

Mark J Cowley

List of Publications by Year in descending order

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Version: 2024-02-01

146
papers

14,794
citations

38742

50
h-index

20358

116
g-index

160
all docs

160
docs citations

160
times ranked

25487
citing authors

#	ARTICLE	IF	CITATIONS
1	Genomic analyses identify molecular subtypes of pancreatic cancer. <i>Nature</i> , 2016, 531, 47-52.	27.8	2,700
2	Whole genomes redefine the mutational landscape of pancreatic cancer. <i>Nature</i> , 2015, 518, 495-501.	27.8	2,132
3	Pancreatic cancer genomes reveal aberrations in axon guidance pathway genes. <i>Nature</i> , 2012, 491, 399-405.	27.8	1,741
4	Whole-genome landscape of pancreatic neuroendocrine tumours. <i>Nature</i> , 2017, 543, 65-71.	27.8	716
5	PINA v2.0: mining interactome modules. <i>Nucleic Acids Research</i> , 2012, 40, D862-D865.	14.5	321
6	The deubiquitinase USP9X suppresses pancreatic ductal adenocarcinoma. <i>Nature</i> , 2012, 486, 266-270.	27.8	297
7	Genome-wide DNA methylation patterns in pancreatic ductal adenocarcinoma reveal epigenetic deregulation of SLIT-ROBO, ITGA2 and MET signaling. <i>International Journal of Cancer</i> , 2014, 135, 1110-1118.	5.1	192
8	Whole genome, transcriptome and methylome profiling enhances actionable target discovery in high-risk pediatric cancer. <i>Nature Medicine</i> , 2020, 26, 1742-1753.	30.7	185
9	Inheritance of coronary artery disease in men: an analysis of the role of the Y chromosome. <i>Lancet, The</i> , 2012, 379, 915-922.	13.7	179
10	Consolidation of the cancer genome into domains of repressive chromatin by long-range epigenetic silencing (LRES) reduces transcriptional plasticity. <i>Nature Cell Biology</i> , 2010, 12, 235-246.	10.3	178
11	Mutation allele burden remains unchanged in chronic myelomonocytic leukaemia responding to hypomethylating agents. <i>Nature Communications</i> , 2016, 7, 10767.	12.8	177
12	Hypermutation In Pancreatic Cancer. <i>Gastroenterology</i> , 2017, 152, 68-74.e2.	1.3	174
13	Next-Generation Sequencing and Emerging Technologies. <i>Seminars in Thrombosis and Hemostasis</i> , 2019, 45, 661-673.	2.7	168
14	Infant High-Grade Gliomas Comprise Multiple Subgroups Characterized by Novel Targetable Gene Fusions and Favorable Outcomes. <i>Cancer Discovery</i> , 2020, 10, 942-963.	9.4	157
15	Differential Regulation of the Let-7 Family of MicroRNAs in CD4+ T Cells Alters IL-10 Expression. <i>Journal of Immunology</i> , 2012, 188, 6238-6246.	0.8	152
16	Histomolecular Phenotypes and Outcome in Adenocarcinoma of the Ampulla of Vater. <i>Journal of Clinical Oncology</i> , 2013, 31, 1348-1356.	1.6	142
17	Whole Genome Sequencing Improves Outcomes of Genetic Testing in Patients With Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2018, 72, 419-429.	2.8	138
18	Whole-exome sequencing reanalysis at 12 months boosts diagnosis and is cost-effective when applied early in Mendelian disorders. <i>Genetics in Medicine</i> , 2018, 20, 1564-1574.	2.4	132

