Vincent Plagnol

List of Publications by Year
in descending order

174 papers	24,874 citations	10351 72 h-index	8138 148 g-index
181 all docs	181 docs citations	181 times ranked	42169 citing authors

#	Article	IF	Citations
1	Bayesian Test for Colocalisation between Pairs of Genetic Association Studies Using Summary Statistics. PLoS Genetics, 2014, 10, e1004383.	1.5	2,012
2	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. Nature Genetics, 2009, 41, 703-707.	9.4	1,513
3	Robust associations of four new chromosome regions from genome-wide analyses of type 1 diabetes. Nature Genetics, 2007, 39, 857-864.	9.4	1,324
4	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	13.7	1,014
5	The Pattern of Polymorphism in Arabidopsis thaliana. PLoS Biology, 2005, 3, e196.	2.6	895
6	Imputation of sequence variants for identification of genetic risks for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet, The, 2011, 377, 641-649.	6.3	845
7	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720.	13.7	737
8	Dense genotyping identifies and localizes multiple common and rare variant association signals in celiac disease. Nature Genetics, 2011, 43, 1193-1201.	9.4	682
9	Shared and Distinct Genetic Variants in Type 1 Diabetes and Celiac Disease. New England Journal of Medicine, 2008, 359, 2767-2777.	13.9	654
10	Phosphoinositide 3-Kinase Î´ Gene Mutation Predisposes to Respiratory Infection and Airway Damage. Science, 2013, 342, 866-871.	6.0	541
11	A robust model for read count data in exome sequencing experiments and implications for copy number variant calling. Bioinformatics, 2012, 28, 2747-2754.	1.8	534
12	Recombination and linkage disequilibrium in Arabidopsis thaliana. Nature Genetics, 2007, 39, 1151-1155.	9.4	497
13	Meta-analysis of genome-wide association study data identifies additional type 1 diabetes risk loci. Nature Genetics, 2008, 40, 1399-1401.	9.4	456
14	Atlas of the clinical genetics of human dilated cardiomyopathy. European Heart Journal, 2015, 36, 1123-1135.	1.0	456
15	Large-scale genetic fine mapping and genotype-phenotype associations implicate polymorphism in the IL2RA region in type 1 diabetes. Nature Genetics, 2007, 39, 1074-1082.	9.4	380
16	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	2.6	343
17	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. American Journal of Human Genetics, 2016, 98, 500-513.	2.6	333
18	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323

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19	Inflammatory Skin and Bowel Disease Linked toADAM17Deletion. New England Journal of Medicine, 2011, 365, 1502-1508.	13.9	285
20	DYX1C1 is required for axonemal dynein assembly and ciliary motility. Nature Genetics, 2013, 45, 995-1003.	9.4	256
21	Cell-specific protein phenotypes for the autoimmune locus IL2RA using a genotype-selectable human bioresource. Nature Genetics, 2009, 41, 1011-1015.	9.4	249
22	Possible Ancestral Structure in Human Populations. PLoS Genetics, 2006, 2, e105.	1.5	235
23	The Effect of Inhaled IFN-Î² on Worsening of Asthma Symptoms Caused by Viral Infections. A Randomized Trial. American Journal of Respiratory and Critical Care Medicine, 2014, 190, 145-154.	2.5	231
24	Genome-Wide Association Analysis of Autoantibody Positivity in Type 1 Diabetes Cases. PLoS Genetics, 2011, 7, e1002216.	1.5	230
25	Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. American Journal of Human Genetics, 2012, 91, 1041-1050.	2.6	224
26	Human Coronavirus OC43 Associated with Fatal Encephalitis. New England Journal of Medicine, 2016, 375, 497-498.	13.9	224
