

Vincent Plagnol

List of Publications by Year in descending order

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Version: 2024-02-01

174
papers

24,874
citations

10351

72
h-index

8138

148
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181
all docs

181
docs citations

181
times ranked

42169
citing authors

#	ARTICLE	IF	CITATIONS
1	Integrated Polygenic Tool Substantially Enhances Coronary Artery Disease Prediction. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003304.	1.6	73
2	MIR-NATs repress MAPT translation and aid proteostasis in neurodegeneration. <i>Nature</i> , 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. <i>American Journal of Cardiology</i> , 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. <i>Frontiers in Genetics</i> , 2020, 11, 562445.	1.1	6
5	Topoisomerase 2Î² mutation impairs early B-cell development. <i>Blood</i> , 2020, 135, 1497-1501.	0.6	18
6	FUS ALS-causative mutations impair FUS autoregulation and splicing factor networks through intron retention. <i>Nucleic Acids Research</i> , 2020, 48, 6889-6905.	6.5	70
7	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. <i>Nature Communications</i> , 2020, 11, 1044.	5.8	81
8	Gain-of-function CEBPE mutation causes noncanonical autoinflammatory inflammasomopathy. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 1364-1376.	1.5	37
9	Downregulated Wnt/Î²-catenin signalling in the Down syndrome hippocampus. <i>Scientific Reports</i> , 2019, 9, 7322.	1.6	20
10	A Transcriptomic Signature of the Hypothalamic Response to Fasting and BDNF Deficiency in Prader-Willi Syndrome. <i>Cell Reports</i> , 2018, 22, 3401-3408.	2.9	81
11	A recurrent splice-site mutation in <i>EPHA2</i> causing congenital posterior nuclear cataract. <i>Ophthalmic Genetics</i> , 2018, 39, 236-241.	0.5	13
12	Missense variants in the X-linked gene <i>PRPS1</i> cause retinal degeneration in females. <i>Human Mutation</i> , 2018, 39, 80-91.	1.1	23
13	Novel PLCG2 Mutation in a Patient With APLAID and Cutis Laxa. <i>Frontiers in Immunology</i> , 2018, 9, 2863.	2.2	64
14	GWAS and colocalization analyses implicate carotid intima-media thickness and carotid plaque loci in cardiovascular outcomes. <i>Nature Communications</i> , 2018, 9, 5141.	5.8	119
15	TDP-43 mutations increase HNRNP A1-7B through gain of splicing function. <i>Brain</i> , 2018, 141, e83-e83.	3.7	7
16	Effects of Collection and Processing Procedures on Plasma Circulating Cell-Free DNA from Cancer Patients. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 883-892.	1.2	81
17	Mice with endogenous <i>TDP</i> 43 mutations exhibit gain of splicing function and characteristics of amyotrophic lateral sclerosis. <i>EMBO Journal</i> , 2018, 37, .	3.5	129
18	Biallelic <i>RIPK1</i> mutations in humans cause severe immunodeficiency, arthritis, and intestinal inflammation. <i>Science</i> , 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	Huntington's disease blood and brain show a common gene expression pattern and share an immune signature with Alzheimer'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 FUS Δ 14 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 Parkinson'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 Within <i>TSC2</i> Exons 25 and 31 Are Very Unlikely to Cause Clinically Diagnosable Tuberous Sclerosis. <i>Human Mutation</i> , 2016, 37, 364-370.	1.1	16
38	Genetic Complexity of Crohn's Disease in Two Large Ashkenazi Jewish Families. <i>Gastroenterology</i> , 2016, 151, 698-709.	0.6	54
39	B17's...Blood transcriptome replicates dysregulation found in human huntington's disease brain and shares an immune signature with alzheimer's disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, A15.1-A15.	0.9	0
40	Biallelic JAK1 mutations in immunodeficient patient with mycobacterial infection. <i>Nature Communications</i> , 2016, 7, 13992.	5.8	104
41	TRNT1 deficiency: clinical, biochemical and molecular genetic features. <i>Orphanet Journal of Rare Diseases</i> , 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 β (IL2RG) gene promoter. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 137, 940-942.e4.	1.5	14
43	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. <i>American Journal of Human Genetics</i> , 2016, 98, 857-868.	2.6	21
44	Marked overlap of four genetic syndromes with dyskeratosis congenita confounds clinical diagnosis. <i>Haematologica</i> , 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. <i>American Journal of Human Genetics</i> , 2016, 99, 430-436.	2.6	27
46	Human Coronavirus OC43 Associated with Fatal Encephalitis. <i>New England Journal of Medicine</i> , 2016, 375, 497-498.	13.9	224
47	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2016, 99, 1305-1315.	2.6	121
48	Expanding the Phenotype of <i>TRNT1</i> -Related Immunodeficiency to Include Childhood Cataract and Inner Retinal Dysfunction. <i>JAMA Ophthalmology</i> , 2016, 134, 1049.	1.4	29
49	Mutations in CPAMD8 Cause a Unique Form of Autosomal-Recessive Anterior Segment Dysgenesis. <i>American Journal of Human Genetics</i> , 2016, 99, 1338-1352.	2.6	47
50	B15's...Innate transcriptional dysregulation is associated with proinflammatory pathway activation in huntington's disease myeloid cells. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, A14.1-A14.	0.9	0
51	Deficiency of the zinc finger protein ZFP106 causes motor and sensory neurodegeneration. <i>Human Molecular Genetics</i> , 2016, 25, 291-307.	1.4	19
52	Post-GWAS methodologies for localisation of functional non-coding variants: ANGPTL3. <i>Atherosclerosis</i> , 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. <i>Journal of Thoracic Oncology</i> , 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. <i>American Journal of Human Genetics</i> , 2016, 98, 75-89.	2.6	70

