Aravinda Chakravarti

List of Publications by Year
in descending order

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316	79,251	2423	567
papers	citations	97 h-index	263 g-index

330	330	330	80436
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
2	Finding the missing heritability of complex diseases. Nature, 2009, 461, 747-753.	13.7	7,490
3	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	13.7	4,137
4	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
5	Discovery and refinement of loci associated with lipid levels. Nature Genetics, 2013, 45, 1274-1283.	9.4	2,641
6	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
7	Defining the role of common variation in the genomic and biological architecture of adult human height. Nature Genetics, 2014, 46, 1173-1186.	9.4	1,818
8	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	13.7	1,788
9	Association between Microdeletion and Microduplication at 16p11.2 and Autism. New England Journal of Medicine, 2008, 358, 667-675.	13.9	1,476
10	Genome-Wide Association Scan Shows Genetic Variants in the FTO Gene Are Associated with Obesity-Related Traits. PLoS Genetics, 2007, 3, e115.	1.5	1,446
11	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
12	DNA duplication associated with Charcot-Marie-Tooth disease type 1A. Cell, 1991, 66, 219-232.	13.5	1,313
13	Genome-wide association study of blood pressure and hypertension. Nature Genetics, 2009, 41, 677-687.	9.4	1,224
14	Patterns of single-nucleotide polymorphisms in candidate genes for blood-pressure homeostasis. Nature Genetics, 1999, 22, 239-247.	9.4	1,040
15	Variations on a Theme: Cataloging Human DNA Sequence Variation. Science, 1997, 278, 1580-1581.	6.0	979
16	Development of Human Protein Reference Database as an Initial Platform for Approaching Systems Biology in Humans. Genome Research, 2003, 13, 2363-2371.	2.4	954
17	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. Nature Genetics, 2018, 50, 1412-1425.	9.4	924
18	A missense mutation of the endothelin-B receptor gene in multigenic hirschsprung's disease. Cell, 1994, 79, 1257-1266.	13.5	877

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19	Common variants associated with plasma triglycerides and risk for coronary artery disease. Nature Genetics, 2013, 45, 1345-1352.	9.4	754
20	A DNA Polymorphism Discovery Resource for Research on Human Genetic Variation: Table 1.. Genome Research, 1998, 8, 1229-1231.	2.4	750
21	New Goals for the U.S. Human Genome Project: 1998-2003. , 1998, 282, 682-689.		711
22	Genomic alterations in cultured human embryonic stem cells. Nature Genetics, 2005, 37, 1099-1103.	9.4	592
23	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
24	A genome-wide linkage and association scan reveals novel loci for autism. Nature, 2009, 461, 802-808.	13.7	570
25	A Common Genetic Variant in the Neurexin Superfamily Member CNTNAP2 Increases Familial Risk of Autism. American Journal of Human Genetics, 2008, 82, 160-164.	2.6	566
26	Population geneticsâ€"making sense out of sequence. Nature Genetics, 1999, 21, 56-60.	9.4	561
27	Meta-analysis identifies six new susceptibility loci for atrial fibrillation. Nature Genetics, 2012, 44, 670-675.	9.4	533
28	Schizophrenia susceptibility loci on chromosomes 13q32 and 8p21. Nature Genetics, 1998, 20, 70-73.	9.4	506
29	A common genetic variant in the NOS1 regulator NOS1AP modulates cardiac repolarization. Nature Genetics, 2006, 38, 644-651.	9.4	500
30	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. Nature Genetics, 2017, 49, 403-415.	9.4	492
31	Automated construction of genetic linkage maps using an expert system (MultiMap): a human genome linkage map. Nature Genetics, 1994, 6, 384-390.	9.4	440
32	A common sex-dependent mutation in a RET enhancer underlies Hirschsprung disease risk. Nature, 2005, 434, 857-863.	13.7	438
33	Common variants in KCNN3 are associated with lone atrial fibrillation. Nature Genetics, 2010, 42, 240-244.	9.4	438
34	Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. Science, 2016, 351, 1166-1171.	6.0	438
35	Haploinsufficiency of telomerase reverse transcriptase leads to anticipation in autosomal dominant dyskeratosis congenita. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 15960-15964.	3.3	423
36	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	9.4	403