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19	Cold adaptation in the marine bacterium, <i>Sphingopyxis alaskensis</i> , assessed using quantitative proteomics. <i>Environmental Microbiology</i> , 2010, 12, 2658-2676.	3.8	130
20	Normalization and Statistical Analysis of Quantitative Proteomics Data Generated by Metabolic Labeling. <i>Molecular and Cellular Proteomics</i> , 2009, 8, 2227-2242.	3.8	111
21	Gene-expression profiling of Gram-positive and Gram-negative sepsis in critically ill patients*. <i>Critical Care Medicine</i> , 2008, 36, 1125-1128.	0.9	110
22	Ampullary Cancers Harbor ELF3 Tumor Suppressor Gene Mutations and Exhibit Frequent WNT Dysregulation. <i>Cell Reports</i> , 2016, 14, 907-919.	6.4	107
23	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. <i>Cell</i> , 2018, 172, 924-936.e11.	28.9	103
24	A <i>SLC39A8</i> variant causes manganese deficiency, and glycosylation and mitochondrial disorders. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 261-269.	3.6	101
25	Tailored first-line and second-line CDK4-targeting treatment combinations in mouse models of pancreatic cancer. <i>Gut</i> , 2018, 67, 2142-2155.	12.1	100
26	Clinical and molecular characterization of HER2 amplified-pancreatic cancer. <i>Genome Medicine</i> , 2013, 5, 78.	8.2	97
27	Neuropilin-2 Promotes Extravasation and Metastasis by Interacting with Endothelial $\alpha 5$ Integrin. <i>Cancer Research</i> , 2013, 73, 4579-4590.	0.9	97
28	qpure: A Tool to Estimate Tumor Cellularity from Genome-Wide Single-Nucleotide Polymorphism Profiles. <i>PLoS ONE</i> , 2012, 7, e45835.	2.5	92
29	The influence of genetic variation on gene expression. <i>Genome Research</i> , 2007, 17, 1707-1716.	5.5	91
30	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. <i>American Journal of Human Genetics</i> , 2016, 99, 1229-1244.	6.2	91
31	Mitochondrial CoQ deficiency is a common driver of mitochondrial oxidants and insulin resistance. <i>ELife</i> , 2018, 7, .	6.0	91
32	Brief Report: Potent clinical and radiological response to larotrectinib in TRK fusion-driven high-grade glioma. <i>British Journal of Cancer</i> , 2018, 119, 693-696.	6.4	90
33	Targeting DNA Damage Response and Replication Stress in Pancreatic Cancer. <i>Gastroenterology</i> , 2021, 160, 362-377.e13.	1.3	90
34	c-Myc and Her2 cooperate to drive a stem-like phenotype with poor prognosis in breast cancer. <i>Oncogene</i> , 2014, 33, 3992-4002.	5.9	88
35	Human Islets Express a Marked Proinflammatory Molecular Signature Prior to Transplantation. <i>Cell Transplantation</i> , 2012, 21, 2063-2078.	2.5	85
36	Influence of atrial fibrillation on microRNA expression profiles in left and right atria from patients with valvular heart disease. <i>Physiological Genomics</i> , 2012, 44, 211-219.	2.3	83

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37	Integrating exome sequencing into a diagnostic pathway for epileptic encephalopathy: Evidence of clinical utility and cost effectiveness. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 186-199.	1.2	83
38	MTOR signaling orchestrates stress-induced mutagenesis, facilitating adaptive evolution in cancer. <i>Science</i> , 2020, 368, 1127-1131.	12.6	83
39	HNF4A and GATA6 Loss Reveals Therapeutically Actionable Subtypes in Pancreatic Cancer. <i>Cell Reports</i> , 2020, 31, 107625.	6.4	78
40	Serp1b2 regulates stromal remodelling and local invasion in pancreatic cancer. <i>Oncogene</i> , 2017, 36, 4288-4298.	5.9	77
41	Maternal obesity and diabetes induces latent metabolic defects and widespread epigenetic changes in isogenic mice. <i>Epigenetics</i> , 2013, 8, 602-611.	2.7	75
42	ELF5 Suppresses Estrogen Sensitivity and Underpins the Acquisition of Antiestrogen Resistance in Luminal Breast Cancer. <i>PLoS Biology</i> , 2012, 10, e1001461.	5.6	74
43	SOX9 regulates ERBB signalling in pancreatic cancer development. <i>Gut</i> , 2015, 64, 1790-1799.	12.1	71
44	Somatic Point Mutation Calling in Low Cellularity Tumors. <i>PLoS ONE</i> , 2013, 8, e74380.	2.5	67
45	Cell and Molecular Determinants of <i>In Vivo</i> Efficacy of the BH3 Mimetic ABT-263 against Pediatric Acute Lymphoblastic Leukemia Xenografts. <i>Clinical Cancer Research</i> , 2014, 20, 4520-4531.	7.0	67
46	A Sustained Dietary Change Increases Epigenetic Variation in Isogenic Mice. <i>PLoS Genetics</i> , 2011, 7, e1001380.	3.5	65
47	Whole-genome sequencing overcomes pseudogene homology to diagnose autosomal dominant polycystic kidney disease. <i>European Journal of Human Genetics</i> , 2016, 24, 1584-1590.	2.8	63
48	Sirtuin-1 Regulates Acinar-to-Ductal Metaplasia and Supports Cancer Cell Viability in Pancreatic Cancer. <i>Cancer Research</i> , 2013, 73, 2357-2367.	0.9	59
49	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. <i>Genetics in Medicine</i> , 2020, 22, 1254-1261.	2.4	59
50	Evaluation of Streck BCT and PAXgene Stabilised Blood Collection Tubes for Cell-Free Circulating DNA Studies in Plasma. <i>Molecular Diagnosis and Therapy</i> , 2017, 21, 563-570.	3.8	58
51	Clinical and pathologic features of familial pancreatic cancer. <i>Cancer</i> , 2014, 120, 3669-3675.	4.1	53
52	Denisovan, modern human and mouse TNFAIP3 alleles tune A20 phosphorylation and immunity. <i>Nature Immunology</i> , 2019, 20, 1299-1310.	14.5	53
53	Diagnostic Yield of Whole Genome Sequencing After Nondiagnostic Exome Sequencing or Gene Panel in Developmental and Epileptic Encephalopathies. <i>Neurology</i> , 2021, 96, e1770-e1782.	1.1	53
54	Genome sequencing as a first-line genetic test in familial dilated cardiomyopathy. <i>Genetics in Medicine</i> , 2019, 21, 650-662.	2.4	52

