Wyeth W Wasserman

List of Publications by Year
in descending order

160	22,244	26630	10158
papers	citations	56	140
		h-index	g-index

192	192	192	40728
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A promoter-level mammalian expression atlas. Nature, 2014, 507, 462-470.	27.8	1,838
2	The clonal and mutational evolution spectrum of primary triple-negative breast cancers. Nature, 2012, 486, 395-399.	27.8	1,778
3	JASPAR: an open-access database for eukaryotic transcription factor binding profiles. Nucleic Acids Research, 2004, 32, 91D-94.	14.5	1,451
4	JASPAR 2018: update of the open-access database of transcription factor binding profiles and its web framework. Nucleic Acids Research, 2018, 46, D260-D266.	14.5	1,232
5	JASPAR 2020: update of the open-access database of transcription factor binding profiles. Nucleic Acids Research, 2020, 48, D87-D92.	14.5	1,039
6	Applied bioinformatics for the identification of regulatory elements. Nature Reviews Genetics, 2004, 5, 276-287.	16.3	1,032
7	JASPAR 2016: a major expansion and update of the open-access database of transcription factor binding profiles. Nucleic Acids Research, 2016, 44, D110-D115.	14.5	968
8	JASPAR 2014: an extensively expanded and updated open-access database of transcription factor binding profiles. Nucleic Acids Research, 2014, 42, D142-D147.	14.5	915
9	JASPAR 2022: the 9th release of the open-access database of transcription factor binding profiles. Nucleic Acids Research, 2022, 50, D165-D173.	14.5	902
10	VPS35 Mutations in Parkinson Disease. American Journal of Human Genetics, 2011, 89, 162-167.	6.2	747
11	Global mapping of binding sites for Nrf2 identifies novel targets in cell survival response through ChIP-Seq profiling and network analysis. Nucleic Acids Research, 2010, 38, 5718-5734.	14.5	653
12	JASPAR 2010: the greatly expanded open-access database of transcription factor binding profiles. Nucleic Acids Research, 2010, 38, D105-D110.	14.5	529
13	Human-mouse genome comparisons to locate regulatory sites. Nature Genetics, 2000, 26, 225-228.	21.4	440
14	ConSite: web-based prediction of regulatory elements using cross-species comparison. Nucleic Acids Research, 2004, 32, W249-W252.	14.5	388
15	Identification of regulatory regions which confer muscle-specific gene expression. Journal of Molecular Biology, 1998, 278, 167-181.	4.2	362
16	oPOSSUM: identification of over-represented transcription factor binding sites in co-expressed genes. Nucleic Acids Research, 2005, 33, 3154-3164.	14.5	360
17	oPOSSUM-3: Advanced Analysis of Regulatory Motif Over-Representation Across Genes or ChIP-Seq Datasets. G3: Genes, Genomes, Genetics, 2012, 2, 987-1002.	1.8	293
18	DNAJC13 mutations in Parkinson disease. Human Molecular Genetics, 2014, 23, 1794-1801.	2.9	258

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19	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	27.0	254
20	ORegAnno: an open-access community-driven resource for regulatory annotation. Nucleic Acids Research, 2007, 36, D107-D113.	14.5	227
21	Identification of conserved regulatory elements by comparative genome analysis. Journal of Biology, 2003, 2, 13.	2.7	222
22	A new generation of JASPAR, the open-access repository for transcription factor binding site profiles. Nucleic Acids Research, 2006, 34, D95-D97.	14.5	200
23	TFCat: the curated catalog of mouse and human transcription factors. Genome Biology, 2009, 10, R29.	9.6	193
24	A Predictive Model for Regulatory Sequences Directing Liver-Specific Transcription. Genome Research, 2001, 11, 1559-1566.	5.5	179
25	Dynamics of the yeast transcriptome during wine fermentation reveals a novel fermentation stress response. FEMS Yeast Research, 2008, 8, 35-52.	2.3	173
26	The Next Generation of Transcription Factor Binding Site Prediction. PLoS Computational Biology, 2013, 9, e1003214.	3.2	160
27	Constrained Binding Site Diversity within Families of Transcription Factors Enhances Pattern Discovery Bioinformatics. Journal of Molecular Biology, 2004, 338, 207-215.	4.2	157
28	TFBS: Computational framework for transcription factor binding site analysis. Bioinformatics, 2002, 18, 1135-1136.	4.1	154
29	oPOSSUM: integrated tools for analysis of regulatory motif over-representation. Nucleic Acids Research, 2007, 35, W245-W252.	14.5	148
30	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
31	Prediction of Nuclear Hormone Receptor Response Elements. Molecular Endocrinology, 2005, 19, 595-606.	3.7	124
32	DNA Shape Features Improve Transcription Factor Binding Site Predictions InÂVivo. Cell Systems, 2016, 3, 278-286.e4.	6.2	119
33	Deep Feature Selection: Theory and Application to Identify Enhancers and Promoters. Journal of Computational Biology, 2016, 23, 322-336.	1.6	118
34	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. Genome Biology, 2020, 21, 102.	8.8	114
35	A SNP in the HTT promoter alters NF-Î¹B binding and is a bidirectional genetic modifier of Huntington disease. Nature Neuroscience, 2015, 18, 807-816.	14.8	113
36	TFBSshape: a motif database for DNA shape features of transcription factor binding sites. Nucleic Acids Research, 2014, 42, D148-D155.	14.5	111

