

Melanie Bahlo

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/63197/publications.pdf>

Version: 2024-02-01

207
papers

14,948
citations

20759

60
h-index

23472

111
g-index

264
all docs

264
docs citations

264
times ranked

23882
citing authors

#	ARTICLE	IF	CITATIONS
1	Atypical development of Broca's area in a large family with inherited stuttering. <i>Brain</i> , 2022, 145, 1177-1188.	3.7	6
2	A family study implicates <i>GBE1</i> in the etiology of autism spectrum disorder. <i>Human Mutation</i> , 2022, 43, 16-29.	1.1	2
3	Infantile-onset myoclonic developmental and epileptic encephalopathy: A new <i>RARS2</i> phenotype. <i>Epilepsia Open</i> , 2022, 7, 170-180.	1.3	5
4	Genetics of reticular pseudodrusen in age-related macular degeneration. <i>Trends in Genetics</i> , 2022, 38, 312-316.	2.9	1
5	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy. <i>Neurology: Genetics</i> , 2022, 8, e652.	0.9	14
6	Population-level genome-wide STR discovery and validation for population structure and genetic diversity assessment of <i>Plasmodium</i> species. <i>PLoS Genetics</i> , 2022, 18, e1009604.	1.5	8
7	Global diversity and balancing selection of 23 leading <i>Plasmodium falciparum</i> candidate vaccine antigens. <i>PLoS Computational Biology</i> , 2022, 18, e1009801.	1.5	14
8	PacBio long-read amplicon sequencing enables scalable high-resolution population allele typing of the complex <i>CYP2D6</i> locus. <i>Communications Biology</i> , 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. <i>Developmental Cell</i> , 2022, 57, 820-836.e6.	3.1	37
10	Germline variants in tumor suppressor <i>FBXW7</i> lead to impaired ubiquitination and a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 601-617.	2.6	16
11	Heterozygous <i>PNPT1</i> Variants Cause Spinocerebellar Ataxia Type 25. <i>Annals of Neurology</i> , 2022, 92, 122-137.	2.8	8
12	Detecting Tandem Repeat Expansions Using Short-Read Sequencing for Clinical Use. <i>Neuromethods</i> , 2022, , 15-42.	0.2	2
13	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. <i>EBioMedicine</i> , 2022, 81, 104079.	2.7	10
14	Spatial centrosome proteome of human neural cells uncovers disease-relevant heterogeneity. <i>Science</i> , 2022, 376, .	6.0	25
15	Founder effect of the TTTCA repeat insertions in <i>SAMD12</i> causing BAFME1. <i>European Journal of Human Genetics</i> , 2021, 29, 343-348.	1.4	14
16	Progressive Myoclonus Epilepsy Caused by a Homozygous Splicing Variant of <i>SLC7A6OS</i> . <i>Annals of Neurology</i> , 2021, 89, 402-407.	2.8	5
17	Transcriptome analysis of a ring chromosome 20 patient cohort. <i>Epilepsia</i> , 2021, 62, e22-e28.	2.6	5
18	Infanticide vs. inherited cardiac arrhythmias. <i>Europace</i> , 2021, 23, 441-450.	0.7	21

