Melanie Bahlo

List of Publications by Year
in descending order

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Version: 2024-02-01

207 papers	14,948 citations	20817 60 h-index	23533 111 g-index
264 all docs	264 docs citations	264 times ranked	23882 citing authors

#	Article	IF	Citations
1	IL28B is associated with response to chronic hepatitis C interferon-Î± and ribavirin therapy. Nature Genetics, 2009, 41, 1100-1104.	21.4	1,808
2	Development of plasmacytoid and conventional dendritic cell subtypes from single precursor cells derived in vitro and in vivo. Nature Immunology, 2007, 8, 1217-1226.	14.5	713
3	Iron-Overloadâ€"Related Disease inHFEHereditary Hemochromatosis. New England Journal of Medicine, 2008, 358, 221-230.	27.0	649
4	Genome-wide association study identifies new multiple sclerosis susceptibility loci on chromosomes 12 and 20. Nature Genetics, 2009, 41, 824-828.	21.4	501
5	Strikingly Different Clinicopathological Phenotypes Determined by Progranulin-Mutation Dosage. American Journal of Human Genetics, 2012, 90, 1102-1107.	6.2	414
6	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. Nature Genetics, 2012, 44, 1188-1190.	21.4	333
7	Genomeâ€wide metaâ€analysis identifies novel multiple sclerosis susceptibility loci. Annals of Neurology, 2011, 70, 897-912.	5.3	314
8	Kufs Disease, the Major Adult Form of Neuronal Ceroid Lipofuscinosis, Caused by Mutations in CLN6. American Journal of Human Genetics, 2011, 88, 566-573.	6.2	253
9	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.	6.2	237
10	Array-Based Gene Discovery with Three Unrelated Subjects Shows SCARB2/LIMP-2 Deficiency Causes Myoclonus Epilepsy and Glomerulosclerosis. American Journal of Human Genetics, 2008, 82, 673-684.	6.2	230
11	The Tasmanian Devil Transcriptome Reveals Schwann Cell Origins of a Clonally Transmissible Cancer. Science, 2010, 327, 84-87.	12.6	222
12	Mutation of the Mitochondrial Tyrosyl-tRNA Synthetase Gene, YARS2, Causes Myopathy, Lactic Acidosis, and Sideroblastic Anemiaâ€"MLASA Syndrome. American Journal of Human Genetics, 2010, 87, 52-59.	6.2	211
13	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with Î±-Synuclein Pathology. American Journal of Human Genetics, 2014, 95, 729-735.	6.2	207
14	Genomewide Linkage Study in 1,176 Affected Sister Pair Families Identifies a Significant Susceptibility Locus for Endometriosis on Chromosome 10q26. American Journal of Human Genetics, 2005, 77, 365-376.	6.2	200
15	Inference from Gene Trees in a Subdivided Population. Theoretical Population Biology, 2000, 57, 79-95.	1.1	193
16	An Î±â€Eâ€catenin (<scp>CTNNA1</scp>) mutation in hereditary diffuse gastric cancer. Journal of Pathology, 2013, 229, 621-629.	4.5	184
17	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. American Journal of Human Genetics, 2019, 105, 151-165.	6.2	170
18	Identification and Analysis of Error Types in High-Throughput Genotyping. American Journal of Human Genetics, 2000, 67, 727-736.	6.2	166

