Melanie Bahlo

List of Publications by Year
in descending order

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207 papers	14,948 citations	23879 60 h-index	26792 111 g-index
264 all docs	264 docs citations	264 times ranked	25926 citing authors

#	Article	IF	Citations
1	Atypical development of Brocaâ€™s area in a large family with inherited stuttering. Brain, 2022, 145, 1177-1188.	3.7	6
2	A family study implicates GBE1 in the etiology of autism spectrum disorder. Human Mutation, 2022, 43, 16-29.	1.1	2
3	Infantileâ€onset myoclonic developmental and epileptic encephalopathy: A new RARS2 phenotype. Epilepsia Open, 2022, 7, 170-180.	1.3	5
4	Genetics of reticular pseudodrusen in age-related macular degeneration. Trends in Genetics, 2022, 38, 312-316.	2.9	1
5	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy. Neurology: Genetics, 2022, 8, e652.	0.9	14
6	Population-level genome-wide STR discovery and validation for population structure and genetic diversity assessment of Plasmodium species. PLoS Genetics, 2022, 18, e1009604.	1.5	8
7	Global diversity and balancing selection of 23 leading Plasmodium falciparum candidate vaccine antigens. PLoS Computational Biology, 2022, 18, e1009801.	1.5	14
8	PacBio long-read amplicon sequencing enables scalable high-resolution population allele typing of the complex CYP2D6 locus. Communications Biology, 2022, 5, 168.	2.0	11
9	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.	3.1	37
10	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	2.6	16
11	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	2.8	8
12	Detecting Tandem Repeat Expansions Using Short-Read Sequencing for Clinical Use. Neuromethods, 2022, , 15-42.	0.2	2
13	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. EBioMedicine, 2022, 81, 104079.	2.7	10
14	Spatial centrosome proteome of human neural cells uncovers disease-relevant heterogeneity. Science, 2022, 376, .	6.0	25
15	Founder effect of the TTTCA repeat insertions in SAMD12 causing BAFME1. European Journal of Human Genetics, 2021, 29, 343-348.	1.4	14
16	Progressive Myoclonus Epilepsy Caused by a Homozygous Splicing Variant of <scp>SLC7A6OS</scp>. Annals of Neurology, 2021, 89, 402-407.	2.8	5
17	Transcriptome analysis of a ring chromosome 20 patient cohort. Epilepsia, 2021, 62, e22-e28.	2.6	5
18	Infanticide vs. inherited cardiac arrhythmias. Europace, 2021, 23, 441-450.	0.7	21

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19	A cross-platform approach identifies genetic regulators of human metabolism and health. Nature Genetics, 2021, 53, 54-64.	9.4	117
20	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.	0.3	16
21	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. Brain Communications, 2021, 3, fcaa235.	1.5	42
22	Comparative genomics revealed adaptive admixture in Cryptosporidium hominis in Africa. Microbial Genomics, 2021, 7, .	1.0	13
23	Contribution of rare genetic variants to drug response in absence epilepsy. Epilepsy Research, 2021, 170, 106537.	0.8	9
24	Genetic disruption of serine biosynthesis is a key driver of macular telangiectasia type 2 aetiology and progression. Genome Medicine, 2021, 13, 39.	3.6	15
25	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. Neurology, 2021, 96, e2251-e2260.	1.5	13
26	Identification of genetic factors influencing metabolic dysregulation and retinal support for MacTel, a retinal disorder. Communications Biology, 2021, 4, 274.	2.0	26
27	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.	2.6	41
28	Expanding the clinical and radiological phenotypes of leukoencephalopathy due to biallelic <scp>HMBS</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 2941-2950.	0.7	2
29	Clonal multi-omics reveals Bcor as a negative regulator of emergency dendritic cell development. Immunity, 2021, 54, 1338-1351.e9.	6.6	25
30	Cutting edge approaches to detecting brain mosaicism associated with common focal epilepsies: implications for diagnosis and potential therapies. Expert Review of Neurotherapeutics, 2021, 21, 1309-1316.	1.4	5
31	A survey of RNA editing at single-cell resolution links interneurons to schizophrenia and autism. Rna, 2021, 27, 1482-1496.	1.6	7
32	Progressive Myoclonus Epilepsies. Neurology: Genetics, 2021, 7, e641.	0.9	20
33	Deletions in VANGL1 are a risk factor for antibody-mediated kidney disease. Cell Reports Medicine, 2021, 2, 100475.	3.3	2
34	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	2.6	33
35	Systemic lipid dysregulation is a risk factor for macular neurodegenerative disease. Scientific Reports, 2020, 10, 12165.	1.6	24
36	SNP barcodes provide higher resolution than microsatellite markers to measure Plasmodium vivax population genetics. Malaria Journal, 2020, 19, 375.	0.8	25

