

# Melanie Bahlo

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

210  
papers

10,904  
citations

52  
h-index

100  
g-index

264  
ext. papers

13,143  
ext. citations

8.4  
avg. IF

5.88  
L-index

#	Paper	IF	Citations
210	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy.. <i>Neurology: Genetics</i> , <b>2022</b> , 8, e652	3.8	1
209	Population-level genome-wide STR discovery and validation for population structure and genetic diversity assessment of Plasmodium species.. <i>PLoS Genetics</i> , <b>2022</b> , 18, e1009604	6	1
208	Global diversity and balancing selection of 23 leading Plasmodium falciparum candidate vaccine antigens.. <i>PLoS Computational Biology</i> , <b>2022</b> , 18, e1009801	5	1
207	PacBio long-read amplicon sequencing enables scalable high-resolution population allele typing of the complex CYP2D6 locus.. <i>Communications Biology</i> , <b>2022</b> , 5, 168	6.7	0
206	Cell-specific cis-regulatory elements and mechanisms of non-coding genetic disease in human retina and retinal organoids.. <i>Developmental Cell</i> , <b>2022</b> ,	10.2	2
205	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome.. <i>American Journal of Human Genetics</i> , <b>2022</b> , 109, 601-617	11	0
204	Detecting Tandem Repeat Expansions Using Short-Read Sequencing for Clinical Use. <i>Neuromethods</i> , <b>2022</b> , 15-42	0.4	0
203	Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery. <i>EBioMedicine</i> , <b>2022</b> , 81, 104079	8.8	1
202	Progressive Myoclonus Epilepsies: Diagnostic Yield With Next-Generation Sequencing in Previously Unsolved Cases. <i>Neurology: Genetics</i> , <b>2021</b> , 7, e641	3.8	2
201	Infantile-onset myoclonic developmental and epileptic encephalopathy: A new RARS2 phenotype. <i>Epilepsia Open</i> , <b>2021</b> ,	4	1
200	Identification of genetic factors influencing metabolic dysregulation and retinal support for MacTel, a retinal disorder. <i>Communications Biology</i> , <b>2021</b> , 4, 274	6.7	5
199	Progressive myoclonus epilepsies-Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. <i>American Journal of Human Genetics</i> , <b>2021</b> , 108, 722-738	11	10
198	Expanding the clinical and radiological phenotypes of leukoencephalopathy due to biallelic HMBS mutations. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 2941-2950	2.5	
197	Clonal multi-omics reveals Bcor as a negative regulator of emergency dendritic cell development. <i>Immunity</i> , <b>2021</b> , 54, 1338-1351.e9	32.3	5
196	Founder effect of the TTTCA repeat insertions in SAMD12 causing BAFME1. <i>European Journal of Human Genetics</i> , <b>2021</b> , 29, 343-348	5.3	3
195	Progressive Myoclonus Epilepsy Caused by a Homozygous Splicing Variant of SLC7A6OS. <i>Annals of Neurology</i> , <b>2021</b> , 89, 402-407	9.4	1
194	Transcriptome analysis of a ring chromosome 20 patient cohort. <i>Epilepsia</i> , <b>2021</b> , 62, e22-e28	6.4	1

193	Infanticide vs. inherited cardiac arrhythmias. <i>Europace</i> , <b>2021</b> , 23, 441-450	3.9	2
192	A cross-platform approach identifies genetic regulators of human metabolism and health. <i>Nature Genetics</i> , <b>2021</b> , 53, 54-64	36.3	18
191	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> , <b>2021</b> , 420, 117260	3.2	4
190	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. <i>Brain Communications</i> , <b>2021</b> , 3, fcaa235	4.5	17
189	Comparative genomics revealed adaptive admixture in in Africa. <i>Microbial Genomics</i> , <b>2021</b> , 7,	4.4	4
188	Contribution of rare genetic variants to drug response in absence epilepsy. <i>Epilepsy Research</i> , <b>2021</b> , 170, 106537	3	5
187	Genetic disruption of serine biosynthesis is a key driver of macular telangiectasia type 2 aetiology and progression. <i>Genome Medicine</i> , <b>2021</b> , 13, 39	14.4	3
186	Association of Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. <i>Neurology</i> , <b>2021</b> , 96, e2251-e2260	6.5	3
185	Cutting edge approaches to detecting brain mosaicism associated with common focal epilepsies: implications for diagnosis and potential therapies. <i>Expert Review of Neurotherapeutics</i> , <b>2021</b> , 21, 1309-1316	4.3	0
184	A survey of RNA editing at single-cell resolution links interneurons to schizophrenia and autism. <i>Rna</i> , <b>2021</b> , 27, 1482-1496	5.8	2
183	Deletions in are a risk factor for antibody-mediated kidney disease.. <i>Cell Reports Medicine</i> , <b>2021</b> , 2, 100475	4.5	1
182	Tracing Autism Traits in Large Multiplex Families to Identify Endophenotypes of the Broader Autism Phenotype. <i>International Journal of Molecular Sciences</i> , <b>2020</b> , 21,	6.3	1
181	Rapid Diagnosis of Spinocerebellar Ataxia 36 in a Three-Generation Family Using Short-Read Whole-Genome Sequencing Data. <i>Movement Disorders</i> , <b>2020</b> , 35, 1675-1679	7	5
180	A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. <i>Nature Communications</i> , <b>2020</b> , 11, 3150	17.4	41
179	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> , <b>2020</b> , 28, 973-978	5.3	9
178	CYLD is a causative gene for frontotemporal dementia - amyotrophic lateral sclerosis. <i>Brain</i> , <b>2020</b> , 143, 783-799	11.2	33
177	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. <i>Epilepsia</i> , <b>2020</b> , 61, e23-e29	6.4	5
176	Callosal agenesis and congenital mirror movements: outcomes associated with DCC mutations. <i>Developmental Medicine and Child Neurology</i> , <b>2020</b> , 62, 758-762	3.3	5