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37	Genome-wide association study of PR interval. Nature Genetics, 2010, 42, 153-159.	9.4	400
38	Variants in ZFHX3 are associated with atrial fibrillation in individuals of European ancestry. Nature Genetics, 2009, 41, 879-881.	9.4	363
39	The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. Nature Genetics, 2016, 48, 1171-1184.	9.4	362
40	Common variants at ten loci modulate the QT interval duration in the QTSCD Study. Nature Genetics, 2009, 41, 407-414.	9.4	356
41	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. PLoS Genetics, 2015, 11, e1005378.	1.5	331
42	Multiple loci influence erythrocyte phenotypes in the CHARGE Consortium. Nature Genetics, 2009, 41, 1191-1198.	9.4	324
43	A BDNF Coding Variant is Associated with the NEO Personality Inventory Domain Neuroticism, a Risk Factor for Depression. Neuropsychopharmacology, 2003, 28, 397-401.	2.8	321
44	Common variants in 22 loci are associated with QRS duration and cardiac ventricular conduction. Nature Genetics, 2010, 42, 1068-1076.	9.4	308
45	A homozygous mutation in the endothelin-3 gene associated with a combined Waardenburg type 2 and Hirschsprung phenotype (Shah-Waardenburg syndrome). Nature Genetics, 1996, 12, 445-447.	9.4	296
46	Genome-Wide Association Study of Coronary Heart Disease and Its Risk Factors in 8,090 African Americans: The NHLBI CARe Project. PLoS Genetics, 2011, 7, e1001300.	1.5	290
47	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	9.4	281
48	Genome-wide association analyses using electronic health records identify new loci influencing blood pressure variation. Nature Genetics, 2017, 49, 54-64.	9.4	281
49	Whole-genome association study identifies STK39 as a hypertension susceptibility gene. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 226-231.	3.3	280
50	Parallel Genotyping of Human SNPs Using Generic High-density Oligonucleotide Tag Arrays. Genome Research, 2000, 10, 853-860.	2.4	273
51	Germline mutations in glial cell line-derived neurotrophic factor (GDNF) and RET in a Hirschsprung disease patient. Nature Genetics, 1996, 14, 341-344.	9.4	269
52	Segregation at three loci explains familial and population risk in Hirschsprung disease. Nature Genetics, 2002, 31, 89-93.	9.4	269
53	High-Throughput Variation Detection and Genotyping Using Microarrays. Genome Research, 2001, 11, 1913-1925.	2.4	258
54	Genome-wide association study and mouse model identify interaction between RET and EDNRB pathways in Hirschsprung disease. Nature Genetics, 2002, 32, 237-244.	9.4	255

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55	Human embryonic stem cells have a unique epigenetic signature. Genome Research, 2006, 16, 1075-1083.	2.4	250
56	Parental Origin and Phenotype of Triploidy in Spontaneous Abortions: Predominance of Diandry and Association with the Partial Hydatidiform Mole. American Journal of Human Genetics, 2000, 66, 1807-1820.	2.6	246
57	Identity-by-descent and association mapping of a recessive gene for Hirschsprung disease on human chromosome 13q22. Human Molecular Genetics, 1994, 3, 1217-1225.	1.4	241
58	Differential Contributions of Rare and Common, Coding and Noncoding Ret Mutations to Multifactorial Hirschsprung Disease Liability. American Journal of Human Genetics, 2010, 87, 60-74.	2.6	230
59	Identifying disease alleles by genome sharing. Nature Genetics, 1999, 23, 25-25.	9.4	228
60	A Bivariate Genome-Wide Approach to Metabolic Syndrome. Diabetes, 2011, 60, 1329-1339.	0.3	226
61	Joint Analysis of Psychiatric Disorders Increases Accuracy of Risk Prediction for Schizophrenia, Bipolar Disorder, and Major Depressive Disorder. American Journal of Human Genetics, 2015, 96, 283-294.	2.6	225
62	Meta-analysis identifies common and rare variants influencing blood pressure and overlapping with metabolic trait loci. Nature Genetics, 2016, 48, 1162-1170.	9.4	223
63	The Human MitoChip: A High-Throughput Sequencing Microarray for Mitochondrial Mutation Detection. Genome Research, 2004, 14, 812-819.	2.4	218
64	Differential Susceptibility to Hypertension Is Due to Selection during the Out-of-Africa Expansion. PLoS Genetics, 2005, 1, e82.	1.5	208
65	The Genome Project-Write. Science, 2016, 353, 126-127.	6.0	194
66	Genome-wide Association Analysis of Blood-Pressure Traits in African-Ancestry Individuals Reveals Common Associated Genes in African and Non-African Populations. American Journal of Human Genetics, 2013, 93, 545-554.	2.6	189
67	Genome-Wide Association Studies of Serum Magnesium, Potassium, and Sodium Concentrations Identify Six Loci Influencing Serum Magnesium Levels. PLoS Genetics, 2010, 6, e1001045.	1.5	185
68	Association of Hypertension Drug Target Genes With Blood Pressure and Hypertension in 86 588 Individuals. Hypertension, 2011, 57, 903-910.	1.3	181
69	Haplotype Inference in Random Population Samples. American Journal of Human Genetics, 2002, 71, 1129-1137.	2.6	176
70	Multiplex PCR of three dinucleotide repeats in the Prader-Willi/Angelman critical region (15q11â€“q13): molecular diagnosis and mechanism of uniparental disomy. Human Molecular Genetics, 1993, 2, 143-151.	1.4	174
71	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	13.7	173
72	Association of genetic variation with systolic and diastolic blood pressure among African Americans: the Candidate Gene Association Resource study. Human Molecular Genetics, 2011, 20, 2273-2284.	1.4	168