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55	Mitochondrial mutations and metabolic adaptation in pancreatic cancer. <i>Cancer & Metabolism</i> , 2017, 5, 2.	5.0	51
56	Whole genome sequencing provides better diagnostic yield and future value than whole exome sequencing. <i>Medical Journal of Australia</i> , 2018, 209, 197-199.	1.7	48
57	Precision Oncology in Surgery. <i>Annals of Surgery</i> , 2020, 272, 366-376.	4.2	48
58	A Preexistent Hypoxic Gene Signature Predicts Impaired Islet Graft Function and Glucose Homeostasis. <i>Cell Transplantation</i> , 2013, 22, 2147-2159.	2.5	47
59	Asparagine Synthetase Deficiency causes reduced proliferation of cells under conditions of limited asparagine. <i>Molecular Genetics and Metabolism</i> , 2015, 116, 178-186.	1.1	47
60	The Medical Genome Reference Bank contains whole genome and phenotype data of 2570 healthy elderly. <i>Nature Communications</i> , 2020, 11, 435.	12.8	47
61	BCL-2 Hypermethylation Is a Potential Biomarker of Sensitivity to Antimitotic Chemotherapy in Endocrine-Resistant Breast Cancer. <i>Molecular Cancer Therapeutics</i> , 2013, 12, 1874-1885.	4.1	45
62	MicroRNA profiling of the pubertal mouse mammary gland identifies miR-184 as a candidate breast tumour suppressor gene. <i>Breast Cancer Research</i> , 2015, 17, 83.	5.0	44
63	The pseudokinase Sgk223 promotes invasion of pancreatic ductal epithelial cells through JAK1/Stat3 signaling. <i>Molecular Cancer</i> , 2015, 14, 139.	19.2	44
64	Whole genome sequencing for the genetic diagnosis of heterogenous dystonia phenotypes. <i>Parkinsonism and Related Disorders</i> , 2019, 69, 111-118.	2.2	44
65	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. <i>Genetics in Medicine</i> , 2019, 21, 2823-2826.	2.4	44
66	Impaired B Cell Development in the Absence of KrÄppel-like Factor 3. <i>Journal of Immunology</i> , 2011, 187, 5032-5042.	0.8	41
67	A De Novo Mutation in the Sodium-Activated Potassium Channel KCNT2 Alters Ion Selectivity and Causes Epileptic Encephalopathy. <i>Cell Reports</i> , 2017, 21, 926-933.	6.4	40
68	An early inflammatory gene profile in visceral adipose tissue in children. <i>Pediatric Obesity</i> , 2011, 6, e360-e363.	3.2	39
69	Cancer-associated noncoding mutations affect RNA G-quadruplex-mediated regulation of gene expression. <i>Scientific Reports</i> , 2017, 7, 708.	3.3	37
70	Analysis of clinically relevant somatic mutations in high-risk head and neck cutaneous squamous cell carcinoma. <i>Modern Pathology</i> , 2018, 31, 275-287.	5.5	37
71	Mutational Patterns in Metastatic Cutaneous Squamous Cell Carcinoma. <i>Journal of Investigative Dermatology</i> , 2019, 139, 1449-1458.e1.	0.7	36
72	ClinSV: clinical grade structural and copy number variant detection from whole genome sequencing data. <i>Genome Medicine</i> , 2021, 13, 32.	8.2	36

