Patrick May

List of Publications by Year
in descending order

143 papers	8,508 citations	50170 46 h-index	58464 82 g-index
175 all docs	175 docs citations	175 times ranked	14746 citing authors

#	Article	IF	Citations
1	<scp>M</scp>ercator: a fast and simple web server for genome scale functional annotation of plant sequence data. Plant, Cell and Environment, 2014, 37, 1250-1258.	2.8	575
2	Identification of Nutrient-Responsive Arabidopsis and Rapeseed MicroRNAs by Comprehensive Real-Time Polymerase Chain Reaction Profiling and Small RNA Sequencing Â Â Â. Plant Physiology, 2009, 150, 1541-1555.	2.3	414
3	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
4	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	2.6	237
5	Stars and Symbiosis: MicroRNA- and MicroRNA*-Mediated Transcript Cleavage Involved in Arbuscular Mycorrhizal Symbiosis Â Â. Plant Physiology, 2011, 156, 1990-2010.	2.3	235
6	Integrated multi-omics of the human gut microbiome in a case study of familial type 1 diabetes. Nature Microbiology, 2017, 2, 16180.	5.9	233
7	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	9.4	230
8	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	9.4	224
9	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. Nature Genetics, 2021, 53, 294-303.	9.4	198
10	Birth mode is associated with earliest strain-conferred gut microbiome functions and immunostimulatory potential. Nature Communications, 2018, 9, 5091.	5.8	190
11	Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Nature Genetics, 2014, 46, 1327-1332.	9.4	178
12	Single-cell sequencing of human midbrain reveals glial activation and a Parkinson-specific neuronal state. Brain, 2022, 145, 964-978.	3.7	177
13	ZIB Structure Prediction Pipeline: Composing a Complex Biological Workflow Through Web Services. Lecture Notes in Computer Science, 2006, , 1148-1158.	1.0	158
14	Expression Pattern Suggests a Role of MiR399 in the Regulation of the Cellular Response to Local Pi Increase During Arbuscular Mycorrhizal Symbiosis. Molecular Plant-Microbe Interactions, 2010, 23, 915-926.	1.4	157
15	Metabolomics- and Proteomics-Assisted Genome Annotation and Analysis of the Draft Metabolic Network of Chlamydomonas reinhardtii. Genetics, 2008, 179, 157-166.	1.2	141
16	IMP: a pipeline forÂreproducible reference-independent integrated metagenomic and metatranscriptomic analyses. Genome Biology, 2016, 17, 260.	3.8	141
17	Genomic Sequence Diversity and Population Structure ofSaccharomyces cerevisiaeAssessed by RAD-seq. G3: Genes, Genomes, Genetics, 2013, 3, 2163-2171.	0.8	132
18	A novel Fanconi anaemia subtype associated with a dominant-negative mutation in RAD51. Nature Communications, 2015, 6, 8829.	5.8	130

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19	Systems genomics evaluation of the SH-SY5Y neuroblastoma cell line as a model for Parkinsonâ€™s disease. BMC Genomics, 2014, 15, 1154.	1.2	126
20	The effects of carbon dioxide and temperature on microRNA expression in Arabidopsis development. Nature Communications, 2013, 4, 2145.	5.8	122
21	Parkinsonâ€™s disease-associated alterations of the gut microbiome predict disease-relevant changes in metabolic functions. BMC Biology, 2020, 18, 62.	1.7	122
22	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
23	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Neurobiology of Aging, 2017, 57, 247.e9-247.e13.	1.5	108
24	Recessive mutations inSLC13A5result in a loss of citrate transport and cause neonatal epilepsy, developmental delay and teeth hypoplasia. Brain, 2015, 138, 3238-3250.	3.7	96
25	Targeted proteomics for Chlamydomonas reinhardtii combined with rapid subcellular protein fractionation, metabolomics and metabolic flux analyses. Molecular BioSystems, 2010, 6, 1018.	2.9	94
26	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	3.7	90
27	Molecular and Clinical Evidence for anARMC5Tumor Syndrome: Concurrent Inactivating Germline and Somatic Mutations Are Associated With Both Primary Macronodular Adrenal Hyperplasia and Meningioma. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E119-E128.	1.8	85
28	Predicting functional effects of missense variants in voltage-gated sodium and calcium channels. Science Translational Medicine, 2020, 12, .	5.8	84
29	Targeted sequencing of 351 candidate genes for epileptic encephalopathy in a large cohort of patients. Molecular Genetics & Genomic Medicine, 2016, 4, 568-580.	0.6	83
30	Comparative integrated omics: identification of key functionalities in microbial community-wide metabolic networks. Npj Biofilms and Microbiomes, 2015, 1, 15007.	2.9	82
31	PathoFact: a pipeline for the prediction of virulence factors and antimicrobial resistance genes in metagenomic data. Microbiome, 2021, 9, 49.	4.9	81
32	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. American Journal of Human Genetics, 2019, 104, 1060-1072.	2.6	78
33	Metformin reverses TRAP1 mutation-associated alterations in mitochondrial function in Parkinsonâ€™s disease. Brain, 2017, 140, 2444-2459.	3.7	76
34	Community-integrated omics links dominance of a microbial generalist to fine-tuned resource usage. Nature Communications, 2014, 5, 5603.	5.8	75
35	Condensing the omics fog of microbial communities. Trends in Microbiology, 2013, 21, 325-333.	3.5	74
36	ChlamyCyc: an integrative systems biology database and web-portal for Chlamydomonas reinhardtii. BMC Genomics, 2009, 10, 209.	1.2	73

