Melanie Bahlo

List of Publications by Citations

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	IL28B is associated with response to chronic hepatitis C interferon-alpha and ribavirin therapy. *Nature Genetics*, **2009**, 41, 1100-4	36.3	1636
209	Development of plasmacytoid and conventional dendritic cell subtypes from single precursor cells derived in vitro and in vivo. *Nature Immunology*, **2007**, 8, 1217-26	19.1	608
208	Iron-overload-related disease in HFE hereditary hemochromatosis. *New England Journal of Medicine* , **2008**, 358, 221-30	59.2	516
207	Genome-wide association study identifies new multiple sclerosis susceptibility loci on chromosomes 12 and 20. *Nature Genetics*, **2009**, 41, 824-8	36.3	432
206	Strikingly different clinicopathological phenotypes determined by progranulin-mutation dosage. *American Journal of Human Genetics*, **2012**, 90, 1102-7	11	336
205	Genome-wide meta-analysis identifies novel multiple sclerosis susceptibility loci. *Annals of Neurology*, **2011**, 70, 897-912	9.4	263
204	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. *Nature Genetics*, **2012**, 44, 1188-90	36.3	253
203	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, 673-84	11	205
202	Mutation of the mitochondrial tyrosyl-tRNA synthetase gene, YARS2, causes myopathy, lactic acidosis, and sideroblastic anemia--MLASA syndrome. *American Journal of Human Genetics*, **2010**, 87, 52-9	11	190
201	The Tasmanian devil transcriptome reveals Schwann cell origins of a clonally transmissible cancer. *Science*, **2010**, 327, 84-7	33.3	188
200	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, 365-76	11	173
199	Inference from gene trees in a subdivided population. *Theoretical Population Biology*, **2000**, 57, 79-95	1.2	156
198	Mutations in RAB39B cause X-linked intellectual disability and early-onset Parkinson disease with □synuclein pathology. *American Journal of Human Genetics*, **2014**, 95, 729-35	11	153
197	Identification and analysis of error types in high-throughput genotyping. *American Journal of Human Genetics*, **2000**, 67, 727-36	11	150
196	A Cav3.2 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, 371-80	6.6	147
195	An □E-catenin (CTNNA1) mutation in hereditary diffuse gastric cancer. *Journal of Pathology*, **2013**, 229, 621-9	9.4	138
194	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. *Nature Genetics*, **2014**, 46, 1239-44	36.3	130

193	Kufs disease, the major adult form of neuronal ceroid lipofuscinosis, caused by mutations in CLN6. *American Journal of Human Genetics*, **2011**, 88, 566-73	11	114
192	Genome-wide linkage analysis of the acute coronary syndrome suggests a locus on chromosome 2. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2002**, 22, 874-8	9.4	109
191	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, 328-37	11	108
190	Human and mouse mutations in WDR35 cause short-rib polydactyly syndromes due to abnormal ciliogenesis. *American Journal of Human Genetics*, **2011**, 88, 508-15	11	106
189	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	11	104
188	A focal epilepsy and intellectual disability syndrome is due to a mutation in TBC1D24. *American Journal of Human Genetics*, **2010**, 87, 371-5	11	100
187	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, 2888-900	5.6	99
186	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. *F1000Research*, **2018**, 7, 1297	3.6	95
185	Serine and Lipid Metabolism in Macular Disease and Peripheral Neuropathy. *New England Journal of Medicine*, **2019**, 381, 1422-1433	59.2	91
184	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. *Human Molecular Genetics*, **2013**, 22, 1417-23	5.6	90
183	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator NPRL3. *Annals of Neurology*, **2016**, 79, 132-7	9.4	90
182	Polycomb repressive complex 2 (PRC2) restricts hematopoietic stem cell activity. *PLoS Biology*, **2008**, 6, e93	9.7	88
181	Loss-of-function mutations of ILDR1 cause autosomal-recessive hearing impairment DFNB42. *American Journal of Human Genetics*, **2011**, 88, 127-37	11	87
180	Dominantly inherited ataxia and dysphonia with dentate calcification: spinocerebellar ataxia type 20. *Brain*, **2004**, 127, 1172-81	11.2	87
179	A mutation in the Golgi Qb-SNARE gene GOSR2 causes progressive myoclonus epilepsy with early ataxia. *American Journal of Human Genetics*, **2011**, 88, 657-63	11	85
178	Genetic dissection of the human leukocyte antigen region by use of haplotypes of Tasmanians with multiple sclerosis. *American Journal of Human Genetics*, **2002**, 70, 1125-37	11	81
177	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	11	80
176	Genome-wide analyses identify common variants associated with macular telangiectasia type 2. *Nature Genetics*, **2017**, 49, 559-567	36.3	75