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37	FLAGS, frequently mutated genes in public exomes. BMC Medical Genomics, 2014, 7, 64.	1.5	108
38	The Transcription Factor Encyclopedia. Genome Biology, 2012, 13, R24.	9.6	103
39	Identification of altered cis-regulatory elements in human disease. Trends in Genetics, 2015, 31, 67-76.	6.7	99
40	The PAZAR database of gene regulatory information coupled to the ORCA toolkit for the study of regulatory sequences. Nucleic Acids Research, 2009, 37, D54-D60.	14.5	97
41	Transcriptional repression of microRNA genes by PML-RARA increases expression of key cancer proteins in acute promyelocytic leukemia. Blood, 2009, 113, 412-421.	1.4	97
42	The SIN3A histone deacetylase complex is required for a complete transcriptional response to hypoxia. Nucleic Acids Research, 2018, 46, 120-133.	14.5	96
43	In Silico Detection of Sequence Variations Modifying Transcriptional Regulation. PLoS Computational Biology, 2008, 4, e5.	3.2	94
44	Exome sequencing identifies mutations in <scp>KIF14</scp> as a novel cause of an autosomal recessive lethal fetal ciliopathy phenotype. Clinical Genetics, 2014, 86, 220-228.	2.0	92
45	PAZAR: a framework for collection and dissemination of cis-regulatory sequence annotation. Genome Biology, 2007, 8, R207.	9.6	89
46	Genome-wide prediction of cis-regulatory regions using supervised deep learning methods. BMC Bioinformatics, 2018, 19, 202.	2.6	88
47	Decoding Human Regulatory Circuits. Genome Research, 2004, 14, 1967-1974.	5.5	86
48	Mitochondrial Carbonic Anhydrase VA Deficiency Resulting from CA5A Alterations Presents with Hyperammonemia in Early Childhood. American Journal of Human Genetics, 2014, 94, 453-461.	6.2	82
49	A regulatory toolbox of MiniPromoters to drive selective expression in the brain. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 16589-16594.	7.1	74
50	Mechanisms underlying p53 regulation of PIK3CA transcription in ovarian surface epithelium and in ovarian cancer. Journal of Cell Science, 2008, 121, 664-674.	2.0	72
51	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in GLS. New England Journal of Medicine, 2019, 380, 1433-1441.	27.0	71
52	The genotypic and phenotypic spectrum of PIGA deficiency. Orphanet Journal of Rare Diseases, 2015, 10, 23.	2.7	70
53	rAAV-compatible MiniPromoters for restricted expression in the brain and eye. Molecular Brain, 2016, 9, 52.	2.6	69
54	Knowledge base and mini-expert platform for the diagnosis of inborn errors of metabolism. Genetics in Medicine, 2018, 20, 151-158.	2.4	67