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37	Patient-derived organoids and orthotopic xenografts of primary and recurrent gliomas represent relevant patient avatars for precision oncology. Acta Neuropathologica, 2020, 140, 919-949.	3.9	72
38	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. Brain, 2016, 139, 2420-2430.	3.7	70
39	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	3.7	69
40	Comprehensive characterization of amino acid positions in protein structures reveals molecular effect of missense variants. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 28201-28211.	3.3	68
41	An integrative approach towards completing genome-scale metabolic networks. Molecular BioSystems, 2009, 5, 1889.	2.9	67
42	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	4.9	67
43	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	4.5	66
44	The RNA Complement of Outer Membrane Vesicles From Salmonella enterica Serovar Typhimurium Under Distinct Culture Conditions. Frontiers in Microbiology, 2018, 9, 2015.	1.5	62
45	Confronting the catalytic dark matter encoded by sequenced genomes. Nucleic Acids Research, 2017, 45, 11495-11514.	6.5	59
46	Spectrum of GABAA receptor variants in epilepsy. Current Opinion in Neurology, 2019, 32, 183-190.	1.8	59
47	Integration of time-series meta-omics data reveals how microbial ecosystems respond to disturbance. Nature Communications, 2020, 11, 5281.	5.8	57
48	Mutations in RHOT1 Disrupt Endoplasmic Reticulumâ€“Mitochondria Contact Sites Interfering with Calcium Homeostasis and Mitochondrial Dynamics in Parkinson's Disease. Antioxidants and Redox Signaling, 2019, 31, 1213-1234.	2.5	56
49	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54
50	Recessive loss-of-function mutations in AP4S1 cause mild fever-sensitive seizures, developmental delay and spastic paraplegia through loss of AP-4 complex assembly. Human Molecular Genetics, 2015, 24, 2218-2227.	1.4	53
51	Simple ClinVar: an interactive web server to explore and retrieve gene and disease variants aggregated in ClinVar database. Nucleic Acids Research, 2019, 47, W99-W105.	6.5	51
52	Statistical and Machine Learning Techniques in Human Microbiome Studies: Contemporary Challenges and Solutions. Frontiers in Microbiology, 2021, 12, 635781.	1.5	51
53	Docking without docking: ISEARCHâ€"prediction of interactions using known interfaces. Proteins: Structure, Function and Bioinformatics, 2007, 69, 839-844.	1.5	50
54	Connectivity independent protein-structure alignment: a hierarchical approach. BMC Bioinformatics, 2006, 7, 510.	1.2	48