175	Severe childhood speech disorder: Gene discovery highlights transcriptional dysregulation. <i>Neurology</i> , <b>2020</b> , 94, e2148-e2167	6.5	28
174	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. <i>Genome Biology</i> , <b>2020</b> , 21, 102	18.3	29
173	The Genetics of Epilepsy. <i>Annual Review of Genomics and Human Genetics</i> , <b>2020</b> , 21, 205-230	9.7	30
172	Multiple sclerosis risk variants regulate gene expression in innate and adaptive immune cells. <i>Life Science Alliance</i> , <b>2020</b> , 3,	5.8	10
171	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. <i>American Journal of Human Genetics</i> , <b>2020</b> , 107, 977-988	11	9
170	Systemic lipid dysregulation is a risk factor for macular neurodegenerative disease. <i>Scientific Reports</i> , <b>2020</b> , 10, 12165	4.9	7
169	SNP barcodes provide higher resolution than microsatellite markers to measure Plasmodium vivax population genetics. <i>Malaria Journal</i> , <b>2020</b> , 19, 375	3.6	8
168	Clinical spectrum of the pentanucleotide repeat expansion in the gene in ataxia syndromes. <i>Neurology</i> , <b>2020</b> , 95, e2912-e2923	6.5	12
167	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. <i>Epilepsia Open</i> , <b>2019</b> , 4, 504-510	4	4
166	Serine and Lipid Metabolism in Macular Disease and Peripheral Neuropathy. <i>New England Journal of Medicine</i> , <b>2019</b> , 381, 1422-1433	59.2	91
165	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. <i>Neuron</i> , <b>2019</b> , 104, 665-679.e8	13.9	16
164	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 151-165	11	80
163	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 2532-2542	8.1	8
162	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> , <b>2019</b> , 64, 308-311	3.6	4
161	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 914-924	11	11
160	Fatal Enteroviral Encephalitis in a Patient with Common Variable Immunodeficiency Harboring a Novel Mutation in NFKB2. <i>Journal of Clinical Immunology</i> , <b>2019</b> , 39, 324-335	5.7	7
159	Dorsal language stream anomalies in an inherited speech disorder. <i>Brain</i> , <b>2019</b> , 142, 966-977	11.2	9
158	Generation of seven iPSC lines from peripheral blood mononuclear cells suitable to investigate Autism Spectrum Disorder. <i>Stem Cell Research</i> , <b>2019</b> , 39, 101516	1.6	4