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55	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	7.6	67
56	MIR@NT@N: a framework integrating transcription factors, microRNAs and their targets to identify sub-network motifs in a meta-regulation network model. BMC Bioinformatics, 2011, 12, 67.	2.6	64
57	Spread of X-chromosome inactivation into autosomal sequences: role for DNA elements, chromatin features and chromosomal domains. Human Molecular Genetics, 2014, 23, 1211-1223.	2.9	60
58	Non-targeted transcription factors motifs are a systemic component of ChIP-seq datasets. Genome Biology, 2014, 15, 412.	8.8	59
59	Regulog Analysis: Detection of Conserved Regulatory Networks Across Bacteria: Application to Staphylococcus aureus. Genome Research, 2004, 14, 1362-1373.	5.5	58
60	Assessment of the ExAC data set for the presence of individuals with pathogenic genotypes implicated in severe Mendelian pediatric disorders. Genetics in Medicine, 2017, 19, 1300-1308.	2.4	58
61	DNA methylation profiling in human Huntington's disease brain. Human Molecular Genetics, 2016, 25, 2013-2030.	2.9	56
62	The role of the clinician in the multiâ€omics era: are you ready?. Journal of Inherited Metabolic Disease, 2018, 41, 571-582.	3.6	55
63	MSCAN: identification of functional clusters of transcription factor binding sites. Nucleic Acids Research, 2004, 32, W195-W198.	14.5	52
64	The identification of cis-regulatory elements: A review from a machine learning perspective. BioSystems, 2015, 138, 6-17.	2.0	51
65	GeneLynx: A Gene-Centric Portal to the Human Genome. Genome Research, 2001, 11, 2151-2157.	5.5	49
66	Improving analysis of transcription factor binding sites within ChIP-Seq data based on topological motif enrichment. BMC Genomics, 2014, 15, 472.	2.8	47
67	Atypical cerebral palsy: genomics analysis enables precision medicine. Genetics in Medicine, 2019, 21, 1621-1628.	2.4	47
68	DeepCAGE Transcriptomics Reveal an Important Role of the Transcription Factor MAFB in the Lymphatic Endothelium. Cell Reports, 2015, 13, 1493-1504.	6.4	46
69	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
70	Targeted CNS delivery using human MiniPromoters and demonstrated compatibility with adeno-associated viral vectors. Molecular Therapy - Methods and Clinical Development, 2014, 1, 5.	4.1	44
71	On the identification of potential regulatory variants within genome wide association candidate SNP sets. BMC Medical Genomics, 2014, 7, 34.	1.5	43
72	TFEA.ChIP: a tool kit for transcription factor binding site enrichment analysis capitalizing on ChIP-seq datasets. Bioinformatics, 2019, 35, 5339-5340.	4.1	41

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73	Secondary neurotransmitter deficiencies in epilepsy caused by voltage-gated sodium channelopathies: A potential treatment target?. Molecular Genetics and Metabolism, 2016, 117, 42-48.	1.1	40
74	De novo dominant variants affecting the motor domain of KIF1A are a cause of PEHO syndrome. European Journal of Human Genetics, 2016, 24, 949-953.	2.8	37
75	Indigenous Genomic Databases: Pragmatic Considerations and Cultural Contexts. Frontiers in Public Health, 2020, 8, 111.	2.7	37
76	Cis-regulatory somatic mutations and gene-expression alteration in B-cell lymphomas. Genome Biology, 2015, 16, 84.	8.8	36
77	Sialic acid catabolism by N-acetylneuraminate pyruvate lyase is essential for muscle function. JCI Insight, 2018, 3, .	5.0	36
78	Strabismus genetics across a spectrum of eye misalignment disorders. Clinical Genetics, 2014, 86, 103-111.	2.0	35
79	Evaluating the impact of single nucleotide variants on transcription factor binding. Nucleic Acids Research, 2016, 44, gkw691.	14.5	35
80	Integration of genomics and metabolomics for prioritization of rare disease variants: a 2018 literature review. Journal of Inherited Metabolic Disease, 2018, 41, 435-445.	3.6	35
81	YY1 binding association with sex-biased transcription revealed through X-linked transcript levels and allelic binding analyses. Scientific Reports, 2016, 6, 37324.	3.3	32
82	Deep Feature Selection: Theory and Application to Identify Enhancers and Promoters. Lecture Notes in Computer Science, 2015, , 205-217.	1.3	32
83	In silico identification of metazoan transcriptional regulatory regions. Die Naturwissenschaften, 2003, 90, 156-166.	1.6	31
84	RMND1 deficiency associated with neonatal lactic acidosis, infantile onset renal failure, deafness, and multiorgan involvement. European Journal of Human Genetics, 2015, 23, 1301-1307.	2.8	28
85	Integrated analysis of yeast regulatory sequences for biologically linked clusters of genes. Functional and Integrative Genomics, 2003, 3, 125-134.	3.5	27
86	Single point mutation in Rabenosyn-5 in a female with intractable seizures and evidence of defective endocytotic trafficking. Orphanet Journal of Rare Diseases, 2014, 9, 141.	2.7	26
87	Deregulated Regulators: Disease-Causing cis Variants in Transcription Factor Genes. Trends in Genetics, 2020, 36, 523-539.	6.7	26
88	Identification of a set of genes showing regionally enriched expression in the mouse brain. BMC Neuroscience, 2008, 9, 66.	1.9	25
89	Inferring novel gene-disease associations using Medical Subject Heading Over-representation Profiles. Genome Medicine, 2012, 4, 75.	8.2	25
90	Quantitative biomedical annotation using medical subject heading over-representation profiles (MeSHOPs). BMC Bioinformatics, 2012, 13, 249.	2.6	24