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19	A cross-platform approach identifies genetic regulators of human metabolism and health. <i>Nature Genetics</i> , 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. <i>Journal of the Neurological Sciences</i> , 2021, 420, 117260.	0.3	16
21	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. <i>Brain Communications</i> , 2021, 3, fcaa235.	1.5	42
22	Comparative genomics revealed adaptive admixture in <i>Cryptosporidium hominis</i> in Africa. <i>Microbial Genomics</i> , 2021, 7, .	1.0	13
23	Contribution of rare genetic variants to drug response in absence epilepsy. <i>Epilepsy Research</i> , 2021, 170, 106537.	0.8	9
24	Genetic disruption of serine biosynthesis is a key driver of macular telangiectasia type 2 aetiology and progression. <i>Genome Medicine</i> , 2021, 13, 39.	3.6	15
25	Association of <i>SLC32A1</i> Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. <i>Neurology</i> , 2021, 96, e2251-e2260.	1.5	13
26	Identification of genetic factors influencing metabolic dysregulation and retinal support for MacTel, a retinal disorder. <i>Communications Biology</i> , 2021, 4, 274.	2.0	26
27	Progressive myoclonus epilepsies—Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. <i>American Journal of Human Genetics</i> , 2021, 108, 722-738.	2.6	41
28	Expanding the clinical and radiological phenotypes of leukoencephalopathy due to biallelic <i>HMBS</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2941-2950.	0.7	2
29	Clonal multi-omics reveals <i>Bcor</i> as a negative regulator of emergency dendritic cell development. <i>Immunity</i> , 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. <i>Expert Review of Neurotherapeutics</i> , 2021, 21, 1309-1316.	1.4	5
31	A survey of RNA editing at single-cell resolution links interneurons to schizophrenia and autism. <i>Rna</i> , 2021, 27, 1482-1496.	1.6	7
32	Progressive Myoclonus Epilepsies. <i>Neurology: Genetics</i> , 2021, 7, e641.	0.9	20
33	Deletions in <i>VANGL1</i> are a risk factor for antibody-mediated kidney disease. <i>Cell Reports Medicine</i> , 2021, 2, 100475.	3.3	2
34	Germline and Mosaic Variants in <i>PRKACA</i> and <i>PRKACB</i> Cause a Multiple Congenital Malformation Syndrome. <i>American Journal of Human Genetics</i> , 2020, 107, 977-988.	2.6	33
35	Systemic lipid dysregulation is a risk factor for macular neurodegenerative disease. <i>Scientific Reports</i> , 2020, 10, 12165.	1.6	24
36	SNP barcodes provide higher resolution than microsatellite markers to measure <i>Plasmodium vivax</i> population genetics. <i>Malaria Journal</i> , 2020, 19, 375.	0.8	25

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37	Clinical spectrum of the pentanucleotide repeat expansion in the <i>RFC1</i> gene in ataxia syndromes. <i>Neurology</i> , 2020, 95, e2912-e2923.	1.5	32
38	Tracing Autism Traits in Large Multiplex Families to Identify Endophenotypes of the Broader Autism Phenotype. <i>International Journal of Molecular Sciences</i> , 2020, 21, 7965.	1.8	3
39	Rapid Diagnosis of Spinocerebellar Ataxia 36 in a Three-Generation Family Using Short-Read Whole-Genome Sequencing Data. <i>Movement Disorders</i> , 2020, 35, 1675-1679.	2.2	12
40	A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. <i>Nature Communications</i> , 2020, 11, 3150.	5.8	75
41	Familial adult myoclonic epilepsy type 1 SAMD12 TTCA repeat expansion arose 17,000 years ago and is present in Sri Lankan and Indian families. <i>European Journal of Human Genetics</i> , 2020, 28, 973-978.	1.4	23
42	CYLD is a causative gene for frontotemporal dementia “ amyotrophic lateral sclerosis. <i>Brain</i> , 2020, 143, 783-799.	3.7	62
43	Inherited <i>RORB</i> pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. <i>Epilepsia</i> , 2020, 61, e23-e29.	2.6	14
44	Callosal agenesis and congenital mirror movements: outcomes associated with <i>DCC</i> mutations. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 758-762.	1.1	11
45	Severe childhood speech disorder. <i>Neurology</i> , 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. <i>Genome Biology</i> , 2020, 21, 102.	3.8	114
47	The Genetics of Epilepsy. <i>Annual Review of Genomics and Human Genetics</i> , 2020, 21, 205-230.	2.5	116
48	Multiple sclerosis risk variants regulate gene expression in innate and adaptive immune cells. <i>Life Science Alliance</i> , 2020, 3, e202000650.	1.3	22
49	Generation of seven iPSC lines from peripheral blood mononuclear cells suitable to investigate Autism Spectrum Disorder. <i>Stem Cell Research</i> , 2019, 39, 101516.	0.3	4
50	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , 2019, 105, 267-282.	2.6	237
51	Intronic ATTTC repeat expansions in <i>STARD7</i> in familial adult myoclonic epilepsy linked to chromosome 2. <i>Nature Communications</i> , 2019, 10, 4920.	5.8	99
52	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. <i>Epilepsia Open</i> , 2019, 4, 504-510.	1.3	11
53	Serine and Lipid Metabolism in Macular Disease and Peripheral Neuropathy. <i>New England Journal of Medicine</i> , 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. <i>Neuron</i> , 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. <i>American Journal of Human Genetics</i> , 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. <i>Genetics in Medicine</i> , 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. <i>Parkinsonism and Related Disorders</i> , 2019, 64, 308-311.	1.1	7
58	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 914-924.	2.6	23
59	Fatal Enteroviral Encephalitis in a Patient with Common Variable Immunodeficiency Harboring a Novel Mutation in NFKB2. <i>Journal of Clinical Immunology</i> , 2019, 39, 324-335.	2.0	14
60	Dorsal language stream anomalies in an inherited speech disorder. <i>Brain</i> , 2019, 142, 966-977.	3.7	16
61	Genetic Analysis of Patients Who Experienced Awareness with Recall while under General Anesthesia. <i>Anesthesiology</i> , 2019, 131, 974-982.	1.3	9
62	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. <i>Nature Communications</i> , 2019, 10, 4919.	5.8	111
63	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. <i>Human Mutation</i> , 2019, 40, 374-379.	1.1	7
64	dtangle: accurate and robust cell type deconvolution. <i>Bioinformatics</i> , 2019, 35, 2093-2099.	1.8	98
65	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. <i>Genetics in Medicine</i> , 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. <i>Molecular Psychiatry</i> , 2019, 24, 1065-1078.	4.1	106
67	Mosaic uniparental disomy results in GM1 gangliosidosis with normal enzyme assay. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 230-234.	0.7	3
68	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. <i>F1000Research</i> , 2018, 7, 1297.	0.8	131
69	Genetic investigation into an increased susceptibility to biliary atresia in an extended New Zealand Māori family. <i>BMC Medical Genomics</i> , 2018, 11, 121.	0.7	7
70	Detecting Expansions of Tandem Repeats in Cohorts Sequenced with Short-Read Sequencing Data. <i>American Journal of Human Genetics</i> , 2018, 103, 858-873.	2.6	93
71	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. <i>F1000Research</i> , 2018, 7, 1297.	0.8	157
72	Identity-by-descent analyses for measuring population dynamics and selection in recombining pathogens. <i>PLoS Genetics</i> , 2018, 14, e1007279.	1.5	86