175	Familial cortical dysplasia type IIA caused by a germline mutation in DEPDC5. *Annals of Clinical and Translational Neurology*, **2015**, 2, 575-80	5.3	73
174	High-resolution characterization of sequence signatures due to non-random cleavage of cell-free DNA. *BMC Medical Genomics*, **2015**, 8, 29	3.7	73
173	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. *Journal of Inherited Metabolic Disease*, **2017**, 40, 261-269	5.4	70
172	Autosomal-recessive congenital cerebellar ataxia is caused by mutations in metabotropic glutamate receptor 1. *American Journal of Human Genetics*, **2012**, 91, 553-64	11	67
171	Reducing the exome search space for mendelian diseases using genetic linkage analysis of exome genotypes. *Genome Biology*, **2011**, 12, R85	18.3	66
170	Comparison of clustering tools in R for medium-sized 10x Genomics single-cell RNA-sequencing data. *F1000Research*, **2018**, 7, 1297	3.6	65
169	A mouse model of harlequin ichthyosis delineates a key role for Abca12 in lipid homeostasis. *PLoS Genetics*, **2008**, 4, e1000192	6	63
168	Cpipe: a shared variant detection pipeline designed for diagnostic settings. *Genome Medicine*, **2015**, 7, 68	14.4	62
167	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, 714-24	11	62
166	A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development. *Molecular Psychiatry*, **2019**, 24, 1065-1078	15.1	62
165	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. *Nature Communications*, **2019**, 10, 4919	17.4	58
164	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	14.4	57
163	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. *Nature Genetics*, **2017**, 49, 511-514	36.3	54
162	Small intragenic deletion in FOXP2 associated with childhood apraxia of speech and dysarthria. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2321-6	2.5	54
161	Hemispheric cortical dysplasia secondary to a mosaic somatic mutation in MTOR. *Neurology*, **2015**, 84, 2029-32	6.5	54
160	Plasmodium vivax populations are more genetically diverse and less structured than sympatric Plasmodium falciparum populations. *PLoS Neglected Tropical Diseases*, **2015**, 9, e0003634	4.8	53
159	Generating linkage mapping files from Affymetrix SNP chip data. *Bioinformatics*, **2009**, 25, 1961-2	7.2	52
158	Detecting Expansions of Tandem Repeats in Cohorts Sequenced with Short-Read Sequencing Data. *American Journal of Human Genetics*, **2018**, 103, 858-873	11	51