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91	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	1.1	24
92	Biologically relevant transfer learning improves transcription factor binding prediction. Genome Biology, 2021, 22, 280.	8.8	24
93	Cytosolic phosphoenolpyruvate carboxykinase deficiency presenting with acute liver failure following gastroenteritis. Molecular Genetics and Metabolism, 2016, 118, 21-27.	1.1	23
94	Cross-species examination of X-chromosome inactivation highlights domains of escape from silencing. Epigenetics and Chromatin, 2021, 14, 12.	3.9	23
95	Towards resolving the transcription factor network controlling myelin gene expression. Nucleic Acids Research, 2011, 39, 7974-7991.	14.5	22
96	New MiniPromoter Ple345 (NEFL) Drives Strong and Specific Expression in Retinal Ganglion Cells of Mouse and Primate Retina. Human Gene Therapy, 2019, 30, 257-272.	2.7	21
97	Improvement of Self-Injury With Dopamine and Serotonin Replacement Therapy in a Patient With a Hemizygous PAK3 Mutation: A New Therapeutic Strategy for Neuropsychiatric Features of an Intellectual Disability Syndrome. Journal of Child Neurology, 2018, 33, 106-113.	1.4	20
98	c-Myc is a novel Leishmania virulence factor by proxy that targets the host miRNA system and is essential for survival in human macrophages. Journal of Biological Chemistry, 2018, 293, 12805-12819.	3.4	20
99	Utilizing Social Media to Study Information-Seeking and Ethical Issues in Gene Therapy. Journal of Medical Internet Research, 2013, 15, e44.	4.3	20
100	Defects in fatty acid amide hydrolase 2 in a male with neurologic and psychiatric symptoms. Orphanet Journal of Rare Diseases, 2015, 10, 38.	2.7	19
101	Gene expression models based on transcription factor binding events confer insight into functional cis-regulatory variants. Bioinformatics, 2019, 35, 2610-2617.	4.1	19
102	Bone health and <scp>SATB2</scp>â€associated syndrome. Clinical Genetics, 2018, 93, 588-594.	2.0	18
103	Gain-of-function KCNJ6 Mutation in a Severe Hyperkinetic Movement Disorder Phenotype. Neuroscience, 2018, 384, 152-164.	2.3	18
104	Human MiniPromoters for ocular-rAAV expression in ON bipolar, cone, corneal, endothelial, MÃ¼ller glial, and PAX6 cells. Gene Therapy, 2021, 28, 351-372.	4.5	18
105	Identification of cis-regulatory sequence variations in individual genome sequences. Genome Medicine, 2011, 3, 65.	8.2	17
106	PAX6 MiniPromoters drive restricted expression from rAAV in the adult mouse retina. Molecular Therapy - Methods and Clinical Development, 2016, 3, 16051.	4.1	17
107	Twenty-Seven Tamoxifen-Inducible iCre-Driver Mouse Strains for Eye and Brain, Including Seventeen Carrying a New Inducible-First Constitutive-Ready Allele. Genetics, 2019, 211, 1155-1177.	2.9	17
108	GeneYenta: A PhenotypeÂBased Rare Disease Case Matching Tool Based on Online Dating Algorithms for the Acceleration of Exome Interpretation. Human Mutation, 2015, 36, 432-438.	2.5	16