157	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 267-282	11	104
156	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. <i>Nature Communications</i> , <b>2019</b> , 10, 4920	17.4	48
155	Genetic Analysis of Patients Who Experienced Awareness with Recall while under General Anesthesia. <i>Anesthesiology</i> , <b>2019</b> , 131, 974-982	4.3	4
154	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. <i>Nature Communications</i> , <b>2019</b> , 10, 4919	17.4	58
153	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. <i>Human Mutation</i> , <b>2019</b> , 40, 374-379	4.7	6
152	dtangle: accurate and robust cell type deconvolution. <i>Bioinformatics</i> , <b>2019</b> , 35, 2093-2099	7.2	30
151	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 948-954	8.1	26
150	A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development. <i>Molecular Psychiatry</i> , <b>2019</b> , 24, 1065-1078	15.1	62
149	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> , <b>2018</b> , 27, 2775-2788	5.6	16
148	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. <i>PLoS Genetics</i> , <b>2018</b> , 14, e1007281	6	27
147	Recent advances in the detection of repeat expansions with short-read next-generation sequencing. <i>F1000Research</i> , <b>2018</b> , 7,	3.6	42
146	Increasingly inbred and fragmented populations of Plasmodium vivax associated with the eastward decline in malaria transmission across the Southwest Pacific. <i>PLoS Neglected Tropical Diseases</i> , <b>2018</b> , 12, e0006146	4.8	16
145	Mosaic uniparental disomy results in GM1 gangliosidosis with normal enzyme assay. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 230-234	2.5	2
144	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. <i>F1000Research</i> , <b>2018</b> , 7, 1297	3.6	65
143	Genetic investigation into an increased susceptibility to biliary atresia in an extended New Zealand Māori family. <i>BMC Medical Genomics</i> , <b>2018</b> , 11, 121	3.7	3
142	Detecting Expansions of Tandem Repeats in Cohorts Sequenced with Short-Read Sequencing Data. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 858-873	11	51
141	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. <i>F1000Research</i> , <b>2018</b> , 7, 1297	3.6	95
140	Identity-by-descent analyses for measuring population dynamics and selection in recombining pathogens. <i>PLoS Genetics</i> , <b>2018</b> , 14, e1007279	6	42

139	Recessive Spondylacarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in MYH3. <i>American Journal of Human Genetics</i> , <b>2018</b> , 102, 1115-1125	11	10
138	Evidence of linkage to chromosome 5p13.2-q11.1 in a large inbred family with genetic generalized epilepsy. <i>Epilepsia</i> , <b>2018</b> , 59, e125-e129	6.4	2
137	EIF2S3 Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. <i>Human Mutation</i> , <b>2017</b> , 38, 409-425	4.7	41
136	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. <i>Nature Genetics</i> , <b>2017</b> , 49, 511-514	36.3	54
135	Genome-wide analyses identify common variants associated with macular telangiectasia type 2. <i>Nature Genetics</i> , <b>2017</b> , 49, 559-567	36.3	75
134	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. <i>Epilepsy Research</i> , <b>2017</b> , 131, 9-14	3	4
133	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. <i>European Journal of Medical Genetics</i> , <b>2017</b> , 60, 437-443	2.6	9
132	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. <i>Journal of Inherited Metabolic Disease</i> , <b>2017</b> , 40, 261-269	5.4	70
131	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 516-524	11	29
130	Neuropathology of childhood-onset basal ganglia degeneration caused by mutation of. <i>Annals of Clinical and Translational Neurology</i> , <b>2017</b> , 4, 859-864	5.3	14
129	brain-coX: investigating and visualising gene co-expression in seven human brain transcriptomic datasets. <i>Genome Medicine</i> , <b>2017</b> , 9, 55	14.4	5
128	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). <i>Neurology</i> , <b>2016</b> , 87, 579-84	6.5	19
127	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. <i>Neurology</i> , <b>2016</b> , 86, 713-22	6.5	22
126	XIBD: software for inferring pairwise identity by descent on the X chromosome. <i>Bioinformatics</i> , <b>2016</b> , 32, 2389-91	7.2	10
125	Structurally conserved erythrocyte-binding domain in Plasmodium provides a versatile scaffold for alternate receptor engagement. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2016</b> , 113, E191-200	11.5	34
124	Mitochondrial Genome Sequence of the Scabies Mite Provides Insight into the Genetic Diversity of Individual Scabies Infections. <i>PLoS Neglected Tropical Diseases</i> , <b>2016</b> , 10, e0004384	4.8	23
123	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2-2q11.2. <i>Human Genetics</i> , <b>2016</b> , 135, 1117-25	6.3	16
122	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 217-27	11	45