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73	Unscrambling cancer genomes via integrated analysis of structural variation and copy number. <i>Cell Genomics</i> , 2022, 2, 100112.	6.5	34
74	Germline variants in familial pituitary tumour syndrome genes are common in young patients and families with additional endocrine tumours. <i>European Journal of Endocrinology</i> , 2017, 176, 635-644.	3.7	33
75	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. <i>Med</i> , 2021, 2, 49-73.e10.	4.4	33
76	Use of Whole-Genome Sequencing for Mitochondrial Disease Diagnosis. <i>Neurology</i> , 2022, 99, .	1.1	33
77	Defining the genetic basis of early onset hereditary spastic paraplegia using whole genome sequencing. <i>Neurogenetics</i> , 2016, 17, 265-270.	1.4	32
78	Proteomic comparison of colorectal tumours and non-neoplastic mucosa from paired patient samples using iTRAQ mass spectrometry. <i>Molecular BioSystems</i> , 2011, 7, 2997.	2.9	31
79	Identification of Novel GH-Regulated Pathway of Lipid Metabolism in Adipose Tissue: A Gene Expression Study in Hypopituitary Men. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011, 96, E1188-E1196.	3.6	31
80	Understanding pancreatic cancer genomes. <i>Journal of Hepato-Biliary-Pancreatic Sciences</i> , 2013, 20, 549-556.	2.6	31
81	Revealing hidden genetic diagnoses in the ocular anterior segment disorders. <i>Genetics in Medicine</i> , 2020, 22, 1623-1632.	2.4	31
82	Clonal expansions of cytotoxic T cells exist in the blood of patients with Waldenström macroglobulinemia but exhibit anergic properties and are eliminated by nucleoside analogue therapy. <i>Blood</i> , 2010, 115, 3580-3588.	1.4	30
83	Detection of Growth Hormone Doping by Gene Expression Profiling of Peripheral Blood. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 4703-4709.	3.6	29
84	Normalization procedures and detection of linkage signal in genetical-genomics experiments. <i>Nature Genetics</i> , 2006, 38, 855-856.	21.4	28
85	Increased Diagnostic Yield of Spastic Paraplegia with or Without Cerebellar Ataxia Through Whole-Genome Sequencing. <i>Cerebellum</i> , 2019, 18, 781-790.	2.5	28
86	Evaluation of the NOD/SCID xenograft model for glucocorticoid-regulated gene expression in childhood B-cell precursor acute lymphoblastic leukemia. <i>BMC Genomics</i> , 2011, 12, 565.	2.8	27
87	Seave: a comprehensive web platform for storing and interrogating human genomic variation. <i>Bioinformatics</i> , 2019, 35, 122-125.	4.1	26
88	Deep multi-region whole-genome sequencing reveals heterogeneity and gene-by-environment interactions in treatment-naive, metastatic lung cancer. <i>Oncogene</i> , 2019, 38, 1661-1675.	5.9	26
89	Extracellular matrix composition significantly influences pancreatic stellate cell gene expression pattern: role of transgelin in PSC function. <i>American Journal of Physiology - Renal Physiology</i> , 2013, 305, G408-G417.	3.4	25
90	Unique presentation of cutis laxa with Leigh-like syndrome due to <i>ECHS1</i> deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 745-747.	3.6	25