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73	Genetic Variations in Nitric Oxide Synthase 1 Adaptor Protein Are Associated With Sudden Cardiac Death in US White Community-Based Populations. Circulation, 2009, 119, 940-951.	1.6	167
74	. . .to a future of genetic medicine. Nature, 2001, 409, 822-823.	13.7	164
75	Polymorphisms in the NOS1APGene Modulate QT Interval Duration and Risk of Arrhythmias in the Long QT Syndrome. Journal of the American College of Cardiology, 2010, 55, 2745-2752.	1.2	163
76	Gene-centric Meta-analysis in 87,736 Individuals of European Ancestry Identifies Multiple Blood-Pressure-Related Loci. American Journal of Human Genetics, 2014, 94, 349-360.	2.6	158
77	Undetected Genotyping Errors Cause Apparent Overtransmission of Common Alleles in the Transmission/Disequilibrium Test. American Journal of Human Genetics, 2003, 72, 598-610.	2.6	157
78	A gene for Hirschsprung disease (megacolon) in the pericentromeric region of human chromosome 10. Nature Genetics, 1993, 4, 351-356.	9.4	154
79	Exhaustive allelic transmission disequilibrium tests as a new approach to genome-wide association studies. Nature Genetics, 2004, 36, 1181-1188.	9.4	154
80	Global implementation of genomic medicine: We are not alone. Science Translational Medicine, 2015, 7, 290ps13.	5.8	146
81	Loss of Î´-catenin function in severe autism. Nature, 2015, 520, 51-56.	13.7	145
82	Loci influencing blood pressure identified using a cardiovascular gene-centric array. Human Molecular Genetics, 2013, 22, 1663-1678.	1.4	141
83	It's raining SNPs, hallelujah?. Nature Genetics, 1998, 19, 216-217.	9.4	139
84	Phenotype variation in two-locus mouse models of Hirschsprung disease: Tissue-specific interaction between Ret and Ednrb. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 1826-1831.	3.3	133
85	Molecular Genetic Anatomy and Risk Profile of Hirschsprungâ€™s Disease. New England Journal of Medicine, 2019, 380, 1421-1432.	13.9	131
86	Nature, nurture and human disease. Nature, 2003, 421, 412-414.	13.7	129
87	A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure. American Journal of Human Genetics, 2018, 102, 375-400.	2.6	123
88	Allele Frequency Distributions in Pooled DNA Samples: Applications to Mapping Complex Diseaseâ€‰Genes. Genome Research, 1998, 8, 111-123.	2.4	120
89	Contribution of rare and common variants determine complex diseasesâ€”Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	0.9	119
90	Functional Loss of Semaphorin 3C and/or Semaphorin 3D and Their Epistatic Interaction with Ret Are Critical to Hirschsprung Disease Liability. American Journal of Human Genetics, 2015, 96, 581-596.	2.6	118