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19	A Mutation in the Golgi Qb-SNARE Gene GOSR2 Causes Progressive Myoclonus Epilepsy with Early Ataxia. American Journal of Human Genetics, 2011, 88, 657-663.	6.2	166
20	Serine and Lipid Metabolism in Macular Disease and Peripheral Neuropathy. New England Journal of Medicine, 2019, 381, 1422-1433.	27.0	166
21	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. Nature Genetics, 2014, 46, 1239-1244.	21.4	165
22	A Ca_v3.2 T-Type Calcium Channel Point Mutation Has Splice-Variant-Specific Effects on Function and Segregates with Seizure Expression in a Polygenic Rat Model of Absence Epilepsy. Journal of Neuroscience, 2009, 29, 371-380.	3.6	164
23	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. F1000Research, 2018, 7, 1297.	1.6	157
24	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. F1000Research, 2018, 7, 1297.	1.6	131
25	Mutations in LOXHD1, an Evolutionarily Conserved Stereociliary Protein, Disrupt Hair Cell Function in Mice and Cause Progressive Hearing Loss in Humans. American Journal of Human Genetics, 2009, 85, 328-337.	6.2	129
26	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	7.6	129
27	Genome-Wide Linkage Analysis of the Acute Coronary Syndrome Suggests a Locus on Chromosome 2. Arteriosclerosis, Thrombosis, and Vascular Biology, 2002, 22, 874-878.	2.4	125
28	Human and Mouse Mutations in WDR35 Cause Short-Rib Polydactyly Syndromes Due to Abnormal Ciliogenesis. American Journal of Human Genetics, 2011, 88, 508-515.	6.2	122
29	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	2.9	120
30	Polycomb Repressive Complex 2 (PRC2) Restricts Hematopoietic Stem Cell Activity. PLoS Biology, 2008, 6, e93.	5.6	118
31	A cross-platform approach identifies genetic regulators of human metabolism and health. Nature Genetics, 2021, 53, 54-64.	21.4	117
32	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator NPRL3. Annals of Neurology, 2016, 79, 132-137.	5.3	116
33	The Genetics of Epilepsy. Annual Review of Genomics and Human Genetics, 2020, 21, 205-230.	6.2	116
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 Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in TBC1D24. American Journal of Human Genetics, 2010, 87, 371-375.	6.2	111
36	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	12.8	111

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37	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	6.2	108
38	High-resolution characterization of sequence signatures due to non-random cleavage of cell-free DNA. BMC Medical Genomics, 2015, 8, 29.	1.5	107
39	Dominantly inherited ataxia and dysphonia with dentate calcification: spinocerebellar ataxia type 20. Brain, 2004, 127, 1172-1181.	7.6	106
40	A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development. Molecular Psychiatry, 2019, 24, 1065-1078.	7.9	106
41	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. Human Molecular Genetics, 2013, 22, 1417-1423.	2.9	105
42	Genome-wide analyses identify common variants associated with macular telangiectasia type 2. Nature Genetics, 2017, 49, 559-567.	21.4	105
43	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. Journal of Inherited Metabolic Disease, 2017, 40, 261-269.	3.6	101
44	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
45	dtangle: accurate and robust cell type deconvolution. Bioinformatics, 2019, 35, 2093-2099.	4.1	98
46	Familial cortical dysplasia type <scp>IIA</scp> caused by a germline mutation in <scp>DEPDC</scp>5. Annals of Clinical and Translational Neurology, 2015, 2, 575-580.	3.7	95
47	Genetic Dissection of the Human Leukocyte Antigen Region by Use of Haplotypes of Tasmanians with Multiple Sclerosis. American Journal of Human Genetics, 2002, 70, 1125-1137.	6.2	93
48	Detecting Expansions of Tandem Repeats in Cohorts Sequenced with Short-Read Sequencing Data. American Journal of Human Genetics, 2018, 103, 858-873.	6.2	93
49	Identity-by-descent analyses for measuring population dynamics and selection in recombining pathogens. PLoS Genetics, 2018, 14, e1007279.	3.5	86
50	Recent advances in the detection of repeat expansions with short-read next-generation sequencing. F1000Research, 2018, 7, 736.	1.6	84
51	Autosomal-Recessive Congenital Cerebellar Ataxia Is Caused by Mutations in Metabotropic Glutamate Receptor 1. American Journal of Human Genetics, 2012, 91, 553-564.	6.2	81
52	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	8.2	78
53	Small intragenic deletion in FOXP2 associated with childhood apraxia of speech and dysarthria. American Journal of Medical Genetics, Part A, 2013, 161, 2321-2326.	1.2	75
54	A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. Nature Communications, 2020, 11, 3150.	12.8	75