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91	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. <i>American Journal of Human Genetics</i> , 2017, 101, 995-1005.	6.2	23
92	Pathogenic variants in <i>PLOD3</i> result in a Stickler syndrome-like connective tissue disorder with vascular complications. <i>Journal of Medical Genetics</i> , 2019, 56, 629-638.	3.2	23
93	Expression of Pro- and Antiapoptotic Molecules of the Bcl-2 Family in Human Islets Postisolation. <i>Cell Transplantation</i> , 2012, 21, 49-60.	2.5	22
94	Somatic mutations in salivary duct carcinoma and potential therapeutic targets. <i>Oncotarget</i> , 2017, 8, 75893-75903.	1.8	22
95	Intra- and inter-individual genetic differences in gene expression. <i>Mammalian Genome</i> , 2009, 20, 281-295.	2.2	21
96	Expanding the spectrum of PEX16 mutations and novel insights into disease mechanisms. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 16, 46-51.	1.1	21
97	High Degree of Genetic Heterogeneity for Hereditary Cerebellar Ataxias in Australia. <i>Cerebellum</i> , 2019, 18, 137-146.	2.5	21
98	JRK is a positive regulator of β -catenin transcriptional activity commonly overexpressed in colon, breast and ovarian cancer. <i>Oncogene</i> , 2016, 35, 2834-2841.	5.9	20
99	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 55-94.	1.6	20
100	Beyond the panel: preconception screening in consanguineous couples using the TruSight One $\text{\textcircled{e}}$ clinical exome $\text{\textcircled{e}}$. <i>Genetics in Medicine</i> , 2019, 21, 608-612.	2.4	20
101	Targeted Therapy of <i>TERT</i> -Rearranged Neuroblastoma with BET Bromodomain Inhibitor and Proteasome Inhibitor Combination Therapy. <i>Clinical Cancer Research</i> , 2021, 27, 1438-1451.	7.0	20
102	Genomic diagnostics in polycystic kidney disease: an assessment of real-world use of whole-genome sequencing. <i>European Journal of Human Genetics</i> , 2021, 29, 760-770.	2.8	20
103	De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 542-552.	6.2	19
104	Efficacy of MEK inhibition in a recurrent malignant peripheral nerve sheath tumor. <i>Npj Precision Oncology</i> , 2021, 5, 9.	5.4	19
105	Integration of genomics, high throughput drug screening, and personalized xenograft models as a novel precision medicine paradigm for high risk pediatric cancer. <i>Cancer Biology and Therapy</i> , 2018, 19, 1078-1087.	3.4	18
106	Development and validation of a targeted gene sequencing panel for application to disparate cancers. <i>Scientific Reports</i> , 2019, 9, 17052.	3.3	18
107	RON is not a prognostic marker for resectable pancreatic cancer. <i>BMC Cancer</i> , 2012, 12, 395.	2.6	17
108	Mutations in the exocyst component EXOC2 cause severe defects in human brain development. <i>Journal of Experimental Medicine</i> , 2020, 217, .	8.5	17

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109	The Antiproliferative Effects of Progestins in T47D Breast Cancer Cells Are Tempered by Progestin Induction of the ETS Transcription Factor Elf5. <i>Molecular Endocrinology</i> , 2010, 24, 1380-1392.	3.7	16
110	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic PNPT1 Variants. <i>Journal of Clinical Medicine</i> , 2019, 8, 2020.	2.4	16
111	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A (<i>KIF1A</i>) in tuberous sclerosis complex. <i>Journal of Medical Genetics</i> , 2020, 57, 100-107.	2.5	16
112	Glutamine addiction promotes glucose oxidation in triple-negative breast cancer. <i>Oncogene</i> , 2022, 41, 4066-4078.	5.9	15
113	Gonadal mosaicism of a novel IQSEC2 variant causing female limited intellectual disability and epilepsy. <i>European Journal of Human Genetics</i> , 2017, 25, 763-767.	2.8	14
114	Molecular patterns in salivary duct carcinoma identify prognostic subgroups. <i>Modern Pathology</i> , 2020, 33, 1896-1909.	5.5	14
115	Genetic dissection of gene regulation in multiple mouse tissues. <i>Mammalian Genome</i> , 2006, 17, 490-495.	2.2	13
116	In vitro and in vivo drug screens of tumor cells identify novel therapies for high-risk child cancer. <i>EMBO Molecular Medicine</i> , 2022, 14, e14608.	6.9	12
117	Cryptic intronic NBAS variant reveals the genetic basis of recurrent liver failure in a child. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 77-82.	1.1	11
118	Population data improves variant interpretation in autosomal dominant polycystic kidney disease. <i>Genetics in Medicine</i> , 2019, 21, 1425-1434.	2.4	11
119	Genome sequencing in congenital cataracts improves diagnostic yield. <i>Human Mutation</i> , 2021, 42, 1173-1183.	2.5	10
120	Oral malignant gastrointestinal neuroectodermal tumour with junctional component mimicking mucosal melanoma. <i>Pathology</i> , 2018, 50, 648-653.	0.6	8
121	Proteogenomic analysis of Inhibitor of Differentiation 4 (ID4) in basal-like breast cancer. <i>Breast Cancer Research</i> , 2020, 22, 63.	5.0	8
122	Application of Genome Sequencing from Blood to Diagnose Mitochondrial Diseases. <i>Genes</i> , 2021, 12, 607.	2.4	8
123	EPC5-Related Vici Syndrome: A Primary Defect of Autophagic Regulation with an Emerging Phenotype Overlapping with Mitochondrial Disorders. <i>JIMD Reports</i> , 2017, 42, 19-29.	1.5	7
124	Genomic stratification and liquid biopsy in a rare adrenocortical carcinoma (ACC) case, with dual lung metastases. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003764.	1.2	7
125	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. <i>Human Mutation</i> , 2019, 40, 374-379.	2.5	7
126	Whole-genome sequencing facilitates patient-specific quantitative PCR-based minimal residual disease monitoring in acute lymphoblastic leukaemia, neuroblastoma and Ewing sarcoma. <i>British Journal of Cancer</i> , 2022, 126, 482-491.	6.4	7