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55	New insights into <scp>C</scp>hlamydomonas reinhardtii hydrogen production processes by combined microarray/<scp>RNA</scp>â€seq transcriptomics. Plant Biotechnology Journal, 2013, 11, 717-733.	4.1	47
56	Identification of pathogenic variant enriched regions across genes and gene families. Genome Research, 2020, 30, 62-71.	2.4	47
57	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
58	Roles of bacteriophages, plasmids and CRISPR immunity in microbial community dynamics revealed using time-series integrated meta-omics. Nature Microbiology, 2021, 6, 123-135.	5.9	47
59	Small RNA profiling of low biomass samples: identification and removal of contaminants. BMC Biology, 2018, 16, 52.	1.7	46
60	Identification and classification of ncRNA molecules using graph properties. Nucleic Acids Research, 2009, 37, e66-e66.	6.5	44
61	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.5	43
62	A rare loss-of-function variant of ADAM17 is associated with late-onset familial Alzheimer disease. Molecular Psychiatry, 2020, 25, 629-639.	4.1	42
63	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. Genome Medicine, 2020, 12, 28.	3.6	42
64	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
65	Distribution pattern of small RNA and degradome reads provides information on miRNA gene structure and regulation. Plant Signaling and Behavior, 2011, 6, 1609-1611.	1.2	39
66	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	2.6	35
67	Critical Assessment of MetaProteome Investigation (CAMPI): a multi-laboratory comparison of established workflows. Nature Communications, 2021, 12, 7305.	5.8	34
68	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	3.7	33
69	Sequenceâ€“structure relationships in RNA loops: establishing the basis for loop homology modeling. Nucleic Acids Research, 2010, 38, 970-980.	6.5	31
70	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	1.5	30
71	Major changes of cell function and toxicant sensitivity in cultured cells undergoing mild, quasi-natural genetic drift. Archives of Toxicology, 2018, 92, 3487-3503.	1.9	27
72	Missing heritability in Parkinsonâ€™s disease: the emerging role of non-coding genetic variation. Journal of Neural Transmission, 2020, 127, 729-748.	1.4	27

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73	Give It AGO: The Search for miRNA-Argonaute Sorting Signals in Arabidopsis thaliana Indicates a Relevance of Sequence Positions Other than the 5â€²-Position Alone. Frontiers in Plant Science, 2012, 3, 272.	1.7	25
74	Persistence of birth mode-dependent effects on gut microbiome composition, immune system stimulation and antimicrobial resistance during the first year of life. ISME Communications, 2021, 1, .	1.7	25
75	Burden of rare variants in synaptic genes in patients with severe tinnitus: An exome based extreme phenotype study. EBioMedicine, 2021, 66, 103309.	2.7	25
76	Genome-wide Association and Meta-analysis of Age at Onset in Parkinson Disease. Neurology, 2022, 99, .	1.5	25
77	A patient-based model of RNA mis-splicing uncovers treatment targets in Parkinsonâ€™s disease. Science Translational Medicine, 2020, 12, .	5.8	24
78	Natural variation of chronological aging in the Saccharomyces cerevisiae species reveals diet-dependent mechanisms of life span control. Npj Aging and Mechanisms of Disease, 2018, 4, 3.	4.5	23
79	Hemap: An Interactive Online Resource for Characterizing Molecular Phenotypes across Hematologic Malignancies. Cancer Research, 2019, 79, 2466-2479.	0.4	23
80	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2020, 107, 683-697.	2.6	23
81	Genetic Architecture of Parkinson's Disease in the Indian Population: Harnessing Genetic Diversity to Address Critical Gaps in Parkinson's Disease Research. Frontiers in Neurology, 2020, 11, 524.	1.1	23
82	Extraction and Analysis of RNA Isolated from Pure Bacteria-Derived Outer Membrane Vesicles. Methods in Molecular Biology, 2018, 1737, 213-230.	0.4	22
83	Exome-wide analysis of mutational burden in patients with typical and atypical Rolandic epilepsy. European Journal of Human Genetics, 2018, 26, 258-264.	1.4	22
84	Mantis: flexible and consensus-driven genome annotation. GigaScience, 2021, 10, .	3.3	22
85	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	1.1	22
86	Breast and prostate cancer risk: The interplay of polygenic risk, rare pathogenic germline variants, and family history. Genetics in Medicine, 2022, 24, 576-585.	1.1	22
87	Enrichment of damaging missense variants in genes related with axonal guidance signalling in sporadic Menièreâ€™s disease. Journal of Medical Genetics, 2020, 57, 82-88.	1.5	21
88	Mendelian Randomisation Study of Smoking, Alcohol, and Coffee Drinking in Relation to Parkinsonâ€™s Disease. Journal of Parkinson's Disease, 2022, 12, 267-282.	1.5	21
89	Genome Sequencing of SARS-CoV-2 Allows Monitoring of Variants of Concern through Wastewater. Water (Switzerland), 2021, 13, 3018.	1.2	21
90	Metabolic networks are NP-hard to reconstruct. Journal of Theoretical Biology, 2008, 254, 807-816.	0.8	20