157	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. *Neurology*, **2016**, 87, 1975-1984	6.5	50
156	A founder mutation in PET100 causes isolated complex IV deficiency in Lebanese individuals with Leigh syndrome. *American Journal of Human Genetics*, **2014**, 94, 209-22	11	49
155	'North Sea' progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. *Brain*, **2013**, 136, 1146-54	11.2	49
154	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, 140-9	4.5	49
153	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
152	Saliva-derived DNA performs well in large-scale, high-density single-nucleotide polymorphism microarray studies. *Cancer Epidemiology Biomarkers and Prevention*, **2010**, 19, 794-8	4	47
151	Agm1/Pgm3-mediated sugar nucleotide synthesis is essential for hematopoiesis and development. *Molecular and Cellular Biology*, **2007**, 27, 5849-59	4.8	45
150	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. *American Journal of Human Genetics*, **2016**, 99, 217-27	11	45
149	A polymorphism in the HLA-DPB1 gene is associated with susceptibility to multiple sclerosis. *PLoS ONE*, **2010**, 5, e13454	3.7	43
148	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, 20652-63	5.4	42
147	Recent advances in the detection of repeat expansions with short-read next-generation sequencing. *F1000Research*, **2018**, 7,	3.6	42
146	Identity-by-descent analyses for measuring population dynamics and selection in recombining pathogens. *PLoS Genetics*, **2018**, 14, e1007279	6	42
145	EIF2S3 Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. *Human Mutation*, **2017**, 38, 409-425	4.7	41
144	A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. *Nature Communications*, **2020**, 11, 3150	17.4	41
143	Genome-wide linkage scan and association study of PARL to the expression of LHON families in Thailand. *Human Genetics*, **2010**, 128, 39-49	6.3	41
142	Multiple sclerosis susceptibility-associated SNPs do not influence disease severity measures in a cohort of Australian MS patients. *PLoS ONE*, **2010**, 5, e10003	3.7	39
141	Mutations in the first MyTH4 domain of MYO15A are a common cause of DFNB3 hearing loss. *Laryngoscope*, **2009**, 119, 727-33	3.6	38
140	Systematic noise degrades gene co-expression signals but can be corrected. *BMC Bioinformatics*, **2015**, 16, 309	3.6	35

139	Dating rare mutations from small samples with dense marker data. *Genetics*, **2014**, 197, 1315-27	4	35
138	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, 573-80	6.3	35
137	Blood pressure QTLs identified by genome-wide linkage analysis and dependence on associated phenotypes. *Physiological Genomics*, **2002**, 8, 99-105	3.6	35
136	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	11.5	34
135	CYLD is a causative gene for frontotemporal dementia - amyotrophic lateral sclerosis. *Brain*, **2020**, 143, 783-799	11.2	33
134	Mutation of the nuclear lamin gene LMNB2 in progressive myoclonus epilepsy with early ataxia. *Human Molecular Genetics*, **2015**, 24, 4483-90	5.6	33
133	A mutation in synaptojanin 2 causes progressive hearing loss in the ENU-mutagenised mouse strain Mozart. *PLoS ONE*, **2011**, 6, e17607	3.7	32
132	A novel splice site mutation in EYA4 causes DFNA10 hearing loss. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1599-604	2.5	32
131	X chromosome association testing in genome wide association studies. *Genetic Epidemiology*, **2011**, 35, 664-70	2.6	31
130	The Genetics of Epilepsy. *Annual Review of Genomics and Human Genetics*, **2020**, 21, 205-230	9.7	30
129	An ethyl-nitrosourea-induced point mutation in phex causes exon skipping, x-linked hypophosphatemia, and rickets. *American Journal of Pathology*, **2002**, 161, 1925-33	5.8	30
128	dtangle: accurate and robust cell type deconvolution. *Bioinformatics*, **2019**, 35, 2093-2099	7.2	30
127	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. *American Journal of Human Genetics*, **2017**, 101, 516-524	11	29
126	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. *Genome Biology*, **2020**, 21, 102	18.3	29
125	Severe childhood speech disorder: Gene discovery highlights transcriptional dysregulation. *Neurology*, **2020**, 94, e2148-e2167	6.5	28
124	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, 2586-90	9.5	28
123	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. *PLoS Genetics*, **2018**, 14, e1007281	6	27
122	Mutations in SH3PXD2B cause Borrone dermato-cardio-skeletal syndrome. *European Journal of Human Genetics*, **2014**, 22, 741-7	5.3	26