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37	Clinical spectrum of the pentanucleotide repeat expansion in the RFC1 gene in ataxia syndromes. Neurology, 2020, 95, e2912-e2923.	1.5	32
38	Tracing Autism Traits in Large Multiplex Families to Identify Endophenotypes of the Broader Autism Phenotype. International Journal of Molecular Sciences, 2020, 21, 7965.	1.8	3
39	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.	2.2	12
40	A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. Nature Communications, 2020, 11, 3150.	5.8	75
41	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.	1.4	23
42	CYLD is a causative gene for frontotemporal dementia â€“ amyotrophic lateral sclerosis. Brain, 2020, 143, 783-799.	3.7	62
43	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. Epilepsia, 2020, 61, e23-e29.	2.6	14
44	Callosal agenesis and congenital mirror movements: outcomes associated with DCC mutations. Developmental Medicine and Child Neurology, 2020, 62, 758-762.	1.1	11
45	Severe childhood speech disorder. Neurology, 2020, 94, e2148-e2167.	1.5	68
46	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. Genome Biology, 2020, 21, 102.	3.8	114
47	The Genetics of Epilepsy. Annual Review of Genomics and Human Genetics, 2020, 21, 205-230.	2.5	116
48	Multiple sclerosis risk variants regulate gene expression in innate and adaptive immune cells. Life Science Alliance, 2020, 3, e202000650.	1.3	22
49	Generation of seven iPSC lines from peripheral blood mononuclear cells suitable to investigate Autism Spectrum Disorder. Stem Cell Research, 2019, 39, 101516.	0.3	4
50	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
51	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
52	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. Epilepsia Open, 2019, 4, 504-510.	1.3	11
53	Serine and Lipid Metabolism in Macular Disease and Peripheral Neuropathy. New England Journal of Medicine, 2019, 381, 1422-1433.	13.9	166
54	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. Neuron, 2019, 104, 665-679.e8.	3.8	43

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55	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.	2.6	170
56	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. Genetics in Medicine, 2019, 21, 2532-2542.	1.1	17
57	Familial early onset Parkinson's disease caused by a homozygous frameshift variant in PARK7: Clinical features and literature update. Parkinsonism and Related Disorders, 2019, 64, 308-311.	1.1	7
58	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	2.6	23
59	Fatal Enteroviral Encephalitis in a Patient with Common Variable Immunodeficiency Harbouring a Novel Mutation in NFKB2. Journal of Clinical Immunology, 2019, 39, 324-335.	2.0	14
60	Dorsal language stream anomalies in an inherited speech disorder. Brain, 2019, 142, 966-977.	3.7	16
61	Genetic Analysis of Patients Who Experienced Awareness with Recall while under General Anesthesia. Anesthesiology, 2019, 131, 974-982.	1.3	9
62	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	5.8	111
63	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. Human Mutation, 2019, 40, 374-379.	1.1	7
64	dtangle: accurate and robust cell type deconvolution. Bioinformatics, 2019, 35, 2093-2099.	1.8	98
65	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. Genetics in Medicine, 2019, 21, 948-954.	1.1	36
66	A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development. Molecular Psychiatry, 2019, 24, 1065-1078.	4.1	106
67	Mosaic uniparental disomy results in GM1 gangliosidosis with normal enzyme assay. American Journal of Medical Genetics, Part A, 2018, 176, 230-234.	0.7	3
68	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. F1000Research, 2018, 7, 1297.	0.8	131
69	Genetic investigation into an increased susceptibility to biliary atresia in an extended New Zealand MÄori family. BMC Medical Genomics, 2018, 11, 121.	0.7	7
70	Detecting Expansions of Tandem Repeats in Cohorts Sequenced with Short-Read Sequencing Data. American Journal of Human Genetics, 2018, 103, 858-873.	2.6	93
71	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. F1000Research, 2018, 7, 1297.	0.8	157
72	Identity-by-descent analyses for measuring population dynamics and selection in recombining pathogens. PLoS Genetics, 2018, 14, e1007279.	1.5	86