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109	Human Enhancers Harboring Specific Sequence Composition, Activity, and Genome Organization Are Linked to the Immune Response. Genetics, 2018, 209, 1055-1071.	2.9	16
110	Usability study of clinical exome analysis software: Top lessons learned and recommendations. Journal of Biomedical Informatics, 2014, 51, 129-136.	4.3	15
111	DeepCAGE transcriptomics identify HOXD10 as transcription factor regulating lymphatic endothelial responses to VEGF-C. Journal of Cell Science, 2016, 129, 2573-85.	2.0	15
112	Impact of next-generation sequencing on diagnosis and management of neurometabolic disorders: current advances and future perspectives. Expert Review of Molecular Diagnostics, 2017, 17, 307-309.	3.1	15
113	Further Validation of the SIGMAR1 c.151+1G>T Mutation as Cause of Distal Hereditary Motor Neuropathy. Child Neurology Open, 2016, 3, 2329048X1666991.	1.1	14
114	A girl with developmental delay, ataxia, cranial nerve palsies, severe respiratory problems in infancyâ€"Expanding NDST1 syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 712-715.	1.2	14
115	Identification of a large intronic transposal insertion in SLC17A5 causing sialic acid storage disease. Orphanet Journal of Rare Diseases, 2017, 12, 28.	2.7	14
116	Multi-Omic Approach to Identify Phenotypic Modifiers Underlying Cerebral Demyelination in X-Linked Adrenoleukodystrophy. Frontiers in Cell and Developmental Biology, 2020, 8, 520.	3.7	14
117	CAGEd-oPOSSUM: motif enrichment analysis from CAGE-derived TSSs. Bioinformatics, 2016, 32, 2858-2860.	4.1	13
118	metPropagate: network-guided propagation of metabolomic information for prioritization of metabolic disease genes. Npj Genomic Medicine, 2020, 5, 25.	3.8	13
119	A de novo mosaic mutation in SPAST with two novel alternative alleles and chromosomal copy number variant in a boy with spastic paraplegia and autism spectrum disorder. European Journal of Medical Genetics, 2017, 60, 548-552.	1.3	12
120	Expansion of the QARS deficiency phenotype with report of a family with isolated supratentorial brain abnormalities. Neurogenetics, 2015, 16, 145-149.	1.4	11
121	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo OPA3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a001156.	1.2	11
122	Identification of novel cerebellar developmental transcriptional regulators with motif activity analysis. BMC Genomics, 2019, 20, 718.	2.8	11
123	Discovery of widespread transcription initiation at microsatellites predictable by sequence-based deep neural network. Nature Communications, 2021, 12, 3297.	12.8	11
124	MANTA2, update of the Mongo database for the analysis of transcription factor binding site alterations. Scientific Data, 2018, 5, 180141.	5.3	11
125	Laboratory Animal Management Assistant (LAMA): a LIMS for active research colonies. Mammalian Genome, 2010, 21, 224-230.	2.2	10
126	Compensating for literature annotation bias when predicting novel drug-disease relationships through Medical Subject Heading Over-representation Profile (MeSHOP) similarity. BMC Medical Genomics, 2013, 6, S3.	1.5	10

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127	A case of splenomegaly in CBL syndrome. European Journal of Medical Genetics, 2017, 60, 374-379.	1.3	10
128	Gene Characterization Index: Assessing the Depth of Gene Annotation. PLoS ONE, 2008, 3, e1440.	2.5	9
129	Combined serial analysis of gene expression and transcription factor binding site prediction identifies novel-candidate-target genes of Nr2e1 in neocortex development. BMC Genomics, 2015, 16, 545.	2.8	9
130	Dynamic software design for clinical exome and genome analyses: insights from bioinformaticians, clinical geneticists, and genetic counselors. Journal of the American Medical Informatics Association: JAMIA, 2016, 23, 257-268.	4.4	9
131	Validation of Skeletal Muscle cis-Regulatory Module Predictions Reveals Nucleotide Composition Bias in Functional Enhancers. PLoS Computational Biology, 2011, 7, e1002256.	3.2	8
132	Identification of non-coding genetic variants in samples from hypoxemic respiratory disease patients that affect the transcriptional response to hypoxia. Nucleic Acids Research, 2016, 44, gkw811.	14.5	8
133	Human progranulin-expressing mice as a novel tool for the development of progranulin-modulating therapeutics. Neurobiology of Disease, 2021, 153, 105314.	4.4	8
134	The genome atlas: navigating a new era of reference genomes. Trends in Genetics, 2021, 37, 807-818.	6.7	8
135	CuboCube: Student creation of a cancer genetics e-textbook using open-access software for social learning. PLoS Biology, 2017, 15, e2001192.	5.6	6
136	Genome sequencing reveals a novel genetic mechanism underlying dihydropyrimidine dehydrogenase deficiency: A novel missense variant c.1700G>A and a large intragenic inversion in DPYD spanning intron 8 to intron 12. Human Mutation, 2018, 39, 947-953.	2.5	6
137	Adult GAMT deficiency: A literature review and report of two siblings. Molecular Genetics and Metabolism Reports, 2021, 27, 100761.	1.1	6
138	Human complete NFAT1 deficiency causes a triad ofÂjoint contractures, osteochondromas, and B-cellÂmalignancy. Blood, 2022, 140, 1858-1874.	1.4	6
139	The Gene Set Builder: collation, curation, and distribution of sets of genes. BMC Bioinformatics, 2005, 6, 305.	2.6	5
140	Mitochondrial Complex III Deficiency with Ketoacidosis and Hyperglycemia Mimicking Neonatal Diabetes. JIMD Reports, 2016, 31, 57-62.	1.5	5
141	Textâ€based phenotypic profiles incorporating biochemical phenotypes of inborn errors of metabolism improve phenomicsâ€based diagnosis. Journal of Inherited Metabolic Disease, 2018, 41, 555-562.	3.6	5
142	Curation and bioinformatic analysis of strabismus genes supports functional heterogeneity and proposes candidate genes with connections to RASopathies. Gene, 2019, 697, 213-226.	2.2	5
143	Non-coding-regulatory regions of human brain genes delineated by bacterial artificial chromosome knock-in mice. BMC Biology, 2013, 11, 106.	3.8	4
144	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. Pediatric Neurology, 2019, 100, 87-91.	2.1	4