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73	Recessive Spondylcarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in MYH3. <i>American Journal of Human Genetics</i> , 2018, 102, 1115-1125.	2.6	18
74	Evidence of linkage to chromosome 5p13.2â€“11.1 in a large inbred family with genetic generalized epilepsy. <i>Epilepsia</i> , 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. <i>Human Molecular Genetics</i> , 2018, 27, 2775-2788.	1.4	25
76	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. <i>PLoS Genetics</i> , 2018, 14, e1007281.	1.5	40
77	Recent advances in the detection of repeat expansions with short-read next-generation sequencing. <i>F1000Research</i> , 2018, 7, 736.	0.8	84
78	Increasingly inbred and fragmented populations of <i>Plasmodium vivax</i> associated with the eastward decline in malaria transmission across the Southwest Pacific. <i>PLoS Neglected Tropical Diseases</i> , 2018, 12, e0006146.	1.3	27
79	<i>EIF2S3</i> Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. <i>Human Mutation</i> , 2017, 38, 409-425.	1.1	57
80	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. <i>Nature Genetics</i> , 2017, 49, 511-514.	9.4	69
81	Genome-wide analyses identify common variants associated with macular telangiectasia type 2. <i>Nature Genetics</i> , 2017, 49, 559-567.	9.4	105
82	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. <i>Epilepsy Research</i> , 2017, 131, 9-14.	0.8	12
83	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. <i>European Journal of Medical Genetics</i> , 2017, 60, 437-443.	0.7	10
84	A <i>SLC39A8</i> variant causes manganese deficiency, and glycosylation and mitochondrial disorders. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 261-269.	1.7	101
85	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. <i>American Journal of Human Genetics</i> , 2017, 101, 516-524.	2.6	43
86	Neuropathology of childhood-onset basal ganglia degeneration caused by mutation of <i>VAC14</i> . <i>Annals of Clinical and Translational Neurology</i> , 2017, 4, 859-864.	1.7	17
87	brain-coX: investigating and visualising gene co-expression in seven human brain transcriptomic datasets. <i>Genome Medicine</i> , 2017, 9, 55.	3.6	13
88	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. <i>Human Genetics</i> , 2016, 135, 1117-1125.	1.8	29
89	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 217-227.	2.6	57
90	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. <i>Neurology: Genetics</i> , 2016, 2, e51.	0.9	19