27	Copy number of FCGR3B, which is associated with systemic lupus erythematosus, correlates with protein expression and immune complex uptake. Journal of Experimental Medicine, 2008, 205, 1573-1582.	4.2	213
28	Genetic complexity in hypertrophic cardiomyopathy revealed by high-throughput sequencing. Journal of Medical Genetics, 2013, 50, 228-239.	1.5	203
29	Dissection of the genetics of Parkinson's disease identifies an additional association 5' of SNCA and multiple associated haplotypes at 17q21. Human Molecular Genetics, 2011, 20, 345-353.	1.4	202
30	Genome-wide analysis of allelic expression imbalance in human primary cells by high-throughput transcriptome resequencing. Human Molecular Genetics, 2010, 19, 122-134.	1.4	197
31	Constitutional Mutations in RTEL1 Cause Severe Dyskeratosis Congenita. American Journal of Human Genetics, 2013, 92, 448-453.	2.6	191
32	Negligible impact of rare autoimmune-locus coding-region variants on missing heritability. Nature, 2013, 498, 232-235.	13.7	184
33	Biallelic RIPK1 mutations in humans cause severe immunodeficiency, arthritis, and intestinal inflammation. Science, 2018, 361, 810-813.	6.0	181
34	Astrovirus VA1/HMO-C: An Increasingly Recognized Neurotropic Pathogen in Immunocompromised Patients. Clinical Infectious Diseases, 2015, 60, 881-888.	2.9	173
35	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	3.7	169
36	Poly(A)-specific ribonuclease deficiency impacts telomere biology and causes dyskeratosis congenita. Journal of Clinical Investigation, 2015, 125, 2151-2160.	3.9	165

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37	RHBDF2 Mutations Are Associated with Tylosis, a Familial Esophageal Cancer Syndrome. American Journal of Human Genetics, 2012, 90, 340-346.	2.6	162
38	The Evolution of Selfing in Arabidopsis thaliana. Science, 2007, 317, 1070-1072.	6.0	160
39	Susceptibility to tuberculosis is associated with variants in the ASAP1 gene encoding a regulator of dendritic cell migration. Nature Genetics, 2015, 47, 523-527.	9.4	156
40	A robust statistical method for case-control association testing with copy number variation. Nature Genetics, 2008, 40, 1245-1252.	9.4	151
41	Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553.	2.8	148
42	Mice with endogenous <scp>TDP</scp> â€43 mutations exhibit gain of splicing function and characteristics of amyotrophic lateral sclerosis. EMBO Journal, 2018, 37, .	3.5	129
43	Recursive splicing in long vertebrate genes. Nature, 2015, 521, 371-375.	13.7	128
44	Exome sequencing identifiesDYNC2H1mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. Journal of Medical Genetics, 2013, 50, 309-323.	1.5	127
45	Novel genotypeâ€“phenotype associations demonstrated by high-throughput sequencing in patients with hypertrophic cardiomyopathy. Heart, 2015, 101, 294-301.	1.2	124
46	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	1.4	122
47	Plasma urate concentration and risk of coronary heart disease: a Mendelian randomisation analysis. Lancet Diabetes and Endocrinology,the, 2016, 4, 327-336.	5.5	122
48	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	2.6	121
49	GWAS and colocalization analyses implicate carotid intima-media thickness and carotid plaque loci in cardiovascular outcomes. Nature Communications, 2018, 9, 5141.	5.8	119
50	Development of a highly sensitive liquid biopsy platform to detect clinically-relevant cancer mutations at low allele fractions in cell-free DNA. PLoS ONE, 2018, 13, e0194630.	1.1	117
51	Recessive Mutations in KCNJ13, Encoding an Inwardly Rectifying Potassium Channel Subunit, Cause Leber Congenital Amaurosis. American Journal of Human Genetics, 2011, 89, 183-190.	2.6	116
