagenesis and clinical genomics		1