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55	Agm1/Pgm3-Mediated Sugar Nucleotide Synthesis Is Essential for Hematopoiesis and Development. Molecular and Cellular Biology, 2007, 27, 5849-5859.	2.3	73
56	Mutations in Contactin-1, a Neural Adhesion and Neuromuscular Junction Protein, Cause a Familial Form of Lethal Congenital Myopathy. American Journal of Human Genetics, 2008, 83, 714-724.	6.2	72
57	Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. Genome Biology, 2011, 12, R85.	9.6	72
58	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology, 2016, 87, 1975-1984.	1.1	71
59	A Mouse Model of Harlequin Ichthyosis Delineates a Key Role for Abca12 in Lipid Homeostasis. PLoS Genetics, 2008, 4, e1000192.	3.5	70
60	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	21.4	69
61	Severe childhood speech disorder. Neurology, 2020, 94, e2148-e2167.	1.1	68
62	Hemispheric cortical dysplasia secondary to a mosaic somatic mutation in MTOR. Neurology, 2015, 84, 2029-2032.	1.1	64
63	Identification of improved IL28B SNPs and haplotypes for prediction of drug response in treatment of hepatitis C using massively parallel sequencing in a cross-sectional European cohort. Genome Medicine, 2011, 3, 57.	8.2	62
64	Plasmodium vivax Populations Are More Genetically Diverse and Less Structured than Sympatric Plasmodium falciparum Populations. PLoS Neglected Tropical Diseases, 2015, 9, e0003634.	3.0	62
65	CYLD is a causative gene for frontotemporal dementia â€“ amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799.	7.6	62
66	A novel association between a SNP in CYBRD1 and serum ferritin levels in a cohort study of HFE hereditary haemochromatosis. British Journal of Haematology, 2009, 147, 140-149.	2.5	61
67	Dating Rare Mutations from Small Samples with Dense Marker Data. Genetics, 2014, 197, 1315-1327.	2.9	61
68	Generating linkage mapping files from Affymetrix SNP chip data. Bioinformatics, 2009, 25, 1961-1962.	4.1	60
69	A Founder Mutation in PET100 Causes Isolated Complex IV Deficiency in Lebanese Individuals with Leigh Syndrome. American Journal of Human Genetics, 2014, 94, 209-222.	6.2	60
70	Proteomic and Metabolomic Analyses of Mitochondrial Complex I-deficient Mouse Model Generated by Spontaneous B2 Short Interspersed Nuclear Element (SINE) Insertion into NADH Dehydrogenase (Ubiquinone) Fe-S Protein 4 (Ndufs4) Gene. Journal of Biological Chemistry, 2012, 287, 20652-20663.	3.4	58
71	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	6.2	57
72	EIF2S3Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. Human Mutation, 2017, 38, 409-425.	2.5	57

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73	A Polymorphism in the HLA-DPB1 Gene Is Associated with Susceptibility to Multiple Sclerosis. PLoS ONE, 2010, 5, e13454.	2.5	55
74	Saliva-Derived DNA Performs Well in Large-Scale, High-Density Single-Nucleotide Polymorphism Microarray Studies. Cancer Epidemiology Biomarkers and Prevention, 2010, 19, 794-798.	2.5	52
75	Systematic noise degrades gene co-expression signals but can be corrected. BMC Bioinformatics, 2015, 16, 309.	2.6	50
76	ALPK3-deficient cardiomyocytes generated from patient-derived induced pluripotent stem cells and mutant human embryonic stem cells display abnormal calcium handling and establish that ALPK3 deficiency underlies familial cardiomyopathy. European Heart Journal, 2016, 37, 2586-2590.	2.2	49
77	Mutations in the first MyTH4 domain of MYO15A are a common cause of DFNB3 hearing loss. Laryngoscope, 2009, 119, 727-733.	2.0	48
78	Multiple Sclerosis Susceptibility-Associated SNPs Do Not Influence Disease Severity Measures in a Cohort of Australian MS Patients. PLoS ONE, 2010, 5, e10003.	2.5	45
79	Genome-wide linkage scan and association study of PARL to the expression of LHON families in Thailand. Human Genetics, 2010, 128, 39-49.	3.8	43
80	X chromosome association testing in genome wide association studies. Genetic Epidemiology, 2011, 35, 664-670.	1.3	43
81	Structurally conserved erythrocyte-binding domain in Plasmodium provides a versatile scaffold for alternate receptor engagement. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E191-200.	7.1	43
82	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	6.2	43
83	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. Neuron, 2019, 104, 665-679.e8.	8.1	43
84	Blood pressure QTLs identified by genome-wide linkage analysis and dependence on associated phenotypes. Physiological Genomics, 2002, 8, 99-105.	2.3	42
85	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. Brain Communications, 2021, 3, fcaa235.	3.3	42
86	Mutation of the nuclear lamin geneLMNB2in progressive myoclonus epilepsy with early ataxia. Human Molecular Genetics, 2015, 24, 4483-4490.	2.9	41
87	Progressive myoclonus epilepsiesâ€"Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. American Journal of Human Genetics, 2021, 108, 722-738.	6.2	41
88	Extended haplotype analysis in the HLA complex reveals an increased frequency of the HFE-C282Y mutation in individuals with multiple sclerosis. Human Genetics, 2004, 114, 573-580.	3.8	40
89	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genetics, 2018, 14, e1007281.	3.5	40
90	A Mutation in Synaptojanin 2 Causes Progressive Hearing Loss in the ENU-Mutagenised Mouse Strain Mozart. PLoS ONE, 2011, 6, e17607.	2.5	39