52	Detecting Ancient Admixture and Estimating Demographic Parameters in Multiple Human Populations. Molecular Biology and Evolution, 2009, 26, 1823-1827.	3.5	113
53	A common single-nucleotide variant in T is strongly associated with chordoma. Nature Genetics, 2012, 44, 1185-1187.	9.4	112
54	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	2.6	109

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55	Fine-Scale Survey of X Chromosome Copy Number Variants and Indels Underlying Intellectual Disability. American Journal of Human Genetics, 2010, 87, 173-188.	2.6	107
56	Whole exome sequencing of familial hypercholesterolaemia patients negative forLDLR/APOB/PCSK9mutations. Journal of Medical Genetics, 2014, 51, 537-544.	1.5	104
57	Biallelic JAK1 mutations in immunodeficient patient with mycobacterial infection. Nature Communications, 2016, 7, 13992.	5.8	104
58	Limited Clinical Utility of Non-invasive Prenatal Testing for Subchromosomal Abnormalities. American Journal of Human Genetics, 2016, 98, 34-44.	2.6	101
59	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	1.5	96
60	Humanized mutant FUS drives progressive motor neuron degeneration without aggregation in â€˜FUSDelta14â€™ knockin mice. Brain, 2017, 140, 2797-2805.	3.7	95
61	Exome Sequencing Identifies Autosomal-Dominant SRP72 Mutations Associated with Familial Aplasia and Myelodysplasia. American Journal of Human Genetics, 2012, 90, 888-892.	2.6	94
62	Neuropathy target esterase impairments cause Oliverâ€“McFarlane and Laurenceâ€“Moon syndromes. Journal of Medical Genetics, 2015, 52, 85-94.	1.5	91
63	LRBA gene deletion in a patient presenting with autoimmunity without hypogammaglobulinemia. Journal of Allergy and Clinical Immunology, 2012, 130, 1428-1432.	1.5	90
64	Analytical validation of a next generation sequencing liquid biopsy assay for high sensitivity broad molecular profiling. PLoS ONE, 2018, 13, e0193802.	1.1	90
65	Statistical independence of the colocalized association signals for type 1 diabetes and RPS26 gene expression on chromosome 12q13. Biostatistics, 2009, 10, 327-334.	0.9	89
66	RP1L1Variants are Associated with a Spectrum of Inherited Retinal Diseases Including Retinitis Pigmentosa and Occult Macular Dystrophy. Human Mutation, 2013, 34, 506-514.	1.1	87
67	DNAJC21 Mutations Link a Cancer-Prone Bone Marrow Failure Syndrome to Corruption in 60S Ribosome Subunit Maturation. American Journal of Human Genetics, 2016, 99, 115-124.	2.6	85
68	Mutations in AQP5, Encoding a Water-Channel Protein, Cause Autosomal-Dominant Diffuse Nonepidermolytic Palmoplantar Keratoderma. American Journal of Human Genetics, 2013, 93, 330-335.	2.6	82
69	A Transcriptomic Signature of the Hypothalamic Response to Fasting and BDNF Deficiency in Prader-Willi Syndrome. Cell Reports, 2018, 22, 3401-3408.	2.9	81
70	Effects of Collection and Processing Procedures on Plasma Circulating Cell-Free DNA from Cancer Patients. Journal of Molecular Diagnostics, 2018, 20, 883-892.	1.2	81
71	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. Nature Communications, 2020, 11, 1044.	5.8	81
72	Genome-wide association study of age-related macular degeneration identifies associated variants in the TNXBâ€“FKBPLâ€“NOTCH4 region of chromosome 6p21.3. Human Molecular Genetics, 2012, 21, 4138-4150.	1.4	80

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73	ISPD gene mutations are a common cause of congenital and limb-girdle muscular dystrophies. Brain, 2013, 136, 269-281.	3.7	80
74	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. American Journal of Human Genetics, 2014, 94, 915-923.	2.6	79