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73	Recessive Spondylocarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in MYH3. American Journal of Human Genetics, 2018, 102, 1115-1125.	2.6	18
74	Evidence of linkage to chromosome 5p13.2â€q11.1 in a large inbred family with genetic generalized epilepsy. Epilepsia, 2018, 59, e125-e129.	2.6	3
75	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.	1.4	25
76	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genetics, 2018, 14, e1007281.	1.5	40
77	Recent advances in the detection of repeat expansions with short-read next-generation sequencing. F1000Research, 2018, 7, 736.	0.8	84
78	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.	1.3	27
79	EIF2S3Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. Human Mutation, 2017, 38, 409-425.	1.1	57
80	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	9.4	69
81	Genome-wide analyses identify common variants associated with macular telangiectasia type 2. Nature Genetics, 2017, 49, 559-567.	9.4	105
82	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. Epilepsy Research, 2017, 131, 9-14.	0.8	12
83	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. European Journal of Medical Genetics, 2017, 60, 437-443.	0.7	10
84	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. Journal of Inherited Metabolic Disease, 2017, 40, 261-269.	1.7	101
85	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	2.6	43
86	Neuropathology of childhoodâ€onset basal ganglia degeneration caused by mutation of VAC14. Annals of Clinical and Translational Neurology, 2017, 4, 859-864.	1.7	17
87	brain-coX: investigating and visualising gene co-expression in seven human brain transcriptomic datasets. Genome Medicine, 2017, 9, 55.	3.6	13
88	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	1.8	29
89	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	2.6	57
90	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. Neurology: Genetics, 2016, 2, e51.	0.9	19

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91	Evaluation of nonâ€coding variation in <scp>GLUT</scp>1 deficiency. Developmental Medicine and Child Neurology, 2016, 58, 1295-1302.	1.1	20
92	Heterozygous mutations in HSD17B4 cause juvenile peroxisomal D-bifunctional protein deficiency. Neurology: Genetics, 2016, 2, e114.	0.9	18
93	Early neuroimaging markers of FOXP2 intragenic deletion. Scientific Reports, 2016, 6, 35192.	1.6	23
94	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.	1.0	49
95	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology, 2016, 87, 1975-1984.	1.5	71
96	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator NPRL3. Annals of Neurology, 2016, 79, 132-137.	2.8	116
97	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). Neurology, 2016, 87, 579-584.	1.5	28
98	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.5	23
99	XIBD: software for inferring pairwise identity by descent on the X chromosome. Bioinformatics, 2016, 32, 2389-2391.	1.8	21
100	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.	3.3	43
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.	1.3	30
102	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Annals of Clinical and Translational Neurology, 2015, 2, 821-830.	1.7	21
103	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. Neurology: Genetics, 2015, 1, e16.	0.9	29
104	Systematic noise degrades gene co-expression signals but can be corrected. BMC Bioinformatics, 2015, 16, 309.	1.2	50
105	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	3.6	78
106	Mutation of the nuclear lamin geneLMNB2in progressive myoclonus epilepsy with early ataxia. Human Molecular Genetics, 2015, 24, 4483-4490.	1.4	41
107	Hemispheric cortical dysplasia secondary to a mosaic somatic mutation in MTOR. Neurology, 2015, 84, 2029-2032.	1.5	64
108	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.	1.7	95