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55	RNA-Seq of Huntington's disease patient myeloid cells reveals innate transcriptional dysregulation associated with proinflammatory pathway activation. <i>Human Molecular Genetics</i> , 2016, 25, ddw142.	1.4	47
56	DNAJC21 Mutations Link a Cancer-Prone Bone Marrow Failure Syndrome to Corruption in 60S Ribosome Subunit Maturation. <i>American Journal of Human Genetics</i> , 2016, 99, 115-124.	2.6	85
57	Plasma urate concentration and risk of coronary heart disease: a Mendelian randomisation analysis. <i>Lancet Diabetes and Endocrinology</i> , 2016, 4, 327-336.	5.5	122
58	Limited Clinical Utility of Non-invasive Prenatal Testing for Subchromosomal Abnormalities. <i>American Journal of Human Genetics</i> , 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. <i>Lancet Neurology</i> , 2016, 15, 585-596.	4.9	77
60	Whole-exome sequencing in the investigation of retinal dystrophy. <i>Lancet</i> , 2016, 387, S52.	6.3	0
61	The ophthalmic presentation of Hermansky-Pudlak syndrome 6. <i>British Journal of Ophthalmology</i> , 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. <i>American Journal of Human Genetics</i> , 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. <i>European Journal of Human Genetics</i> , 2016, 24, 985-991.	1.4	33
64	Diagnostic yield of molecular autopsy in patients with sudden arrhythmic death syndrome using targeted exome sequencing. <i>Europace</i> , 2016, 18, 888-896.	0.7	69
65	CHCHD10 Pro34Ser is not a highly penetrant pathogenic variant for amyotrophic lateral sclerosis and frontotemporal dementia. <i>Brain</i> , 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. <i>Blood</i> , 2015, 126, 1527-1535.	0.6	66
67	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. <i>EBioMedicine</i> , 2015, 2, 1063-1070.	2.7	74
68	Disease Expression in Autosomal Recessive Retinal Dystrophy Associated With Mutations in the DRAM2 Gene. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 8083.		13
69	Exome Sequencing of 75 Individuals from Multiply Affected Coeliac Families and Large Scale Resequencing Follow Up. <i>PLoS ONE</i> , 2015, 10, e0116845.	1.1	8
70	Poly(A)-specific ribonuclease deficiency impacts telomere biology and causes dyskeratosis congenita. <i>Journal of Clinical Investigation</i> , 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. <i>Nature Genetics</i> , 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. <i>Human Molecular Genetics</i> , 2015, 24, 1774-1790.	1.4	20