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91	A novel splice site mutation in EYA4 causes DFNA10 hearing loss. American Journal of Medical Genetics, Part A, 2007, 143A, 1599-1604.	1.2	38
92	An Ethyl-Nitrosourea-Induced Point Mutation in Phex Causes Exon Skipping, X-Linked Hypophosphatemia, and Rickets. American Journal of Pathology, 2002, 161, 1925-1933.	3.8	37
93	Deficiency of 5-hydroxyisourate hydrolase causes hepatomegaly and hepatocellular carcinoma in mice. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 16625-16630.	7.1	37
94	Cell-specific cis-regulatory elements and mechanisms of non-coding genetic disease in human retina and retinal organoids. Developmental Cell, 2022, 57, 820-836.e6.	7.0	37
95	Familial Adult Myoclonic Epilepsy. Archives of Neurology, 2012, 69, 474.	4.5	36
96	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. Genetics in Medicine, 2019, 21, 948-954.	2.4	36
97	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	6.2	33
98	GNE myopathy in Roma patients homozygous for the p.I618T founder mutation. Neuromuscular Disorders, 2015, 25, 713-718.	0.6	32
99	Clinical spectrum of the pentanucleotide repeat expansion in the RFC1 gene in ataxia syndromes. Neurology, 2020, 95, e2912-e2923.	1.1	32
100	Mutations in SH3PXD2B cause Borrone dermato-cardio-skeletal syndrome. European Journal of Human Genetics, 2014, 22, 741-747.	2.8	30
101	Mitochondrial Genome Sequence of the Scabies Mite Provides Insight into the Genetic Diversity of Individual Scabies Infections. PLoS Neglected Tropical Diseases, 2016, 10, e0004384.	3.0	30
102	Coalescence time for two genes from a subdivided population. Journal of Mathematical Biology, 2001, 43, 397-410.	1.9	29
103	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. Neurology: Genetics, 2015, 1, e16.	1.9	29
104	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	3.8	29
105	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). Neurology, 2016, 87, 579-584.	1.1	28
106	Whole exome sequencing combined with linkage analysis identifies a novel 3â€‰bp deletion in NR5A1. European Journal of Human Genetics, 2015, 23, 486-493.	2.8	27
107	Increasingly inbred and fragmented populations of Plasmodium vivax associated with the eastward decline in malaria transmission across the Southwest Pacific. PLoS Neglected Tropical Diseases, 2018, 12, e0006146.	3.0	27
108	A novel X-linked form of congenital fiber-type disproportion. Annals of Neurology, 2005, 58, 767-772.	5.3	26

