Vincent Plagnol

List of Publications by Year
in descending order

174	24,874	10373 72	8156 148
papers	citations	h-index	g-index

181	181	181	42169
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Integrated Polygenic Tool Substantially Enhances Coronary Artery Disease Prediction. Circulation Genomic and Precision Medicine, 2021, 14, e003304.	1.6	73
2	MIR-NATs repress MAPT translation and aid proteostasis in neurodegeneration. Nature, 2021, 594, 117-123.	13.7	29
3	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
4	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
5	Topoisomerase 2Î² mutation impairs early B-cell development. Blood, 2020, 135, 1497-1501.	0.6	18
6	FUS ALS-causative mutations impair FUS autoregulation and splicing factor networks through intron retention. Nucleic Acids Research, 2020, 48, 6889-6905.	6.5	70
7	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. Nature Communications, 2020, 11, 1044.	5.8	81
8	Gain-of-function CEBPE mutation causes noncanonical autoinflammatory inflammasomopathy. Journal of Allergy and Clinical Immunology, 2019, 144, 1364-1376.	1.5	37
9	Downregulated Wnt/Î²-catenin signalling in the Down syndrome hippocampus. Scientific Reports, 2019, 9, 7322.	1.6	20
10	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
11	A recurrent splice-site mutation in EPHA2 causing congenital posterior nuclear cataract. Ophthalmic Genetics, 2018, 39, 236-241.	0.5	13
12	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	1.1	23
13	Novel PLCG2 Mutation in a Patient With APLAID and Cutis Laxa. Frontiers in Immunology, 2018, 9, 2863.	2.2	64
14	GWAS and colocalization analyses implicate carotid intima-media thickness and carotid plaque loci in cardiovascular outcomes. Nature Communications, 2018, 9, 5141.	5.8	119
15	TDP-43 mutations increase HNRNP A1-7B through gain of splicing function. Brain, 2018, 141, e83-e83.	3.7	7
16	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
17	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
18	Biallelic RIPK1 mutations in humans cause severe immunodeficiency, arthritis, and intestinal inflammation. Science, 2018, 361, 810-813.	6.0	181

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19	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	1.5	66
20	Peripheral tissues reprogram CD8+ T cells for pathogenicity during graft-versus-host disease. JCI Insight, 2018, 3, .	2.3	23
21	Analytical validation of a next generation sequencing liquid biopsy assay for high sensitivity broad molecular profiling. PLoS ONE, 2018, 13, e0193802.	1.1	90
22	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
23	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
24	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
25	Bidirectional nucleolar dysfunction in C9orf72 frontotemporal lobar degeneration. Acta Neuropathologica Communications, 2017, 5, 29.	2.4	43
26	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	1.8	40
27	P2.03b-093 Validation and Performance of a Standardized ctDNA NGS Assay across Two Laboratories. Journal of Thoracic Oncology, 2017, 12, S992-S993.	0.5	0
28	P3.02b-102 Osimertinib Benefit in ctDNA T790M Positive, EGFR-Mutant NSCLC Patients. Journal of Thoracic Oncology, 2017, 12, S1254-S1255.	0.5	3
29	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
30	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
31	Humanized mutant FUS drives progressive motor neuron degeneration without aggregation in â€˜FUSDelta14â€™ knockin mice. Brain, 2017, 140, 2797-2805.	3.7	95
32	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
33	Deep sequencing reveals persistence of cell-associated mumps vaccine virus in chronic encephalitis. Acta Neuropathologica, 2017, 133, 139-147.	3.9	41
34	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323
35	Genetic variants alter T-bet binding and gene expression in mucosal inflammatory disease. PLoS Genetics, 2017, 13, e1006587.	1.5	40
36	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