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91	High-throughput tetrad analysis. Nature Methods, 2013, 10, 671-675.	9.0	20
92	Non-Coding RNAs in the Brain-Heart Axis: The Case of Parkinsonâ€™s Disease. International Journal of Molecular Sciences, 2020, 21, 6513.	1.8	19
93	Comparative analysis of miRNAs and their targets across four plant species. BMC Research Notes, 2011, 4, 483.	0.6	18
94	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	3.7	18
95	Genetic evaluation of dementia with Lewy bodies implicates distinct disease subgroups. Brain, 2022, 145, 1757-1762.	3.7	17
96	Proteomic Analysis of Dhh1 Complexes Reveals a Role for Hsp40 Chaperone Ydj1 in Yeast P-Body Assembly. G3: Genes, Genomes, Genetics, 2015, 5, 2497-2511.	0.8	16
97	Recessive mutations in SLC35A3 cause early onset epileptic encephalopathy with skeletal defects. American Journal of Medical Genetics, Part A, 2017, 173, 1119-1123.	0.7	16
98	PTGL--a web-based database application for protein topologies. Bioinformatics, 2004, 20, 3277-3279.	1.8	15
99	Dairy Intake and Parkinson's Disease: A Mendelian Randomization Study. Movement Disorders, 2022, 37, 857-864.	2.2	15
100	Understanding the role of genetic variability in LRRK2 in Indian population. Movement Disorders, 2019, 34, 496-505.	2.2	14
101	MISCAST: MIssense variant to protein StruCture Analysis web SuiTe. Nucleic Acids Research, 2020, 48, W132-W139.	6.5	14
102	PTGL: a database for secondary structure-based protein topologies. Nucleic Acids Research, 2010, 38, D326-D330.	6.5	13
103	Rare ABCA7 variants in 2 German families with Alzheimer disease. Neurology: Genetics, 2018, 4, e224.	0.9	12
104	Assessment of genetic variant burden in epilepsy-associated brain lesions. European Journal of Human Genetics, 2019, 27, 1738-1744.	1.4	12
105	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.5	11
106	Excess of singleton loss-of-function variants in Parkinsonâ€™s disease contributes to genetic risk. Journal of Medical Genetics, 2020, 57, 617-623.	1.5	10
107	Integration of Proteomic and Metabolomic Profiling as well as Metabolic Modeling for the Functional Analysis of Metabolic Networks. Methods in Molecular Biology, 2011, 694, 341-363.	0.4	10
108	A Protein Prioritization Approach Tailored for the FA/BRCA Pathway. PLoS ONE, 2013, 8, e62017.	1.1	8