121	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. *Genetics in Medicine*, **2019**, 21, 948-954	8.1	26
120	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. *Neurology: Genetics*, **2015**, 1, e16	3.8	25
119	Familial adult myoclonic epilepsy: recognition of mild phenotypes and refinement of the 2q locus. *Archives of Neurology*, **2012**, 69, 474-81		25
118	A novel X-linked form of congenital fiber-type disproportion. *Annals of Neurology*, **2005**, 58, 767-72	9.4	25
117	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, 16625-30	11.5	24
116	Whole exome sequencing combined with linkage analysis identifies a novel 3 bp deletion in NR5A1. *European Journal of Human Genetics*, **2015**, 23, 486-93	5.3	23
115	An ENU-induced mutation of Cdh23 causes congenital hearing loss, but no vestibular dysfunction, in mice. *American Journal of Pathology*, **2011**, 179, 903-14	5.8	23
114	Mitochondrial Genome Sequence of the Scabies Mite Provides Insight into the Genetic Diversity of Individual Scabies Infections. *PLoS Neglected Tropical Diseases*, **2016**, 10, e0004384	4.8	23
113	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. *Neurology*, **2016**, 86, 713-22	6.5	22
112	Harnessing gene expression networks to prioritize candidate epileptic encephalopathy genes. *PLoS ONE*, **2014**, 9, e102079	3.7	22
111	GNE myopathy in Roma patients homozygous for the p.I618T founder mutation. *Neuromuscular Disorders*, **2015**, 25, 713-8	2.9	21
110	Coalescence time for two genes from a subdivided population. *Journal of Mathematical Biology*, **2001**, 43, 397-410	2	21
109	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). *Neurology*, **2016**, 87, 579-84	6.5	19
108	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 821-30	5.3	19
107	Evaluation of non-coding variation in GLUT1 deficiency. *Developmental Medicine and Child Neurology*, **2016**, 58, 1295-1302	3.3	19
106	Autosomal dominant congenital spinal muscular atrophy: a true form of spinal muscular atrophy caused by early loss of anterior horn cells. *Brain*, **2012**, 135, 1714-23	11.2	18
105	A novel mutation in COCH-implications for genotype-phenotype correlations in DFNA9 hearing loss. *Laryngoscope*, **2010**, 120, 2489-93	3.6	18
104	SNP selection for genes of iron metabolism in a study of genetic modifiers of hemochromatosis. *BMC Medical Genetics*, **2008**, 9, 18	2.1	18

103	On the utility of data from the International HapMap Project for Australian association studies. *Human Genetics*, **2006**, 119, 220-2	6.3	18
102	A cross-platform approach identifies genetic regulators of human metabolism and health. *Nature Genetics*, **2021**, 53, 54-64	36.3	18
101	Use of copy number deletion polymorphisms to assess DNA chimerism. *Clinical Chemistry*, **2014**, 60, 1105-14	5.5	17
100	Cochlear implants for DFNA17 deafness. *Laryngoscope*, **2006**, 116, 2211-5	3.6	17
99	Identifying nineteenth century genealogical links from genotypes. *Human Genetics*, **2005**, 117, 188-99	6.3	17
98	Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. *Brain Communications*, **2021**, 3, fcaa235	4.5	17
97	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. *Neuron*, **2019**, 104, 665-679.e8	13.9	16
96	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	5.6	16
95	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	4.8	16
94	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2-2q11.2. *Human Genetics*, **2016**, 135, 1117-25	6.3	16
93	Investigating and correcting plasma DNA sequencing coverage bias to enhance aneuploidy discovery. *PLoS ONE*, **2014**, 9, e86993	3.7	15
92	Autosomal dominant vasovagal syncope: clinical features and linkage to chromosome 15q26. *Neurology*, **2013**, 80, 1485-93	6.5	15
91	In silico prioritization based on coexpression can aid epileptic encephalopathy gene discovery. *Neurology: Genetics*, **2016**, 2, e51	3.8	15
90	Early neuroimaging markers of FOXP2 intragenic deletion. *Scientific Reports*, **2016**, 6, 35192	4.9	15
89	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. *Human Molecular Genetics*, **2015**, 24, 2000-10	5.6	14
88	Neuropathology of childhood-onset basal ganglia degeneration caused by mutation of. *Annals of Clinical and Translational Neurology*, **2017**, 4, 859-864	5.3	14
87	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, 555-8	2.5	14
86	Heterozygous mutations in cause juvenile peroxisomal D-bifunctional protein deficiency. *Neurology: Genetics*, **2016**, 2, e114	3.8	14