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109	High-resolution characterization of sequence signatures due to non-random cleavage of cell-free DNA. BMC Medical Genomics, 2015, 8, 29.	0.7	107
110	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	1.4	25
111	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. Epilepsy Research, 2015, 114, 98-105.	0.8	7
112	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.	0.7	17
113	Plasmodium vivax Populations Are More Genetically Diverse and Less Structured than Sympatric Plasmodium falciparum Populations. PLoS Neglected Tropical Diseases, 2015, 9, e0003634.	1.3	62
114	Whole exome sequencing combined with linkage analysis identifies a novel 3â€‰bp deletion in NR5A1. European Journal of Human Genetics, 2015, 23, 486-493.	1.4	27
115	GNE myopathy in Roma patients homozygous for the p.I618T founder mutation. Neuromuscular Disorders, 2015, 25, 713-718.	0.3	32
116	Investigating and Correcting Plasma DNA Sequencing Coverage Bias to Enhance Aneuploidy Discovery. PLoS ONE, 2014, 9, e86993.	1.1	24
117	Harnessing Gene Expression Networks to Prioritize Candidate Epileptic Encephalopathy Genes. PLoS ONE, 2014, 9, e102079.	1.1	25
118	Identification of a Novel RNF213 Variant in a Family with Heterogeneous Intracerebral Vasculopathy. International Journal of Stroke, 2014, 9, E26-E27.	2.9	9
119	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.	2.6	207
120	Mutations in SH3PXD2B cause Borrone dermato-cardio-skeletal syndrome. European Journal of Human Genetics, 2014, 22, 741-747.	1.4	30
121	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.	0.6	11
122	Dating Rare Mutations from Small Samples with Dense Marker Data. Genetics, 2014, 197, 1315-1327.	1.2	61
123	Using familial information for variant filtering in high-throughput sequencing studies. Human Genetics, 2014, 133, 1331-1341.	1.8	10
124	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. Nature Genetics, 2014, 46, 1239-1244.	9.4	165
125	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.	1.4	120
126	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.	2.6	60

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127	Use of Copy Number Deletion Polymorphisms to Assess DNA Chimerism. Clinical Chemistry, 2014, 60, 1105-1114.	1.5	20
128	Small intragenic deletion in FOXP2 associated with childhood apraxia of speech and dysarthria. American Journal of Medical Genetics, Part A, 2013, 161, 2321-2326.	0.7	75
129	An Î±â€Ÿâ€catenin (<scp>CTNNA1</scp>) mutation in hereditary diffuse gastric cancer. Journal of Pathology, 2013, 229, 621-629.	2.1	184
130	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	3.7	129
131	Autosomal dominant vasovagal syncope. Neurology, 2013, 80, 1485-1493.	1.5	20
132	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. Human Molecular Genetics, 2013, 22, 1417-1423.	1.4	105
133	Challenges of diagnostic exome sequencing in an inbred founder population. Molecular Genetics & Genomic Medicine, 2013, 1, 71-76.	0.6	16
134	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.	3.7	21
135	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.	1.6	58
136	Familial Adult Myoclonic Epilepsy. Archives of Neurology, 2012, 69, 474.	4.9	36
137	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. Nature Genetics, 2012, 44, 1188-1190.	9.4	333
138	Autosomal-Recessive Congenital Cerebellar Ataxia Is Caused by Mutations in Metabotropic Glutamate Receptor 1. American Journal of Human Genetics, 2012, 91, 553-564.	2.6	81
139	Strikingly Different Clinicopathological Phenotypes Determined by Progranulin-Mutation Dosage. American Journal of Human Genetics, 2012, 90, 1102-1107.	2.6	414
140	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.	3.6	62
141	An ENU-Induced Mutation of Cdh23 Causes Congenital Hearing Loss, but No Vestibular Dysfunction, in Mice. American Journal of Pathology, 2011, 179, 903-914.	1.9	26
142	Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. Genome Biology, 2011, 12, R85.	13.9	72
143	Human and Mouse Mutations in WDR35 Cause Short-Rib Polydactyly Syndromes Due to Abnormal Ciliogenesis. American Journal of Human Genetics, 2011, 88, 508-515.	2.6	122
144	A Mutation in Synaptojanin 2 Causes Progressive Hearing Loss in the ENU-Mutagenised Mouse Strain Mozart. PLoS ONE, 2011, 6, e17607.	1.1	39