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109	An ENU-Induced Mutation of Cdh23 Causes Congenital Hearing Loss, but No Vestibular Dysfunction, in Mice. American Journal of Pathology, 2011, 179, 903-914.	3.8	26
110	Identification of genetic factors influencing metabolic dysregulation and retinal support for MacTel, a retinal disorder. Communications Biology, 2021, 4, 274.	4.4	26
111	Harnessing Gene Expression Networks to Prioritize Candidate Epileptic Encephalopathy Genes. PLoS ONE, 2014, 9, e102079.	2.5	25
112	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	2.9	25
113	Functional analysis of a hypomorphic allele shows that MMP14 catalytic activity is the prime determinant of the Winchester syndrome phenotype. Human Molecular Genetics, 2018, 27, 2775-2788.	2.9	25
114	SNP barcodes provide higher resolution than microsatellite markers to measure Plasmodium vivax population genetics. Malaria Journal, 2020, 19, 375.	2.3	25
115	Clonal multi-omics reveals Bcor as a negative regulator of emergency dendritic cell development. Immunity, 2021, 54, 1338-1351.e9.	14.3	25
116	Spatial centrosome proteome of human neural cells uncovers disease-relevant heterogeneity. Science, 2022, 376, .	12.6	25
117	Investigating and Correcting Plasma DNA Sequencing Coverage Bias to Enhance Aneuploidy Discovery. PLoS ONE, 2014, 9, e86993.	2.5	24
118	Systemic lipid dysregulation is a risk factor for macular neurodegenerative disease. Scientific Reports, 2020, 10, 12165.	3.3	24
119	Early neuroimaging markers of FOXP2 intragenic deletion. Scientific Reports, 2016, 6, 35192.	3.3	23
120	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.1	23
121	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
122	Familial adult myoclonic epilepsy type 1 SAMD12 TTTCA repeat expansion arose 17,000 years ago and is present in Sri Lankan and Indian families. European Journal of Human Genetics, 2020, 28, 973-978.	2.8	23
123	Multiple sclerosis risk variants regulate gene expression in innate and adaptive immune cells. Life Science Alliance, 2020, 3, e202000650.	2.8	22
124	Autosomal dominant congenital spinal muscular atrophy: a true form of spinal muscular atrophy caused by early loss of anterior horn cells. Brain, 2012, 135, 1714-1723.	7.6	21
125	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Annals of Clinical and Translational Neurology, 2015, 2, 821-830.	3.7	21
126	XIBD: software for inferring pairwise identity by descent on the X chromosome. Bioinformatics, 2016, 32, 2389-2391.	4.1	21

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127	Infanticide vs. inherited cardiac arrhythmias. Europace, 2021, 23, 441-450.	1.7	21
128	Cochlear Implants for DFNA17 Deafness. Laryngoscope, 2006, 116, 2211-2215.	2.0	20
129	A novel mutation in COCHâ€"implications for genotypeâ€phenotype correlations in DFNA9 hearing loss. Laryngoscope, 2010, 120, 2489-2493.	2.0	20
130	Autosomal dominant vasovagal syncope. Neurology, 2013, 80, 1485-1493.	1.1	20
131	Use of Copy Number Deletion Polymorphisms to Assess DNA Chimerism. Clinical Chemistry, 2014, 60, 1105-1114.	3.2	20
132	Evaluation of nonâ€coding variation in <scp>GLUT</scp>1 deficiency. Developmental Medicine and Child Neurology, 2016, 58, 1295-1302.	2.1	20
133	Progressive Myoclonus Epilepsies. Neurology: Genetics, 2021, 7, e641.	1.9	20
134	On the utility of data from the International HapMap Project for Australian association studies. Human Genetics, 2006, 119, 220-222.	3.8	19
135	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. Neurology: Genetics, 2016, 2, e51.	1.9	19
136	SNP selection for genes of iron metabolism in a study of genetic modifiers of hemochromatosis. BMC Medical Genetics, 2008, 9, 18.	2.1	18
137	A novel splice site mutation in the RDX gene causes DFNB24 hearing loss in an Iranian family. American Journal of Medical Genetics, Part A, 2009, 149A, 555-558.	1.2	18
138	Heterozygous mutations in HSD17B4 cause juvenile peroxisomal D-bifunctional protein deficiency. Neurology: Genetics, 2016, 2, e114.	1.9	18
139	Recessive Spondylocarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in MYH3. American Journal of Human Genetics, 2018, 102, 1115-1125.	6.2	18
140	Identifying nineteenth century genealogical links from genotypes. Human Genetics, 2005, 117, 188-199.	3.8	17
141	Identical by descent L1CAM mutation in two apparently unrelated families with intellectual disability without L1 syndrome. European Journal of Medical Genetics, 2015, 58, 364-368.	1.3	17
142	Neuropathology of childhoodâ€onset basal ganglia degeneration caused by mutation of VAC14. Annals of Clinical and Translational Neurology, 2017, 4, 859-864.	3.7	17
143	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. Genetics in Medicine, 2019, 21, 2532-2542.	2.4	17
144	Challenges of diagnostic exome sequencing in an inbred founder population. Molecular Genetics & Genomic Medicine, 2013, 1, 71-76.	1.2	16