85	A recombination hotspot leads to sequence variability within a novel gene (AK005651) and contributes to type 1 diabetes susceptibility. *Genome Research*, **2010**, 20, 1629-38	9.7	13
84	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, 177-86	3.2	12
83	Clinical spectrum of the pentanucleotide repeat expansion in the gene in ataxia syndromes. *Neurology*, **2020**, 95, e2912-e2923	6.5	12
82	Pathogenic Variants in GPC4 Cause Keipert Syndrome. *American Journal of Human Genetics*, **2019**, 104, 914-924	11	11
81	Identical by descent L1CAM mutation in two apparently unrelated families with intellectual disability without L1 syndrome. *European Journal of Medical Genetics*, **2015**, 58, 364-8	2.6	11
80	Challenges of diagnostic exome sequencing in an inbred founder population. *Molecular Genetics & Genomic Medicine*, **2013**, 1, 71-6	2.3	11
79	XIBD: software for inferring pairwise identity by descent on the X chromosome. *Bioinformatics*, **2016**, 32, 2389-91	7.2	10
78	Candidate disease gene prediction using Gentrepid: application to a genome-wide association study on coronary artery disease. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 44-57	2.3	10
77	Mapping of the Plasmodium chabaudi resistance locus char2. *Infection and Immunity*, **2006**, 74, 5814-9	3.7	10
76	Detecting genome wide haplotype sharing using SNP or microsatellite haplotype data. *Human Genetics*, **2006**, 119, 38-50	6.3	10
75	Multiple sclerosis risk variants regulate gene expression in innate and adaptive immune cells. *Life Science Alliance*, **2020**, 3,	5.8	10
74	Progressive myoclonus epilepsies-Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. *American Journal of Human Genetics*, **2021**, 108, 722-738	11	10
73	Recessive Spondylocarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in MYH3. *American Journal of Human Genetics*, **2018**, 102, 1115-1125	11	10
72	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. *European Journal of Medical Genetics*, **2017**, 60, 437-443	2.6	9
71	Dorsal language stream anomalies in an inherited speech disorder. *Brain*, **2019**, 142, 966-977	11.2	9
70	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	5.3	9
69	Identification of a novel RNF213 variant in a family with heterogeneous intracerebral vasculopathy. *International Journal of Stroke*, **2014**, 9, E26-7	6.3	9
68	Analysis of genome-wide association study data using the protein knowledge base. *BMC Genetics*, **2011**, 12, 98	2.6	9