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127	A Novel Orthotopic Patient-Derived Xenograft Model of Radiation-Induced Glioma Following Medulloblastoma. <i>Cancers</i> , 2020, 12, 2937.	3.7	6
128	RLIM Is a Candidate Dosage-Sensitive Gene for Individuals with Varying Duplications of Xq13, Intellectual Disability, and Distinct Facial Features. <i>American Journal of Human Genetics</i> , 2020, 107, 1157-1169.	6.2	6
129	Childhood acute myeloid leukemia shows a high level of germline predisposition. <i>Blood</i> , 2021, 138, 2293-2298.	1.4	5
130	Hierarchical Bayes variable selection and microarray experiments. <i>Journal of Multivariate Analysis</i> , 2007, 98, 852-872.	1.0	4
131	Recurrent <i>SPECC1</i> – <i>NTRK</i> fusions in pediatric sarcoma and brain tumors. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a005710.	1.2	4
132	A G316A Polymorphism in the Ornithine Decarboxylase Gene Promoter Modulates MYCN-Driven Childhood Neuroblastoma. <i>Cancers</i> , 2021, 13, 1807.	3.7	4
133	Whole Genome Sequencing, Focused Assays and Functional Studies Increasing Understanding in Cryptic Inherited Retinal Dystrophies. <i>International Journal of Molecular Sciences</i> , 2022, 23, 3905.	4.1	4
134	Decompensation of cardiorespiratory function and emergence of anemia during pregnancy in a case of mitochondrial myopathy, lactic acidosis, and sideroblastic anemia 2 with compound heterozygous <i>YARS2</i> pathogenic variants. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2226-2230.	1.2	4
135	Reversible Suppression of Lymphoproliferation and Thrombocytopenia with Rapamycin in a Patient with Common Variable Immunodeficiency. <i>Journal of Clinical Immunology</i> , 2018, 38, 159-162.	3.8	3
136	Serum microRNA expression during neoadjuvant chemoradiation for rectal cancer.. <i>Journal of Clinical Oncology</i> , 2017, 35, e15081-e15081.	1.6	3
137	Clonal Expansions of Cytotoxic T Cells in the Blood of Patients with Waldenstrom's Macroglobulinaemia Are Anergic and Disappear After Nucleoside Analogue Therapy.. <i>Blood</i> , 2009, 114, 1820-1820.	1.4	3
138	Intra- and inter-individual genetic differences in gene expression. <i>Nature Precedings</i> , 2008, , .	0.1	2
139	Measurable residual disease analysis in paediatric acute lymphoblastic leukaemia patients with ABL-class fusions. <i>British Journal of Cancer</i> , 2022, 127, 908-915.	6.4	2
140	The Cancer Molecular Screening and Therapeutics Program (MoST): Actionable mutation frequencies in a population with rare and less common cancers.. <i>Journal of Clinical Oncology</i> , 2019, 37, 3136-3136.	1.6	1
141	Abstract 1942: Change in serum microRNA expression during neoadjuvant chemoradiation for rectal cancer. , 2016, , .		1
142	A Pre-Existent Hypoxic Gene Signature Predicts Impaired Islet Graft Function and Glucose Homeostasis. <i>Transplantation</i> , 2012, 94, 710.	1.0	0
143	Response to Brodehl et al.. <i>Genetics in Medicine</i> , 2019, 21, 1248-1249.	2.4	0
144	Different types of disease-causing noncoding variants revealed by genomic and gene expression analyses in families with X-linked intellectual disability. <i>Human Mutation</i> , 2021, 42, 835-847.	2.5	0

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145	Abstract LB-73: SOX9 regulates EGFR/ERBB signaling in pancreatic cancer. , 2014, , .		0
146	The Molecular Screening and Therapeutics (MoST) Program: A precision medicine framework for biomarker-driven signal-seeking clinical studies for rare cancers.. Journal of Clinical Oncology, 2017, 35, TPS2621-TPS2621.	1.6	0