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145	Dorsal language stream anomalies in an inherited speech disorder. Brain, 2019, 142, 966-977.	7.6	16
146	The clinical utility of exome sequencing and extended bioinformatic analyses in adolescents and adults with a broad range of neurological phenotypes: an Australian perspective. Journal of the Neurological Sciences, 2021, 420, 117260.	0.6	16
147	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
148	Genetic disruption of serine biosynthesis is a key driver of macular telangiectasia type 2 aetiology and progression. Genome Medicine, 2021, 13, 39.	8.2	15
149	Molecular characterization of a novel Xâ€linked syndrome involving developmental delay and deafness. American Journal of Medical Genetics, Part A, 2007, 143A, 2564-2575.	1.2	14
150	A recombination hotspot leads to sequence variability within a novel gene (AK005651) and contributes to type 1 diabetes susceptibility. Genome Research, 2010, 20, 1629-1638.	5.5	14
151	Fatal Enteroviral Encephalitis in a Patient with Common Variable Immunodeficiency Harbouring a Novel Mutation in NFKB2. Journal of Clinical Immunology, 2019, 39, 324-335.	3.8	14
152	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. Epilepsia, 2020, 61, e23-e29.	5.1	14
153	Founder effect of the TTTCA repeat insertions in SAMD12 causing BAFME1. European Journal of Human Genetics, 2021, 29, 343-348.	2.8	14
154	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy. Neurology: Genetics, 2022, 8, e652.	1.9	14
155	Global diversity and balancing selection of 23 leading Plasmodium falciparum candidate vaccine antigens. PLoS Computational Biology, 2022, 18, e1009801.	3.2	14
156	Analysis of extended HLA haplotypes in multiple sclerosis and narcolepsy families confirms a predisposing effect for the class I region in Tasmanian MS patients. Immunogenetics, 2007, 59, 177-186.	2.4	13
157	brain-coX: investigating and visualising gene co-expression in seven human brain transcriptomic datasets. Genome Medicine, 2017, 9, 55.	8.2	13
158	Comparative genomics revealed adaptive admixture in Cryptosporidium hominis in Africa. Microbial Genomics, 2021, 7, .	2.0	13
159	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. Neurology, 2021, 96, e2251-e2260.	1.1	13
160	Comparing the frequency of common genetic variants and haplotypes between carriers and non-carriers of BRCA1 and BRCA2deleterious mutations in Australian women diagnosed with breast cancer before 40 years of age. BMC Cancer, 2010, 10, 466.	2.6	12
161	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. Epilepsy Research, 2017, 131, 9-14.	1.6	12
162	Rapid Diagnosis of Spinocerebellar Ataxia 36 in a <scp>Threeâ€Generation</scp> Family Using <scp>Shortâ€Read Wholeâ€Genome</scp> Sequencing Data. Movement Disorders, 2020, 35, 1675-1679.	3.9	12

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163	Mapping of the Plasmodium chabaudi Resistance Locus char2. Infection and Immunity, 2006, 74, 5814-5819.	2.2	11
164	Candidate disease gene prediction using Gentrepid: application to a genomeâ€wide association study on coronary artery disease. Molecular Genetics & Genomic Medicine, 2014, 2, 44-57.	1.2	11
165	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. Epilepsia Open, 2019, 4, 504-510.	2.4	11
166	Callosal agenesis and congenital mirror movements: outcomes associated with DCC mutations. Developmental Medicine and Child Neurology, 2020, 62, 758-762.	2.1	11
167	PacBio long-read amplicon sequencing enables scalable high-resolution population allele typing of the complex CYP2D6 locus. Communications Biology, 2022, 5, 168.	4.4	11
168	Detecting genome wide haplotype sharing using SNP or microsatellite haplotype data. Human Genetics, 2006, 119, 38-50.	3.8	10
169	Analysis of genome-wide association study data using the protein knowledge base. BMC Genetics, 2011, 12, 98.	2.7	10
170	Using familial information for variant filtering in high-throughput sequencing studies. Human Genetics, 2014, 133, 1331-1341.	3.8	10
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