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73	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. <i>American Journal of Human Genetics</i> , 2015, 96, 938-947.	2.6	109
74	Variants in KCNJ11 and BAD do not predict response to ketogenic dietary therapies for epilepsy. <i>Epilepsy Research</i> , 2015, 118, 22-28.	0.8	6
75	Biallelic <i>CLPB</i> mutations cause cataract, renal cysts, nephrocalcinosis and 3-methylglutaconic aciduria, a novel disorder of mitochondrial protein disaggregation. <i>Journal of Inherited Metabolic Disease</i> , 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. <i>American Journal of Human Genetics</i> , 2015, 96, 440-447.	2.6	36
77	Astrovirus VA1/HMO-C: An Increasingly Recognized Neurotropic Pathogen in Immunocompromised Patients. <i>Clinical Infectious Diseases</i> , 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. <i>Investigative Ophthalmology and Visual Science</i> , 2015, 56, 578-586.	3.3	33
79	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. <i>Neurobiology of Aging</i> , 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. <i>Archives of Oral Biology</i> , 2015, 60, 982-988.	0.8	12
81	Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. <i>American Journal of Human Genetics</i> , 2015, 96, 948-954.	2.6	42
82	Recursive splicing in long vertebrate genes. <i>Nature</i> , 2015, 521, 371-375.	13.7	128
83	Mutations in TUBGCP4 Alter Microtubule Organization via the β -Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. <i>American Journal of Human Genetics</i> , 2015, 96, 666-674.	2.6	60
84	Tubular aggregates caused by serine active site containing 1 (<i>SERAC1</i>) mutations in a patient with a mitochondrial encephalopathy. <i>Neuropathology and Applied Neurobiology</i> , 2015, 41, 399-402.	1.8	10
85	Signal transducer and activator of transcription 2 deficiency is a novel disorder of mitochondrial fission. <i>Brain</i> , 2015, 138, 2834-2846.	3.7	78
86	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , 2015, 526, 82-90.	13.7	1,014
87	Bayesian mixture analysis for metagenomic community profiling. <i>Bioinformatics</i> , 2015, 31, 2930-2938.	1.8	31
88	Novel genotype-phenotype associations demonstrated by high-throughput sequencing in patients with hypertrophic cardiomyopathy. <i>Heart</i> , 2015, 101, 294-301.	1.2	124
89	Neuropathy target esterase impairments cause Oliver-McFarlane and Laurence-Moon syndromes. <i>Journal of Medical Genetics</i> , 2015, 52, 85-94.	1.5	91
90	Atlas of the clinical genetics of human dilated cardiomyopathy. <i>European Heart Journal</i> , 2015, 36, 1123-1135.	1.0	456