75	PTPN22 Trp620 Explains the Association of Chromosome 1p13 With Type 1 Diabetes and Shows a Statistical Interaction With HLA Class II Genotypes. Diabetes, 2008, 57, 1730-1737.	0.3	78
76	Signal transducer and activator of transcription 2 deficiency is a novel disorder of mitochondrial fission. Brain, 2015, 138, 2834-2846.	3.7	78
77	Deletions at 22q11.2 in idiopathic Parkinson's disease: a combined analysis of genome-wide association data. Lancet Neurology, The, 2016, 15, 585-596.	4.9	77
78	Use of targeted exome sequencing as a diagnostic tool for Familial Hypercholesterolaemia. Journal of Medical Genetics, 2012, 49, 644-649.	1.5	74
79	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	2.7	74
80	Genome-wide association study in multiple human prion diseases suggests genetic risk factors additional to PRNP. Human Molecular Genetics, 2012, 21, 1897-1906.	1.4	73
81	Integrated Polygenic Tool Substantially Enhances Coronary Artery Disease Prediction. Circulation Genomic and Precision Medicine, 2021, 14, e003304.	1.6	73
82	Autosomal-Dominant Corneal Endothelial Dystrophies CHED1 and PPCD1 Are Allelic Disorders Caused by Non-coding Mutations in the Promoter of OVOL2. American Journal of Human Genetics, 2016, 98, 75-89.	2.6	70
83	FUS ALS-causative mutations impair FUS autoregulation and splicing factor networks through intron retention. Nucleic Acids Research, 2020, 48, 6889-6905.	6.5	70
84	CNV Analysis in Tourette Syndrome Implicates Large Genomic Rearrangements in COL8A1 and NRXN1. PLoS ONE, 2013, 8, e59061.	1.1	70
85	Diagnostic yield of molecular autopsy in patients with sudden arrhythmic death syndrome using targeted exome sequencing. Europace, 2016, 18, 888-896.	0.7	69
86	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	2.6	67
87	Immunodeficiency and severe susceptibility to bacterial infection associated with a loss-of-function homozygous mutation of MKL1. Blood, 2015, 126, 1527-1535.	0.6	66
88	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	1.5	66
89	TRNT1 deficiency: clinical, biochemical and molecular genetic features. Orphanet Journal of Rare Diseases, 2016, 11, 90.	1.2	64
90	Novel PLCG2 Mutation in a Patient With APLAID and Cutis Laxa. Frontiers in Immunology, 2018, 9, 2863.	2.2	64

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91	A Method to Address Differential Bias in Genotyping in Large-Scale Association Studies. PLoS Genetics, 2007, 3, e74.	1.5	63
92	A Homozygous Mutation in the<scp>TUB</scp>Gene Associated with Retinal Dystrophy and Obesity. Human Mutation, 2014, 35, 289-293.	1.1	63
93	Cryptogenic multifocal ulcerating stenosing enteritis associated with homozygous deletion mutations in cytosolic phospholipase A2-Î±. Gut, 2014, 63, 96-104.	6.1	62
94	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	2.6	61
95	Mutations in TUBGCP4 Alter Microtubule Organization via the Î³-Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. American Journal of Human Genetics, 2015, 96, 666-674.	2.6	60
96	The Phenotypic Variability of Retinal Dystrophies Associated With Mutations in CRX, With Report of a Novel Macular Dystrophy Phenotype. Investigative Ophthalmology and Visual Science, 2014, 55, 6934-6944.	3.3	59
97	ERCC6L2 Mutations Link a Distinct Bone-Marrow-Failure Syndrome to DNA Repair and Mitochondrial Function. American Journal of Human Genetics, 2014, 94, 246-256.	2.6	58
98	Interactions between Idd5.1/Ctla4 and Other Type 1 Diabetes Genes. Journal of Immunology, 2007, 179, 8341-8349.	0.4	54
99	Genetic Complexity of Crohnâ€™s Disease in Two Large Ashkenazi Jewish Families. Gastroenterology, 2016, 151, 698-709.	0.6	54