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91	The road ahead in genetics and genomics. Nature Reviews Genetics, 2020, 21, 581-596.	7.7	118
92	Identification of a Sudden Cardiac Death Susceptibility Locus at 2q24.2 through Genome-Wide Association in European Ancestry Individuals. PLoS Genetics, 2011, 7, e1002158.	1.5	117
93	Future of genetics of mood disorders research. Biological Psychiatry, 2002, 52, 457-477.	0.7	116
94	Associations Between Hypertension and Genes in the Renin-Angiotensin System. Hypertension, 2003, 41, 1027-1034.	1.3	116
95	Sequence variations in the public human genome data reflect a bottlenecked population history. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 376-381.	3.3	113
96	52 Genetic Loci Influencing MyocardialÂMass. Journal of the American College of Cardiology, 2016, 68, 1435-1448.	1.2	113
97	Trans-Ethnic Fine-Mapping of Lipid Loci Identifies Population-Specific Signals and Allelic Heterogeneity That Increases the Trait Variance Explained. PLoS Genetics, 2013, 9, e1003379.	1.5	112
98	Enhancer Variants Synergistically Drive Dysfunction of a Gene Regulatory Network In Hirschsprung Disease. Cell, 2016, 167, 355-368.e10.	13.5	112
99	Gene-Age Interactions in Blood Pressure Regulation: A Large-Scale Investigation with the CHARGE, Global BPgen, and ICBP Consortia. American Journal of Human Genetics, 2014, 95, 24-38.	2.6	109
100	Drug-Sensitized Zebrafish Screen Identifies Multiple Genes, Including GINS3 , as Regulators of Myocardial Repolarization. Circulation, 2009, 120, 553-559.	1.6	106
101	Multiple Loci Are Associated with White Blood Cell Phenotypes. PLoS Genetics, 2011, 7, e1002113.	1.5	106
102	Associations of Mitochondrial and Nuclear Mitochondrial Variants and Genes with Seven Metabolic Traits. American Journal of Human Genetics, 2019, 104, 112-138.	2.6	106
103	Multiple Genes for Essential-Hypertension Susceptibility on Chromosome 1q. American Journal of Human Genetics, 2007, 80, 253-264.	2.6	102
104	Pleiotropic Skeletal and Ocular Phenotypes of the Mouse Mutation Congenital Hydrocephalus (ch/Mf1) Arise from a Winged Helix/Forkhead Transcription Factor Gene. Human Molecular Genetics, 1999, 8, 625-637.	1.4	100
105	Genetics and Genomics for the Prevention and Treatment of Cardiovascular Disease: Update. Circulation, 2013, 128, 2813-2851.	1.6	100
106	EDNRB/EDN3 and Hirschsprung Disease Type II. Pigment Cell & Melanoma Research, 2001, 14, 161-169.	4.0	97
107	Two-Locus models of disease. Genetic Epidemiology, 1992, 9, 347-365.	0.6	96
108	Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries. PLoS ONE, 2018, 13, e0198166.	1.1	94