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145	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	2.6	108
146	Kufs Disease, the Major Adult Form of Neuronal Ceroid Lipofuscinosis, Caused by Mutations in CLN6. American Journal of Human Genetics, 2011, 88, 566-573.	2.6	253
147	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.	2.6	166
148	Analysis of genome-wide association study data using the protein knowledge base. BMC Genetics, 2011, 12, 98.	2.7	10
149	Genomeâ€wide metaâ€analysis identifies novel multiple sclerosis susceptibility loci. Annals of Neurology, 2011, 70, 897-912.	2.8	314
150	X chromosome association testing in genome wide association studies. Genetic Epidemiology, 2011, 35, 664-670.	0.6	43
151	The Tasmanian Devil Transcriptome Reveals Schwann Cell Origins of a Clonally Transmissible Cancer. Science, 2010, 327, 84-87.	6.0	222
152	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.	2.6	211
153	A Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in TBC1D24. American Journal of Human Genetics, 2010, 87, 371-375.	2.6	111
154	Genome-wide linkage scan and association study of PARL to the expression of LHON families in Thailand. Human Genetics, 2010, 128, 39-49.	1.8	43
155	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.	1.1	12
156	A novel mutation in COCHâ€"implications for genotypeâ€phenotype correlations in DFNA9 hearing loss. Laryngoscope, 2010, 120, 2489-2493.	1.1	20
157	Evidence for a common genetic aetiology in highâ€risk families with multiple haematological malignancy subtypes. British Journal of Haematology, 2010, 150, 456-462.	1.2	7
158	Multiple Sclerosis Susceptibility-Associated SNPs Do Not Influence Disease Severity Measures in a Cohort of Australian MS Patients. PLoS ONE, 2010, 5, e10003.	1.1	45
159	A Polymorphism in the HLA-DPB1 Gene Is Associated with Susceptibility to Multiple Sclerosis. PLoS ONE, 2010, 5, e13454.	1.1	55
160	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.	2.4	14
161	Saliva-Derived DNA Performs Well in Large-Scale, High-Density Single-Nucleotide Polymorphism Microarray Studies. Cancer Epidemiology Biomarkers and Prevention, 2010, 19, 794-798.	1.1	52
162	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.	3.3	37

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163	Generating linkage mapping files from Affymetrix SNP chip data. Bioinformatics, 2009, 25, 1961-1962.	1.8	60
164	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.	1.7	164
165	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.	0.7	18
166	Mutations in the first MyTH4 domain of MYO15A are a common cause of DFNB3 hearing loss. Laryngoscope, 2009, 119, 727-733.	1.1	48
167	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.	1.2	61
168	Genome-wide association study identifies new multiple sclerosis susceptibility loci on chromosomes 12 and 20. Nature Genetics, 2009, 41, 824-828.	9.4	501
169	IL28B is associated with response to chronic hepatitis C interferon-Î± and ribavirin therapy. Nature Genetics, 2009, 41, 1100-1104.	9.4	1,808
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