100	A Frameshift in CSF2RB Predominant Among Ashkenazi Jews Increases Risk for Crohn's Disease and Reduces Monocyte Signaling via GM-CSF. Gastroenterology, 2016, 151, 710-723.e2.	0.6	51
101	Extreme Clonality in Lymphoblastoid Cell Lines with Implications for Allele Specific Expression Analyses. PLoS ONE, 2008, 3, e2966.	1.1	50
102	Validation of an Integrated Risk Tool, Including Polygenic Risk Score, for Atherosclerotic Cardiovascular Disease in Multiple Ethnicities and Ancestries. American Journal of Cardiology, 2021, 148, 157-164.	0.7	48
103	Mutations in CPAMD8 Cause a Unique Form of Autosomal-Recessive Anterior Segment Dysgenesis. American Journal of Human Genetics, 2016, 99, 1338-1352.	2.6	47
104	RNA-Seq of Huntingtonâ€™s disease patient myeloid cells reveals innate transcriptional dysregulation associated with proinflammatory pathway activation. Human Molecular Genetics, 2016, 25, ddw142.	1.4	47
105	Biâ€allelic CLPB mutations cause cataract, renal cysts, nephrocalcinosis and 3â€methylglutaconic aciduria, a novel disorder of mitochondrial protein disaggregation. Journal of Inherited Metabolic Disease, 2015, 38, 211-219.	1.7	46
106	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. PLoS ONE, 2013, 8, e70724.	1.1	45
107	Huntingtonâ€™s disease blood and brain show a common gene expression pattern and share an immune signature with Alzheimerâ€™s disease. Scientific Reports, 2017, 7, 44849.	1.6	45
108	Bidirectional nucleolar dysfunction in C9orf72 frontotemporal lobar degeneration. Acta Neuropathologica Communications, 2017, 5, 29.	2.4	43

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109	Exome sequencing identifies MPL as a causative gene in familial aplastic anemia. Haematologica, 2012, 97, 524-528.	1.7	42
110	Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. American Journal of Human Genetics, 2015, 96, 948-954.	2.6	42
111	Deep sequencing reveals persistence of cell-associated mumps vaccine virus in chronic encephalitis. Acta Neuropathologica, 2017, 133, 139-147.	3.9	41
112	Biallelic Mutations in PLA2G5, Encoding Group V Phospholipase A2, Cause Benign Fleck Retina. American Journal of Human Genetics, 2011, 89, 782-791.	2.6	40
113	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	1.8	40
114	Genetic variants alter T-bet binding and gene expression in mucosal inflammatory disease. PLoS Genetics, 2017, 13, e1006587.	1.5	40
115	Population Genomics of Cardiometabolic Traits: Design of the University College London-London School of Hygiene and Tropical Medicine-Edinburgh-Bristol (UCLEB) Consortium. PLoS ONE, 2013, 8, e71345.	1.1	39
116	Immunodeficiency and disseminated mycobacterial infection associated with homozygous nonsense mutation of IKKÎ². Journal of Allergy and Clinical Immunology, 2014, 134, 215-218.e3.	1.5	37
117	Gain-of-function CEBPE mutation causes noncanonical autoinflammatory inflammasomopathy. Journal of Allergy and Clinical Immunology, 2019, 144, 1364-1376.	1.5	37
118	Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. Neurobiology of Aging, 2014, 35, 1491-1498.	1.5	36
119	Loss-of-Function Mutations in CAST Cause Peeling Skin, Leukonychia, Acral Punctate Keratoses, Cheilitis, and Knuckle Pads. American Journal of Human Genetics, 2015, 96, 440-447.	2.6	36
120	Study of the genetic variability in a Parkinson's Disease gene: EIF4G1. Neuroscience Letters, 2012, 518, 19-22.	1.0	35
121	Marked overlap of four genetic syndromes with dyskeratosis congenita confounds clinical diagnosis. Haematologica, 2016, 101, 1180-1189.	1.7	34
122	Brittle Cornea Syndrome ZNF469 Mutation Carrier Phenotype and Segregation Analysis of Rare ZNF469 Variants in Familial Keratoconus. Investigative Ophthalmology and Visual Science, 2015, 56, 578-586.	3.3	33