121	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. <i>Neurology: Genetics</i> , <b>2016</b> , 2, e51	3.8	15
120	Evaluation of non-coding variation in GLUT1 deficiency. <i>Developmental Medicine and Child Neurology</i> , <b>2016</b> , 58, 1295-1302	3.3	19
119	Heterozygous mutations in cause juvenile peroxisomal D-bifunctional protein deficiency. <i>Neurology: Genetics</i> , <b>2016</b> , 2, e114	3.8	14
118	Early neuroimaging markers of FOXP2 intragenic deletion. <i>Scientific Reports</i> , <b>2016</b> , 6, 35192	4.9	15
117	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> , <b>2016</b> , 37, 2586-90	9.5	28
116	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. <i>Neurology</i> , <b>2016</b> , 87, 1975-1984	6.5	50
115	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator NPRL3. <i>Annals of Neurology</i> , <b>2016</b> , 79, 132-7	9.4	90
114	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 2000-10	5.6	14
113	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. <i>Epilepsy Research</i> , <b>2015</b> , 114, 98-105	3	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> , <b>2015</b> , 58, 364-8	2.6	11
111	Plasmodium vivax populations are more genetically diverse and less structured than sympatric Plasmodium falciparum populations. <i>PLoS Neglected Tropical Diseases</i> , <b>2015</b> , 9, e0003634	4.8	53
110	Whole exome sequencing combined with linkage analysis identifies a novel 3 bp deletion in NR5A1. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 486-93	5.3	23
109	GNE myopathy in Roma patients homozygous for the p.I618T founder mutation. <i>Neuromuscular Disorders</i> , <b>2015</b> , 25, 713-8	2.9	21
108	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. <i>Annals of Clinical and Translational Neurology</i> , <b>2015</b> , 2, 821-30	5.3	19
107	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. <i>Neurology: Genetics</i> , <b>2015</b> , 1, e16	3.8	25
106	Systematic noise degrades gene co-expression signals but can be corrected. <i>BMC Bioinformatics</i> , <b>2015</b> , 16, 309	3.6	35
105	Cpipe: a shared variant detection pipeline designed for diagnostic settings. <i>Genome Medicine</i> , <b>2015</b> , 7, 68	14.4	62
104	Mutation of the nuclear lamin gene LMNB2 in progressive myoclonus epilepsy with early ataxia. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 4483-90	5.6	33

103	Hemispheric cortical dysplasia secondary to a mosaic somatic mutation in MTOR. <i>Neurology</i> , <b>2015</b> , 84, 2029-32	6.5	54
102	Familial cortical dysplasia type IIA caused by a germline mutation in DEPDC5. <i>Annals of Clinical and Translational Neurology</i> , <b>2015</b> , 2, 575-80	5.3	73
101	High-resolution characterization of sequence signatures due to non-random cleavage of cell-free DNA. <i>BMC Medical Genomics</i> , <b>2015</b> , 8, 29	3.7	73
100	Using familial information for variant filtering in high-throughput sequencing studies. <i>Human Genetics</i> , <b>2014</b> , 133, 1331-41	6.3	8
99	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. <i>Nature Genetics</i> , <b>2014</b> , 46, 1239-44	36.3	130
98	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> , <b>2014</b> , 23, 2888-900	5.6	99
97	A founder mutation in PET100 causes isolated complex IV deficiency in Lebanese individuals with Leigh syndrome. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 209-22	11	49
96	Use of copy number deletion polymorphisms to assess DNA chimerism. <i>Clinical Chemistry</i> , <b>2014</b> , 60, 1105-14	5.4	17
95	Investigating and correcting plasma DNA sequencing coverage bias to enhance aneuploidy discovery. <i>PLoS ONE</i> , <b>2014</b> , 9, e86993	3.7	15
94	Harnessing gene expression networks to prioritize candidate epileptic encephalopathy genes. <i>PLoS ONE</i> , <b>2014</b> , 9, e102079	3.7	22
93	Identification of a novel RNF213 variant in a family with heterogeneous intracerebral vasculopathy. <i>International Journal of Stroke</i> , <b>2014</b> , 9, E26-7	6.3	9
92	Mutations in RAB39B cause X-linked intellectual disability and early-onset Parkinson disease with $\alpha$ -synuclein pathology. <i>American Journal of Human Genetics</i> , <b>2014</b> , 95, 729-35	11	153
91	Mutations in SH3PXD2B cause Borrone dermato-cardio-skeletal syndrome. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22, 741-7	5.3	26
90	Candidate disease gene prediction using Gentrepid: application to a genome-wide association study on coronary artery disease. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2014</b> , 2, 44-57	2.3	10
89	Dating rare mutations from small samples with dense marker data. <i>Genetics</i> , <b>2014</b> , 197, 1315-27	4	35
88	Small intragenic deletion in FOXP2 associated with childhood apraxia of speech and dysarthria. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 2321-6	2.5	54
87	An E-catenin (CTNNA1) mutation in hereditary diffuse gastric cancer. <i>Journal of Pathology</i> , <b>2013</b> , 229, 621-9	9.4	138
86	'North Sea' progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. <i>Brain</i> , <b>2013</b> , 136, 1146-54	11.2	49