67	Molecular characterization of a novel X-linked syndrome involving developmental delay and deafness. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2564-75	2.5	9
66	SNPs in putative regulatory regions identified by human mouse comparative sequencing and transcription factor binding site data. *Mammalian Genome*, **2002**, 13, 554-7	3.2	9
65	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. *American Journal of Human Genetics*, **2020**, 107, 977-988	11	9
64	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. *Genetics in Medicine*, **2019**, 21, 2532-2542	8.1	8
63	Using familial information for variant filtering in high-throughput sequencing studies. *Human Genetics*, **2014**, 133, 1331-41	6.3	8
62	Keipert syndrome (Nasodigitoacoustic syndrome) is X-linked and maps to Xq22.2-Xq28. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2236-41	2.5	8
61	Multipoint approximations of identity-by-descent probabilities for accurate linkage analysis of distantly related individuals. *American Journal of Human Genetics*, **2008**, 82, 607-22	11	8
60	Cross-platform genetic discovery of small molecule products of metabolism and application to clinical outcomes		8
59	SNP barcodes provide higher resolution than microsatellite markers to measure Plasmodium vivax population genetics. *Malaria Journal*, **2020**, 19, 375	3.6	8
58	Fatal Enteroviral Encephalitis in a Patient with Common Variable Immunodeficiency Harbouring a Novel Mutation in NFKB2. *Journal of Clinical Immunology*, **2019**, 39, 324-335	5.7	7
57	Myoclonic occipital photosensitive epilepsy with dystonia (MOPED): A familial epilepsy syndrome. *Epilepsy Research*, **2015**, 114, 98-105	3	7
56	Evidence for a common genetic aetiology in high-risk families with multiple haematological malignancy subtypes. *British Journal of Haematology*, **2010**, 150, 456-62	4.5	7
55	Systemic lipid dysregulation is a risk factor for macular neurodegenerative disease. *Scientific Reports*, **2020**, 10, 12165	4.9	7
54	Comparing the frequency of common genetic variants and haplotypes between carriers and non-carriers of BRCA1 and BRCA2 deleterious mutations in Australian women diagnosed with breast cancer before 40 years of age. *BMC Cancer*, **2010**, 10, 466	4.8	6
53	Reanalysis and optimisation of bioinformatic pipelines is critical for mutation detection. *Human Mutation*, **2019**, 40, 374-379	4.7	6
52	Rapid Diagnosis of Spinocerebellar Ataxia 36 in a Three-Generation Family Using Short-Read Whole-Genome Sequencing Data. *Movement Disorders*, **2020**, 35, 1675-1679	7	5
51	Inherited RORB pathogenic variants: Overlap of photosensitive genetic generalized and occipital lobe epilepsy. *Epilepsia*, **2020**, 61, e23-e29	6.4	5
50	Callosal agenesis and congenital mirror movements: outcomes associated with DCC mutations. *Developmental Medicine and Child Neurology*, **2020**, 62, 758-762	3.3	5

49	brain-coX: investigating and visualising gene co-expression in seven human brain transcriptomic datasets. *Genome Medicine*, **2017**, 9, 55	14.4	5
48	Segregating sites in a gene conversion model with mutation. *Theoretical Population Biology*, **1998**, 54, 243-56	1.2	5
47	Identification of genetic factors influencing metabolic dysregulation and retinal support for MacTel, a retinal disorder. *Communications Biology*, **2021**, 4, 274	6.7	5
46	Clonal multi-omics reveals Bcor as a negative regulator of emergency dendritic cell development. *Immunity*, **2021**, 54, 1338-1351.e9	32.3	5
45	Contribution of rare genetic variants to drug response in absence epilepsy. *Epilepsy Research*, **2021**, 170, 106537	3	5
44	SCN1A clinical spectrum includes the self-limited focal epilepsies of childhood. *Epilepsy Research*, **2017**, 131, 9-14	3	4
43	Epidemiology and etiology of infantile developmental and epileptic encephalopathies in Tasmania. *Epilepsia Open*, **2019**, 4, 504-510	4	4
42	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	3.6	4
41	Generation of seven iPSC lines from peripheral blood mononuclear cells suitable to investigate Autism Spectrum Disorder. *Stem Cell Research*, **2019**, 39, 101516	1.6	4
40	Probabilistic analysis of recessive mutagenesis screen strategies. *Mammalian Genome*, **2007**, 18, 5-22	3.2	4
39	Genetic Analysis of Patients Who Experienced Awareness with Recall while under General Anesthesia. *Anesthesiology*, **2019**, 131, 974-982	4.3	4
38	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	3.2	4
37	Comparative genomics revealed adaptive admixture in in Africa. *Microbial Genomics*, **2021**, 7,	4.4	4
36	The advantages of dense marker sets for linkage analysis with very large families. *Human Genetics*, **2007**, 121, 459-68	6.3	3
35	Identity-by-descent analyses for measuring population dynamics and selection in recombining pathogens		3
34	Detecting tandem repeat expansions in cohorts sequenced with short-read sequencing data		3
33	Genetic Disruption of Serine Biosynthesis is a Key Driver of Macular Telangiectasia Type 2 Etiology and Progression		3
32	SIS-seq, a molecular time machine connects single cell fate with gene programs		3

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