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91	Association of CHRD1 Mutations and Variants with X-linked Megalocornea, Neuhäuser 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 <i>TUB</i> 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- ζ . Gut, 2014, 63, 96-104.	6.1	62
97	The Effect of Inhaled IFN- γ 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	Parkinson'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 for <i>LDLR</i> , <i>APOB</i> , <i>PCSK9</i> mutations. 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 IKK ζ . 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 $\hat{\Gamma}$ Gene Mutation Predisposes to Respiratory Infection and Airway Damage. <i>Science</i> , 2013, 342, 866-871.	6.0	541
110	Kohlschütter-TÄ¶niz Syndrome: Mutations in <i>ROGDI</i> and Evidence of Genetic Heterogeneity. <i>Human Mutation</i> , 2013, 34, 296-300.	1.1	24
111	Mutations in AQP5, Encoding a Water-Channel Protein, Cause Autosomal-Dominant Diffuse Nonepidermolytic Palmoplantar Keratoderma. <i>American Journal of Human Genetics</i> , 2013, 93, 330-335.	2.6	82
112	Constitutional Mutations in RTEL1 Cause Severe Dyskeratosis Congenita. <i>American Journal of Human Genetics</i> , 2013, 92, 448-453.	2.6	191
113	Graphical modelling of molecular networks underlying sporadic inclusion body myositis. <i>Molecular BioSystems</i> , 2013, 9, 1736.	2.9	10
114	Negligible impact of rare autoimmune-locus coding-region variants on missing heritability. <i>Nature</i> , 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. <i>Journal of Investigative Dermatology</i> , 2013, 133, 573-576.	0.3	25
116	<i>RP1L1</i> Variants are Associated with a Spectrum of Inherited Retinal Diseases Including Retinitis Pigmentosa and Occult Macular Dystrophy. <i>Human Mutation</i> , 2013, 34, 506-514.	1.1	87
117	Genetic complexity in hypertrophic cardiomyopathy revealed by high-throughput sequencing. <i>Journal of Medical Genetics</i> , 2013, 50, 228-239.	1.5	203
118	Exome sequencing reveals a novel partial deletion in the progranulin gene causing primary progressive aphasia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 1411-1412.	0.9	9
119	ISPD gene mutations are a common cause of congenital and limb-girdle muscular dystrophies. <i>Brain</i> , 2013, 136, 269-281.	3.7	80
120	Exome sequencing identifies <i>DYNC2H1</i> mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. <i>Journal of Medical Genetics</i> , 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. <i>Human Molecular Genetics</i> , 2013, 22, 1039-1049.	1.4	122
122	Mutations in the autoregulatory domain of $\hat{\Gamma}$ tubulin 4a cause hereditary dystonia. <i>Annals of Neurology</i> , 2013, 73, 546-553.	2.8	148
123	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. <i>PLoS ONE</i> , 2013, 8, e70724.	1.1	45
124	CNV Analysis in Tourette Syndrome Implicates Large Genomic Rearrangements in COL8A1 and NRXN1. <i>PLoS ONE</i> , 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. <i>PLoS ONE</i> , 2013, 8, e71345.	1.1	39
126	Clinical characteristics of early retinal disease due to CDHR1 mutation. <i>Molecular Vision</i> , 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 TNXB-“FKBPL”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 Family-Based Association Test for Copy Number Variant Data in the Case of Non-Linear Intensity-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 to ADAM17 Deletion. 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. <i>Human Molecular Genetics</i> , 2011, 20, 345-353.	1.4	202
146	Genome-Wide Association Analysis of Autoantibody Positivity in Type 1 Diabetes Cases. <i>PLoS Genetics</i> , 2011, 7, e1002216.	1.5	230
147	Fine-Scale Survey of X Chromosome Copy Number Variants and Indels Underlying Intellectual Disability. <i>American Journal of Human Genetics</i> , 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. <i>Nature</i> , 2010, 464, 713-720.	13.7	737
149	Genome-wide analysis of allelic expression imbalance in human primary cells by high-throughput transcriptome resequencing. <i>Human Molecular Genetics</i> , 2010, 19, 122-134.	1.4	197
150	Fluorescence Intensity Normalisation: Correcting for Time Effects in Large-Scale Flow Cytometric Analysis. <i>Advances in Bioinformatics</i> , 2009, 2009, 1-6.	5.7	12
151	Detecting Ancient Admixture and Estimating Demographic Parameters in Multiple Human Populations. <i>Molecular Biology and Evolution</i> , 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. <i>Biostatistics</i> , 2009, 10, 327-334.	0.9	89
153	F.5. Cell-specific CD25 Expression is Determined by Type 1 Diabetes Associated IL2RA Haplotypes. <i>Clinical Immunology</i> , 2009, 131, S94.	1.4	0
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