85	Autosomal dominant vasovagal syncope: clinical features and linkage to chromosome 15q26. <i>Neurology</i> , <b>2013</b> , 80, 1485-93	6.5	15
84	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 1417-23	5.6	90
83	Challenges of diagnostic exome sequencing in an inbred founder population. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2013</b> , 1, 71-6	2.3	11
82	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. <i>Nature Genetics</i> , <b>2012</b> , 44, 1188-90	36.3	253
81	Autosomal-recessive congenital cerebellar ataxia is caused by mutations in metabotropic glutamate receptor 1. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 553-64	11	67
80	Strikingly different clinicopathological phenotypes determined by progranulin-mutation dosage. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 1102-7	11	336
79	Autosomal dominant congenital spinal muscular atrophy: a true form of spinal muscular atrophy caused by early loss of anterior horn cells. <i>Brain</i> , <b>2012</b> , 135, 1714-23	11.2	18
78	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. <i>Journal of Biological Chemistry</i> , <b>2012</b> , 287, 20652-63	5.4	42
77	Familial adult myoclonic epilepsy: recognition of mild phenotypes and refinement of the 2q locus. <i>Archives of Neurology</i> , <b>2012</b> , 69, 474-81		25
76	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> , <b>2011</b> , 3, 57	14.4	57
75	An ENU-induced mutation of Cdh23 causes congenital hearing loss, but no vestibular dysfunction, in mice. <i>American Journal of Pathology</i> , <b>2011</b> , 179, 903-14	5.8	23
74	Reducing the exome search space for mendelian diseases using genetic linkage analysis of exome genotypes. <i>Genome Biology</i> , <b>2011</b> , 12, R85	18.3	66
73	Human and mouse mutations in WDR35 cause short-rib polydactyly syndromes due to abnormal ciliogenesis. <i>American Journal of Human Genetics</i> , <b>2011</b> , 88, 508-15	11	106
72	A mutation in synaptojanin 2 causes progressive hearing loss in the ENU-mutagenised mouse strain Mozart. <i>PLoS ONE</i> , <b>2011</b> , 6, e17607	3.7	32
71	Loss-of-function mutations of ILDR1 cause autosomal-recessive hearing impairment DFNB42. <i>American Journal of Human Genetics</i> , <b>2011</b> , 88, 127-37	11	87
70	Kufs disease, the major adult form of neuronal ceroid lipofuscinosis, caused by mutations in CLN6. <i>American Journal of Human Genetics</i> , <b>2011</b> , 88, 566-73	11	114
69	A mutation in the Golgi Qb-SNARE gene GOSR2 causes progressive myoclonus epilepsy with early ataxia. <i>American Journal of Human Genetics</i> , <b>2011</b> , 88, 657-63	11	85
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50	Mutations in the first MyTH4 domain of MYO15A are a common cause of DFNB3 hearing loss. <i>Laryngoscope</i> , <b>2009</b> , 119, 727-33	3.6	38

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47	IL28B is associated with response to chronic hepatitis C interferon-alpha and ribavirin therapy. <i>Nature Genetics</i> , <b>2009</b> , 41, 1100-4	36.3	1636
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44	A mouse model of harlequin ichthyosis delineates a key role for Abca12 in lipid homeostasis. <i>PLoS Genetics</i> , <b>2008</b> , 4, e1000192	6	63
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39	Mutations in contactin-1, a neural adhesion and neuromuscular junction protein, cause a familial form of lethal congenital myopathy. <i>American Journal of Human Genetics</i> , <b>2008</b> , 83, 714-24	11	62
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10	Real time, field-deployable whole genome sequencing of malaria parasites using nanopore technology		1
9	Identity-by-descent analyses for measuring population dynamics and selection in recombining pathogens		3
8	Detecting tandem repeat expansions in cohorts sequenced with short-read sequencing data		3
7	Cross-platform genetic discovery of small molecule products of metabolism and application to clinical outcomes		8
6	Genetic Disruption of Serine Biosynthesis is a Key Driver of Macular Telangiectasia Type 2 Etiology and Progression		3
5	SIS-seq, a molecular time machine connects single cell fate with gene programs		3
4	ExpansionHunter Denovo: A computational method for locating known and novel repeat expansions in short-read sequencing data		1
3	UKB.COVID19: an R package for UK Biobank COVID-19 data processing and analysis. <i>F1000Research</i> , 10, 830	3.6	
2	Multi-omic Analysis of Developing Human Retina and Organoids Reveals Cell-Specific Cis-Regulatory Elements and Mechanisms of Non-Coding Genetic Disease Risk		2
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