123	Heterozygous deletions at the ZEB1 locus verify haploinsufficiency as the mechanism of disease for posterior polymorphous corneal dystrophy type 3. European Journal of Human Genetics, 2016, 24, 985-991.	1.4	33
124	Bayesian mixture analysis for metagenomic community profiling. Bioinformatics, 2015, 31, 2930-2938.	1.8	31
125	Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049.	1.4	29
126	MIR-NATs repress MAPT translation and aid proteostasis in neurodegeneration. Nature, 2021, 594, 117-123.	13.7	29

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127	RAPIDR: an analysis package for non-invasive prenatal testing of aneuploidy. Bioinformatics, 2014, 30, 2965-2967.	1.8	28
128	Association of CHRDL1 Mutations and Variants with X-linked Megalocornea, NeuhÃuser Syndrome and Central Corneal Thickness. PLoS ONE, 2014, 9, e104163.	1.1	27
129	Loss-of-Function Mutations in SERPINB8 Linked to Exfoliative Ichthyosis with Impaired Mechanical Stability of Intercellular Adhesions. American Journal of Human Genetics, 2016, 99, 430-436.	2.6	27
130	Relative Influences of Crossing Over and Gene Conversion on the Pattern of Linkage Disequilibrium in Arabidopsis thaliana. Genetics, 2006, 172, 2441-2448.	1.2	26
131	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	2.6	26
132	Digital PCR analysis of circulating tumor DNA: a biomarker for chondrosarcoma diagnosis, prognostication, and residual disease detection. Cancer Medicine, 2017, 6, 2194-2202.	1.3	26
133	Targeted Sequence Capture and High-Throughput Sequencing in the Molecular Diagnosis of Ichthyosis and Other Skin Diseases. Journal of Investigative Dermatology, 2013, 133, 573-576.	0.3	25
134	KohlschÃ¼tter-TÃ¶nz Syndrome: Mutations inROGDIand Evidence of Genetic Heterogeneity. Human Mutation, 2013, 34, 296-300.	1.1	24
135	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	1.1	23
136	Peripheral tissues reprogram CD8+ T cells for pathogenicity during graft-versus-host disease. JCI Insight, 2018, 3, .	2.3	23
137	Clinical characteristics of early retinal disease due to CDHR1 mutation. Molecular Vision, 2013, 19, 2250-9.	1.1	22
138	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. American Journal of Human Genetics, 2016, 98, 857-868.	2.6	21
139	Detection and correction of artefacts in estimation of rare copy number variants and analysis of rare deletions in type 1 diabetes. Human Molecular Genetics, 2015, 24, 1774-1790.	1.4	20
140	Downregulated Wnt/Î²-catenin signalling in the Down syndrome hippocampus. Scientific Reports, 2019, 9, 7322.	1.6	20
141	Deficiency of the zinc finger protein ZFP106 causes motor and sensory neurodegeneration. Human Molecular Genetics, 2016, 25, 291-307.	1.4	19
142	Topoisomerase 2Î² mutation impairs early B-cell development. Blood, 2020, 135, 1497-1501.	0.6	18
143	A Genome-Wide Assessment of the Role of Untagged Copy Number Variants in Type 1 Diabetes. PLoS Genetics, 2014, 10, e1004367.	1.5	17
144	Profilin1 E117G is a moderate risk factor for amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 506-508.	0.9	17

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145	Variants WithinTSC2Exons 25 and 31 Are Very Unlikely to Cause Clinically Diagnosable Tuberous Sclerosis. Human Mutation, 2016, 37, 364-370.	1.1	16
146	Post-GWAS methodologies for localisation of functional non-coding variants: ANGPTL3. Atherosclerosis, 2016, 246, 193-201.	0.4	15
147	Common variable immunodeficiency and natural killer cell lymphopenia caused by Ets-binding site mutation in the IL-2 receptor Î³ (IL2RG) gene promoter. Journal of Allergy and Clinical Immunology, 2016, 137, 940-942.e4.	1.5	14