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109	Identification, Recovery, and Refinement of Hitherto Undescribed Population-Level Genomes from the Human Gastrointestinal Tract. Frontiers in Microbiology, 2016, 7, 884.	1.5	8
110	Guidelineâ€based and bioinformatic reassessment of lesionâ€associated gene and variant pathogenicity in focal human epilepsies. Epilepsia, 2018, 59, 2145-2152.	2.6	8
111	Replication of a Novel Parkinson's Locus in a European Ancestry Population. Movement Disorders, 2021, 36, 1689-1695.	2.2	8
112	Challenges, Strategies, and Perspectives for Reference-Independent Longitudinal Multi-Omic Microbiome Studies. Frontiers in Genetics, 2021, 12, 666244.	1.1	8
113	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
114	JAIL: a structure-based interface library for macromolecules. Nucleic Acids Research, 2009, 37, D338-D341.	6.5	7
115	The new protein topology graph library web server. Bioinformatics, 2016, 32, 474-476.	1.8	7
116	Machine learning-based identification and characterization of 15 novel pathogenic SUOX missense mutations. Molecular Genetics and Metabolism, 2021, 134, 188-194.	0.5	7
117	Distinct gene-set burden patterns underlie common generalized and focal epilepsies. EBioMedicine, 2021, 72, 103588.	2.7	7
118	Genome-wide linkage analysis of families with primary hyperhidrosis. PLoS ONE, 2020, 15, e0244565.	1.1	7
119	Benchmarking Low-Frequency Variant Calling With Long-Read Data on Mitochondrial DNA. Frontiers in Genetics, 0, 13, .	1.1	7
120	Modeling RNA loops using sequence homology and geometric constraints. Bioinformatics, 2010, 26, 1671-1672.	1.8	6
121	Rare gene deletions in genetic generalized and Rolandic epilepsies. PLoS ONE, 2018, 13, e0202022.	1.1	6
122	Unification of functional annotation descriptions using text mining. Biological Chemistry, 2021, 402, 983-990.	1.2	6
123	Functional meta-omics provide critical insights into long- and short-read assemblies. Briefings in Bioinformatics, 2021, 22, .	3.2	6
124	Algebraic connectivity may explain the evolution of gene regulatory networks. Journal of Theoretical Biology, 2010, 267, 7-14.	0.8	5
125	Variant Score Rankerâ€”a web application for intuitive missense variant prioritization. Bioinformatics, 2019, 35, 4478-4479.	1.8	5
126	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	2.6	5

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127	POMO - Plotting Omics analysis results for Multiple Organisms. BMC Genomics, 2013, 14, 918.	1.2	4
128	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. New England Journal of Medicine, 2019, 380, e24.	13.9	4
129	The Interaction between <scp>HLAâ€DRB1</scp> and Smoking in Parkinson's Disease Revisited. Movement Disorders, 2022, 37, 1929-1937.	2.2	4
130	BSA4Yeast: Web-based quantitative trait locus linkage analysis and bulk segregant analysis of yeast sequencing data. GigaScience, 2019, 8, .	3.3	3
131	Systematic characterization of human gut microbiome-secreted molecules by integrated multi-omics. ISME Communications, 2021, 1, 82.	1.7	3
132	Variant-DB: A Tool for Efficiently Exploring Millions of Human Genetic Variants and Their Annotations. Lecture Notes in Computer Science, 2017, , 22-28.	1.0	2
133	Reply: No evidence for rare TRAP1 mutations influencing the risk of idiopathic Parkinsonâ€™s disease. Brain, 2018, 141, e17-e17.	3.7	2
134	Genomic analysis of AlphaFold2-predicted structures identifies maps of 3D essential sites in 243 neurodevelopmental disorder-associated proteins. Biophysical Journal, 2022, 121, 165a-166a.	0.2	2
135	Accelerated microRNA-Precursor Detection Using the Smith-Waterman Algorithm on FPGAs. , 2007, , 19-32.		1
136	Hierarchical Representation of Supersecondary Structures Using a Graph-Theoretical Approach. Methods in Molecular Biology, 2012, 932, 7-33.	0.4	0
137	[P2â€“108]: IDENTIFICATION OF A RARE GENE VARIANT THAT IS ASSOCIATED WITH FAMILIAL ALZHEIMER DISEASE AND REGULATES APP EXPRESSION. Alzheimer's and Dementia, 2017, 13, P648.	0.4	0
138	Functional Interpretation of Single Amino Acid Substitutions in 1,330 Disease-Associated Genes. Biophysical Journal, 2019, 116, 420a-421a.	0.2	0
139	Evaluation of the Molecular Pathogenesis of Adrenocortical Tumors by Whole-Genome Sequencing. Journal of the Endocrine Society, 2021, 5, A68-A68.	0.1	0
140	Evaluation of Cell Line Suitability for Disease Specific Perturbation Experiments. Studies in Classification, Data Analysis, and Knowledge Organization, 2015, , 297-307.	0.1	0
141	Mapping the Gene Expression Diversity Across Hematological and Lymphoid Malignancies. Blood, 2015, 126, 4805-4805.	0.6	0
142	Burden of Rare Variants in Synaptic Genes in Patients with Severe Tinnitus: An Exome Based Extreme Phenotype Study. SSRN Electronic Journal, 0, , .	0.4	0
143	Benchmarking of univariate pleiotropy detection methods applied to epilepsy. Human Mutation, 2022, 43, 1314-1332.	1.1	0