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91	Evaluation of non-coding variation in <i>GLUT1</i> deficiency. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 1295-1302.	1.1	20
92	Heterozygous mutations in <i>HSD17B4</i> cause juvenile peroxisomal D-bifunctional protein deficiency. <i>Neurology: Genetics</i> , 2016, 2, e114.	0.9	18
93	Early neuroimaging markers of FOXP2 intragenic deletion. <i>Scientific Reports</i> , 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. <i>European Heart Journal</i> , 2016, 37, 2586-2590.	1.0	49
95	Dominant <i>KCNA2</i> mutation causes episodic ataxia and pharmaco-responsive epilepsy. <i>Neurology</i> , 2016, 87, 1975-1984.	1.5	71
96	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator <i>NPRL3</i> . <i>Annals of Neurology</i> , 2016, 79, 132-137.	2.8	116
97	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). <i>Neurology</i> , 2016, 87, 579-584.	1.5	28
98	Multiplex families with epilepsy. <i>Neurology</i> , 2016, 86, 713-722.	1.5	23
99	XIBD: software for inferring pairwise identity by descent on the X chromosome. <i>Bioinformatics</i> , 2016, 32, 2389-2391.	1.8	21
100	Structurally conserved erythrocyte-binding domain in <i>Plasmodium</i> provides a versatile scaffold for alternate receptor engagement. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 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. <i>PLoS Neglected Tropical Diseases</i> , 2016, 10, e0004384.	1.3	30
102	<i>PRIMA1</i> mutation: a new cause of nocturnal frontal lobe epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 821-830.	1.7	21
103	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to <i>AMPD2</i> loss. <i>Neurology: Genetics</i> , 2015, 1, e16.	0.9	29
104	Systematic noise degrades gene co-expression signals but can be corrected. <i>BMC Bioinformatics</i> , 2015, 16, 309.	1.2	50
105	Cpipe: a shared variant detection pipeline designed for diagnostic settings. <i>Genome Medicine</i> , 2015, 7, 68.	3.6	78
106	Mutation of the nuclear lamin gene <i>LMNB2</i> in progressive myoclonus epilepsy with early ataxia. <i>Human Molecular Genetics</i> , 2015, 24, 4483-4490.	1.4	41
107	Hemispheric cortical dysplasia secondary to a mosaic somatic mutation in <i>MTOR</i> . <i>Neurology</i> , 2015, 84, 2029-2032.	1.5	64
108	Familial cortical dysplasia type IIa caused by a germline mutation in <i>DEPDC5</i> . <i>Annals of Clinical and Translational Neurology</i> , 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. <i>BMC Medical Genomics</i> , 2015, 8, 29.	0.7	107
110	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. <i>Human Molecular Genetics</i> , 2015, 24, 2000-2010.	1.4	25
111	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. <i>Epilepsy Research</i> , 2015, 114, 98-105.	0.8	7
112	Identical by descent L1CAM mutation in two apparently unrelated families with intellectual disability without L1 syndrome. <i>European Journal of Medical Genetics</i> , 2015, 58, 364-368.	0.7	17
113	Plasmodium vivax Populations Are More Genetically Diverse and Less Structured than Sympatric Plasmodium falciparum Populations. <i>PLoS Neglected Tropical Diseases</i> , 2015, 9, e0003634.	1.3	62
114	Whole exome sequencing combined with linkage analysis identifies a novel 3â€%bp deletion in NR5A1. <i>European Journal of Human Genetics</i> , 2015, 23, 486-493.	1.4	27
115	GNE myopathy in Roma patients homozygous for the p.I618T founder mutation. <i>Neuromuscular Disorders</i> , 2015, 25, 713-718.	0.3	32
116	Investigating and Correcting Plasma DNA Sequencing Coverage Bias to Enhance Aneuploidy Discovery. <i>PLoS ONE</i> , 2014, 9, e86993.	1.1	24
117	Harnessing Gene Expression Networks to Prioritize Candidate Epileptic Encephalopathy Genes. <i>PLoS ONE</i> , 2014, 9, e102079.	1.1	25
118	Identification of a Novel RNF213 Variant in a Family with Heterogeneous Intracerebral Vasculopathy. <i>International Journal of Stroke</i> , 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. <i>American Journal of Human Genetics</i> , 2014, 95, 729-735.	2.6	207
120	Mutations in SH3PXD2B cause Borrone dermato-cardio-skeletal syndrome. <i>European Journal of Human Genetics</i> , 2014, 22, 741-747.	1.4	30
121	Candidate disease gene prediction using <i>Gentrepid</i> : application to a genome-wide association study on coronary artery disease. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 44-57.	0.6	11
122	Dating Rare Mutations from Small Samples with Dense Marker Data. <i>Genetics</i> , 2014, 197, 1315-1327.	1.2	61
123	Using familial information for variant filtering in high-throughput sequencing studies. <i>Human Genetics</i> , 2014, 133, 1331-1341.	1.8	10
124	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. <i>Nature Genetics</i> , 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. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	1.4	120
126	A Founder Mutation in PET100 Causes Isolated Complex IV Deficiency in Lebanese Individuals with Leigh Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 209-222.	2.6	60