148	Disease Expression in Autosomal Recessive Retinal Dystrophy Associated With Mutations in theDRAM2Gene. , 2015, 56, 8083.		13
149	A recurrent splice-site mutation in EPHA2 causing congenital posterior nuclear cataract. Ophthalmic Genetics, 2018, 39, 236-241.	0.5	13
150	Fluorescence Intensity Normalisation: Correcting for Time Effects in Large-Scale Flow Cytometric Analysis. Advances in Bioinformatics, 2009, 2009, 1-6.	5.7	12
151	A novel frameshift MSX1 mutation in a Saudi family with autosomal dominant premolar and third molar agenesis. Archives of Oral Biology, 2015, 60, 982-988.	0.8	12
152	Graphical modelling of molecular networks underlying sporadic inclusion body myositis. Molecular BioSystems, 2013, 9, 1736.	2.9	10
153	Tubular aggregates caused by serine active site containing 1 (<scp>SERAC1</scp>) mutations in a patient with a mitochondrial encephalopathy. Neuropathology and Applied Neurobiology, 2015, 41, 399-402.	1.8	10
154	Exome sequencing reveals a novel partial deletion in the progranulin gene causing primary progressive aphasia. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 1411-1412.	0.9	9
155	Exome Sequencing of 75 Individuals from Multiply Affected Coeliac Families and Large Scale Resequencing Follow Up. PLoS ONE, 2015, 10, e0116845.	1.1	8
156	CHCHD10Pro34Ser is not a highly penetrant pathogenic variant for amyotrophic lateral sclerosis and frontotemporal dementia. Brain, 2016, 139, e9-e9.	3.7	7
157	TDP-43 mutations increase HNRNP A1-7B through gain of splicing function. Brain, 2018, 141, e83-e83.	3.7	7
158	Association tests and software for copy number variant data. Human Genomics, 2009, 3, 191.	1.4	6
159	Variants in KCNJ11 and BAD do not predict response to ketogenic dietary therapies for epilepsy. Epilepsy Research, 2015, 118, 22-28.	0.8	6
160	The ophthalmic presentation of Hermanskyâ€“Pudlak syndrome 6. British Journal of Ophthalmology, 2016, 100, 1521-1524.	2.1	6
161	A Comparison of Low Read Depth QuantSeq 3â€² Sequencing to Total RNA-Seq in FUS Mutant Mice. Frontiers in Genetics, 2020, 11, 562445.	1.1	6
162	Validity of the Familyâ€Based Association Test for Copy Number Variant Data in the Case of Nonâ€Linear Intensityâ€Genotype Relationship. Genetic Epidemiology, 2012, 36, 895-898.	0.6	3

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163	P3.02b-102 Osimertinib Benefit in ctDNA T790M Positive, EGFR-Mutant NSCLC Patients. Journal of Thoracic Oncology, 2017, 12, S1254-S1255.	0.5	3
164	Copy Number Variant Association Studies. , 2011, , 215-230.		1
165	A method to address differential bias in genotyping in large scale association studies. PLoS Genetics, 2005, preprint, e74.	1.5	1
166	Possible ancestral structure in human populations. PLoS Genetics, 2005, preprint, e105.	1.5	1
167	A Genome-wide Assessment of the Genetic Basis of Type 1 Diabetes. Clinical Immunology, 2007, 123, S136.	1.4	0
168	F.5. Cell-specific CD25 Expression is Determined by Type 1 Diabetes Associated IL2RA Haplotypes. Clinical Immunology, 2009, 131, S94.	1.4	0
169	B17â€...Blood transcriptome replicates dysregulation found in human huntingtonâ€™s disease brain and shares an immune signature with alzheimerâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A15.1-A15.	0.9	0
170	B15â€...Innate transcriptional dysregulation is associated with proinflammatory pathway activation in huntingtonâ€™s disease myeloid cells. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A14.1-A14.	0.9	0
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