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37	Variants WithinTSC2Exons 25 and 31 Are Very Unlikely to Cause Clinically Diagnosable Tuberous Sclerosis. Human Mutation, 2016, 37, 364-370.	1.1	16
38	Genetic Complexity of Crohnâ€™s Disease in Two Large Ashkenazi Jewish Families. Gastroenterology, 2016, 151, 698-709.	0.6	54
39	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
40	Biallelic JAK1 mutations in immunodeficient patient with mycobacterial infection. Nature Communications, 2016, 7, 13992.	5.8	104
41	TRNT1 deficiency: clinical, biochemical and molecular genetic features. Orphanet Journal of Rare Diseases, 2016, 11, 90.	1.2	64
42	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
43	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
44	Marked overlap of four genetic syndromes with dyskeratosis congenita confounds clinical diagnosis. Haematologica, 2016, 101, 1180-1189.	1.7	34
45	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
46	Human Coronavirus OC43 Associated with Fatal Encephalitis. New England Journal of Medicine, 2016, 375, 497-498.	13.9	224
47	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	2.6	121
48	Expanding the Phenotype ofTRNT1-Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. JAMA Ophthalmology, 2016, 134, 1049.	1.4	29
49	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
50	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
51	Deficiency of the zinc finger protein ZFP106 causes motor and sensory neurodegeneration. Human Molecular Genetics, 2016, 25, 291-307.	1.4	19
52	Post-GWAS methodologies for localisation of functional non-coding variants: ANGPTL3. Atherosclerosis, 2016, 246, 193-201.	0.4	15
53	Liquid biopsies could be superior to tumor biopsy to provide a molecular profile in non-small cell lung cancer (NSCLC) patients. Journal of Thoracic Oncology, 2016, 11, S37.	0.5	0
54	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

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55	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
56	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
57	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
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	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
60	Whole-exome sequencing in the investigation of retinal dystrophy. Lancet, The, 2016, 387, S52.	6.3	0
61	The ophthalmic presentation of Hermanskyâ€“Pudlak syndrome 6. British Journal of Ophthalmology, 2016, 100, 1521-1524.	2.1	6
62	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
63	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
64	Diagnostic yield of molecular autopsy in patients with sudden arrhythmic death syndrome using targeted exome sequencing. Europace, 2016, 18, 888-896.	0.7	69
65	CHCHD10Pro34Ser is not a highly penetrant pathogenic variant for amyotrophic lateral sclerosis and frontotemporal dementia. Brain, 2016, 139, e9-e9.	3.7	7
66	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
67	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	2.7	74
68	Disease Expression in Autosomal Recessive Retinal Dystrophy Associated With Mutations in theDRAM2Gene. , 2015, 56, 8083.		13
69	Exome Sequencing of 75 Individuals from Multiply Affected Coeliac Families and Large Scale Resequencing Follow Up. PLoS ONE, 2015, 10, e0116845.	1.1	8
70	Poly(A)-specific ribonuclease deficiency impacts telomere biology and causes dyskeratosis congenita. Journal of Clinical Investigation, 2015, 125, 2151-2160.	3.9	165
71	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
72	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

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73	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	2.6	109
74	Variants in KCNJ11 and BAD do not predict response to ketogenic dietary therapies for epilepsy. Epilepsy Research, 2015, 118, 22-28.	0.8	6
75	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
76	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
77	Astrovirus VA1/HMO-C: An Increasingly Recognized Neurotropic Pathogen in Immunocompromised Patients. Clinical Infectious Diseases, 2015, 60, 881-888.	2.9	173
78	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
79	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	1.5	96
80	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
81	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
82	Recursive splicing in long vertebrate genes. Nature, 2015, 521, 371-375.	13.7	128
83	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
84	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
85	Signal transducer and activator of transcription 2 deficiency is a novel disorder of mitochondrial fission. Brain, 2015, 138, 2834-2846.	3.7	78
86	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	13.7	1,014
87	Bayesian mixture analysis for metagenomic community profiling. Bioinformatics, 2015, 31, 2930-2938.	1.8	31
88	Novel genotypeâ€“phenotype associations demonstrated by high-throughput sequencing in patients with hypertrophic cardiomyopathy. Heart, 2015, 101, 294-301.	1.2	124
89	Neuropathy target esterase impairments cause Oliverâ€“McFarlane and Laurenceâ€–Moon syndromes. Journal of Medical Genetics, 2015, 52, 85-94.	1.5	91
90	Atlas of the clinical genetics of human dilated cardiomyopathy. European Heart Journal, 2015, 36, 1123-1135.	1.0	456