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127	Use of Copy Number Deletion Polymorphisms to Assess DNA Chimerism. <i>Clinical Chemistry</i> , 2014, 60, 1105-1114.	1.5	20
128	Small intragenic deletion in <i>FOXP2</i> associated with childhood apraxia of speech and dysarthria. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2321-2326.	0.7	75
129	An <i>adenomatous polyposis coli</i> (<i>APC</i>) mutation in hereditary diffuse gastric cancer. <i>Journal of Pathology</i> , 2013, 229, 621-629.	2.1	184
130	"North Sea" progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. <i>Brain</i> , 2013, 136, 1146-1154.	3.7	129
131	Autosomal dominant vasovagal syncope. <i>Neurology</i> , 2013, 80, 1485-1493.	1.5	20
132	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. <i>Human Molecular Genetics</i> , 2013, 22, 1417-1423.	1.4	105
133	Challenges of diagnostic exome sequencing in an inbred founder population. <i>Molecular Genetics & Genomic Medicine</i> , 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. <i>Brain</i> , 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 (<i>Ndufs4</i>) Gene. <i>Journal of Biological Chemistry</i> , 2012, 287, 20652-20663.	1.6	58
136	Familial Adult Myoclonic Epilepsy. <i>Archives of Neurology</i> , 2012, 69, 474.	4.9	36
137	Missense mutations in the sodium-gated potassium channel gene <i>KCNT1</i> cause severe autosomal dominant nocturnal frontal lobe epilepsy. <i>Nature Genetics</i> , 2012, 44, 1188-1190.	9.4	333
138	Autosomal-Recessive Congenital Cerebellar Ataxia Is Caused by Mutations in Metabotropic Glutamate Receptor 1. <i>American Journal of Human Genetics</i> , 2012, 91, 553-564.	2.6	81
139	Strikingly Different Clinicopathological Phenotypes Determined by Progranulin-Mutation Dosage. <i>American Journal of Human Genetics</i> , 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. <i>Genome Medicine</i> , 2011, 3, 57.	3.6	62
141	An ENU-Induced Mutation of <i>Cdh23</i> Causes Congenital Hearing Loss, but No Vestibular Dysfunction, in Mice. <i>American Journal of Pathology</i> , 2011, 179, 903-914.	1.9	26
142	Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. <i>Genome Biology</i> , 2011, 12, R85.	13.9	72
143	Human and Mouse Mutations in <i>WDR35</i> Cause Short-Rib Polydactyly Syndromes Due to Abnormal Ciliogenesis. <i>American Journal of Human Genetics</i> , 2011, 88, 508-515.	2.6	122
144	A Mutation in Synaptotagmin 2 Causes Progressive Hearing Loss in the ENU-Mutagenised Mouse Strain Mozart. <i>PLoS ONE</i> , 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	Genome-wide meta-analysis identifies novel multiple sclerosis susceptibility loci. Annals of Neurology, 2011, 70, 897-912.	2.8	314
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