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145	Discovery and Expansion of Gene Modules by Seeking Isolated Groups in a Random Graph Process. PLoS ONE, 2008, 3, e3358.	2.5	3
146	The variability conundrum in neurometabolic degenerative diseases. Molecular Genetics and Metabolism, 2020, 131, 367-369.	1.1	3
147	GeneBreaker: Variant simulation to improve the diagnosis of Mendelian rare genetic diseases. Human Mutation, 2021, 42, 346-358.	2.5	3
148	Portal for Families Overcoming Neurodevelopmental Disorders (PFOND): Implementation of a Software Framework for Facilitated Community Website Creation by Nontechnical Volunteers. JMIR Research Protocols, 2013, 2, e25.	1.0	3
149	Computational Analysis of Transcriptional Regulation Sites at the HTT Gene Locus. Journal of Huntington's Disease, 2018, 7, 223-237.	1.9	2
150	Development and user evaluation of a rare disease gene prioritization workflow based on cognitive ergonomics. Journal of the American Medical Informatics Association: JAMIA, 2019, 26, 124-133.	4.4	2
151	Exome sequencing enables diagnosis of X-linked hypohidrotic ectodermal dysplasia in patient with eosinophilic esophagitis and severe atopy. Allergy, Asthma and Clinical Immunology, 2021, 17, 9.	2.0	2
152	Introduction to Genomic Analysis Workshop: A catalyst for engaging life-science researchers in high throughput analysis. F1000Research, 2019, 8, 1221.	1.6	2
153	Linkage analysis identifies an isolated strabismus locus at 14q12 overlapping with FOXG1 syndrome region. Journal of Medical Genetics, 2022, 59, 46-55.	3.2	2
154	IDENTIFICATION OF OVER-REPRESENTED COMBINATIONS OF TRANSCRIPTION FACTOR BINDING SITES IN SETS OF CO-EXPRESSED GENES. , 2005, , .		1
155	Secondary biogenic amine deficiencies: genetic etiology, therapeutic interventions, and clinical effects. Neurogenetics, 2021, 22, 251-262.	1.4	1
156	Knowledge Base of Inborn Errors of Metabolism (IEMbase): A Practical Approach. , 2022, , 1449-1455.		1
157	Exome sequencing pilot study in children with carbamazepineâ€induced serious skin reactions. Clinical and Translational Allergy, 2014, 4, P119.	3.2	0
158	Demonstrating the utility of flexible sequence queries against indexed short reads with FlexTyper. PLoS Computational Biology, 2021, 17, e1008815.	3.2	0
159	Targeting AXL Kinase Sensitizes Acute Myeloid Leukemia Stem and Progenitor Cells to Venetoclax Treatment. Blood, 2020, 136, 20-20.	1.4	0
160	RevUP: an online scoring system for regulatory variants implicated in rare diseases. Bioinformatics, 2022, 38, 2664-2666.	4.1	0