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91	Association of CHRDL1 Mutations and Variants with X-linked Megalocornea, NeuhÃuser Syndrome and Central Corneal Thickness. PLoS ONE, 2014, 9, e104163.	1.1	27
92	A Genome-Wide Assessment of the Role of Untagged Copy Number Variants in Type 1 Diabetes. PLoS Genetics, 2014, 10, e1004367.	1.5	17
93	Profilin1 E117G is a moderate risk factor for amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 506-508.	0.9	17
94	Bayesian Test for Colocalisation between Pairs of Genetic Association Studies Using Summary Statistics. PLoS Genetics, 2014, 10, e1004383.	1.5	2,012
95	A Homozygous Mutation in the<scp>TUB</scp>Gene Associated with Retinal Dystrophy and Obesity. Human Mutation, 2014, 35, 289-293.	1.1	63
96	Cryptogenic multifocal ulcerating stenosing enteritis associated with homozygous deletion mutations in cytosolic phospholipase A2-Î±. Gut, 2014, 63, 96-104.	6.1	62
97	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
98	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	3.7	169
99	RAPIDR: an analysis package for non-invasive prenatal testing of aneuploidy. Bioinformatics, 2014, 30, 2965-2967.	1.8	28
100	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
101	Whole exome sequencing of familial hypercholesterolaemia patients negative forLDLR/APOB/PCSK9mutations. Journal of Medical Genetics, 2014, 51, 537-544.	1.5	104
102	Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. Neurobiology of Aging, 2014, 35, 1491-1498.	1.5	36
103	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	2.6	67
104	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
105	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
106	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
107	A Systems Immunology Approach to Graft-Versus-Host Disease. Blood, 2014, 124, 3812-3812.	0.6	0
108	DYX1C1 is required for axonemal dynein assembly and ciliary motility. Nature Genetics, 2013, 45, 995-1003.	9.4	256

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109	Phosphoinositide 3-Kinase Î´ Gene Mutation Predisposes to Respiratory Infection and Airway Damage. Science, 2013, 342, 866-871.	6.0	541
110	KohlschÃ¼tter-TÃ¶nz Syndrome: Mutations inROGDIand Evidence of Genetic Heterogeneity. Human Mutation, 2013, 34, 296-300.	1.1	24
111	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
112	Constitutional Mutations in RTEL1 Cause Severe Dyskeratosis Congenita. American Journal of Human Genetics, 2013, 92, 448-453.	2.6	191
113	Graphical modelling of molecular networks underlying sporadic inclusion body myositis. Molecular BioSystems, 2013, 9, 1736.	2.9	10
114	Negligible impact of rare autoimmune-locus coding-region variants on missing heritability. Nature, 2013, 498, 232-235.	13.7	184
115	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
116	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
117	Genetic complexity in hypertrophic cardiomyopathy revealed by high-throughput sequencing. Journal of Medical Genetics, 2013, 50, 228-239.	1.5	203
118	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
119	ISPD gene mutations are a common cause of congenital and limb-girdle muscular dystrophies. Brain, 2013, 136, 269-281.	3.7	80
120	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
121	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
122	Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553.	2.8	148
123	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. PLoS ONE, 2013, 8, e70724.	1.1	45
124	CNV Analysis in Tourette Syndrome Implicates Large Genomic Rearrangements in COL8A1 and NRXN1. PLoS ONE, 2013, 8, e59061.	1.1	70
125	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
126	Clinical characteristics of early retinal disease due to CDHR1 mutation. Molecular Vision, 2013, 19, 2250-9.	1.1	22

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127	Use of targeted exome sequencing as a diagnostic tool for Familial Hypercholesterolaemia. Journal of Medical Genetics, 2012, 49, 644-649.	1.5	74
128	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
129	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
130	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
131	Exome sequencing identifies MPL as a causative gene in familial aplastic anemia. Haematologica, 2012, 97, 524-528.	1.7	42
132	Study of the genetic variability in a Parkinson's Disease gene: EIF4G1. Neuroscience Letters, 2012, 518, 19-22.	1.0	35
133	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
134	LRBA gene deletion in a patient presenting with autoimmunity without hypogammaglobulinemia. Journal of Allergy and Clinical Immunology, 2012, 130, 1428-1432.	1.5	90
135	Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. American Journal of Human Genetics, 2012, 91, 1041-1050.	2.6	224
136	A common single-nucleotide variant in T is strongly associated with chordoma. Nature Genetics, 2012, 44, 1185-1187.	9.4	112
137	RHBDF2 Mutations Are Associated with Tylosis, a Familial Esophageal Cancer Syndrome. American Journal of Human Genetics, 2012, 90, 340-346.	2.6	162
138	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
139	Inflammatory Skin and Bowel Disease Linked toADAM17Deletion. New England Journal of Medicine, 2011, 365, 1502-1508.	13.9	285
140	Dense genotyping identifies and localizes multiple common and rare variant association signals in celiac disease. Nature Genetics, 2011, 43, 1193-1201.	9.4	682
141	Copy Number Variant Association Studies. , 2011, , 215-230.		1
142	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
143	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
144	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

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145	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
146	Genome-Wide Association Analysis of Autoantibody Positivity in Type 1 Diabetes Cases. PLoS Genetics, 2011, 7, e1002216.	1.5	230
147	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
148	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
149	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
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	Detecting Ancient Admixture and Estimating Demographic Parameters in Multiple Human Populations. Molecular Biology and Evolution, 2009, 26, 1823-1827.	3.5	113
152	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
153	F.5. Cell-specific CD25 Expression is Determined by Type 1 Diabetes Associated IL2RA Haplotypes. Clinical Immunology, 2009, 131, S94.	1.4	0
154	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
155	Cell-specific protein phenotypes for the autoimmune locus IL2RA using a genotype-selectable human bioresource. Nature Genetics, 2009, 41, 1011-1015.	9.4	249
156	Association tests and software for copy number variant data. Human Genomics, 2009, 3, 191.	1.4	6
157	A robust statistical method for case-control association testing with copy number variation. Nature Genetics, 2008, 40, 1245-1252.	9.4	151
158	Meta-analysis of genome-wide association study data identifies additional type 1 diabetes risk loci. Nature Genetics, 2008, 40, 1399-1401.	9.4	456
159	Shared and Distinct Genetic Variants in Type 1 Diabetes and Celiac Disease. New England Journal of Medicine, 2008, 359, 2767-2777.	13.9	654
160	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
161	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
162	Extreme Clonality in Lymphoblastoid Cell Lines with Implications for Allele Specific Expression Analyses. PLoS ONE, 2008, 3, e2966.	1.1	50

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163	A Method to Address Differential Bias in Genotyping in Large-Scale Association Studies. PLoS Genetics, 2007, 3, e74.	1.5	63
164	Interactions between Idd5.1/Ctla4 and Other Type 1 Diabetes Genes. Journal of Immunology, 2007, 179, 8341-8349.	0.4	54
165	The Evolution of Selfing in Arabidopsis thaliana. Science, 2007, 317, 1070-1072.	6.0	160
166	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
167	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
168	Recombination and linkage disequilibrium in Arabidopsis thaliana. Nature Genetics, 2007, 39, 1151-1155.	9.4	497
169	A Genome-wide Assessment of the Genetic Basis of Type 1 Diabetes. Clinical Immunology, 2007, 123, S136.	1.4	0
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