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109	Drift variances of FSTand GST statistics obtained from a finite number of isolated populations. Theoretical Population Biology, 1977, 11, 307-325.	0.5	93
110	An integrated genetic linkage map of the laboratory rat. Mammalian Genome, 1998, 9, 521-530.	1.0	92
111	Endothelinâ€“3 frameshift mutation in congenital central hypoventilation syndrome. Nature Genetics, 1996, 13, 395-396.	9.4	89
112	Mitochondrial DNA Variants of Respiratory Complex I that Uniquely Characterize Haplogroup T2 Are Associated with Increased Risk of Age-Related Macular Degeneration. PLoS ONE, 2009, 4, e5508.	1.1	89
113	Mean and variance of FST in a finite number of incompletely isolated populations. Theoretical Population Biology, 1977, 11, 291-306.	0.5	88
114	Genomics in Sudden Cardiac Death. Circulation Research, 2004, 94, 712-723.	2.0	88
115	Single-trait and multi-trait genome-wide association analyses identify novel loci for blood pressure in African-ancestry populations. PLoS Genetics, 2017, 13, e1006728.	1.5	88
116	Positional Identification of Hypertension Susceptibility Genes on Chromosome 2. Hypertension, 2004, 43, 477-482.	1.3	85
117	Linkage Disequilibrium Analysis of Biallelic DNA Markers, Human Quantitative Trait Loci, and Threshold-Defined Case and Control Subjects. American Journal of Human Genetics, 2000, 67, 1208-1218.	2.6	84
118	A genetic linkage map of 17 markers on human chromosome 21. Genomics, 1989, 4, 579-591.	1.3	82
119	A genetic linkage map of 27 markers on human chromosome 21. Genomics, 1991, 9, 407-419.	1.3	82
120	On consanguineous marriages and the genetic load. Human Genetics, 1977, 36, 47-54.	1.8	81
121	Novel Approach Identifies SNPs in SLC2A10 and KCNK9 with Evidence for Parent-of-Origin Effect on Body Mass Index. PLoS Genetics, 2014, 10, e1004508.	1.5	80
122	PATTERNS OFGENETICVARIATION INMENDELIAN ANDCOMPLEXTRAITS. Annual Review of Genomics and Human Genetics, 2000, 1, 387-407.	2.5	78
123	Identifying Allelic Loss and Homozygous Deletions in Pancreatic Cancer without Matched Normals Using High-Density Single-Nucleotide Polymorphism Arrays. Cancer Research, 2006, 66, 7920-7928.	0.4	78
124	Combined admixture mapping and association analysis identifies a novel blood pressure genetic locus on 5p13: contributions from the CARe consortium. Human Molecular Genetics, 2011, 20, 2285-2295.	1.4	77
125	Mendelian disorders and multifactorial traits: the big divide or one for all?. Nature Reviews Genetics, 2010, 11, 380-384.	7.7	76
126	Evaluation of the RET regulatory landscape reveals the biological relevance of a HSCR-implicated enhancer. Human Molecular Genetics, 2005, 14, 3837-3845.	1.4	75

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127	A Linkage Map of Human Chromosome 21: 43 PCR Markers at Average Intervals of 2.5 cM. Genomics, 1993, 16, 562-571.	1.3	73
128	Effects of Long-Term Averaging of Quantitative Blood Pressure Traits on the Detection of Genetic Associations. American Journal of Human Genetics, 2014, 95, 49-65.	2.6	73
129	Safety issues in cell-based intervention trials. Fertility and Sterility, 2003, 80, 1077-1085.	0.5	72
130	An Enhancer Polymorphism at the Cardiomyocyte Intercalated Disc Protein NOS1AP Locus Is a Major Regulator of the QT Interval. American Journal of Human Genetics, 2014, 94, 854-869.	2.6	72
131	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	3.8	72
132	Linkage Disequilibrium and Haplotype Diversity in the Genes of the Renin-Angiotensin System: Findings From the Family Blood Pressure Program. Genome Research, 2003, 13, 173-181.	2.4	71
133	Associations between Genetic Variants in the NOS1AP (CAPON) Gene and Cardiac Repolarization in the Old Order Amish. Human Heredity, 2007, 64, 214-219.	0.4	71
134	Down syndrome consequent to a cryptic maternal 12p;21q chromosome translocation. American Journal of Medical Genetics Part A, 1995, 56, 67-71.	2.4	70
135	Understanding cardiovascular disease through the lens of genome-wide association studies. Trends in Genetics, 2009, 25, 387-394.	2.9	68
136	DNA polymorphism haplotypes of the human apolipoprotein APOA1-APOC3-APOA4 gene cluster. Human Genetics, 1988, 80, 265-273.	1.8	67
137	A graphical representation of genetic and physical maps: The Marey map. Genomics, 1991, 11, 219-222.	1.3	67
138	Distilling Pathophysiology from Complex Disease Genetics. Cell, 2013, 155, 21-26.	13.5	67
139	Phylogeny of human Î²-globin haplotypes and its implications for recent human evolution. American Journal of Physical Anthropology, 1990, 81, 113-130.	2.1	66
140	Population variation in total genetic risk of Hirschsprung disease from common RET, SEMA3 and NRG1 susceptibility polymorphisms. Human Molecular Genetics, 2015, 24, 2997-3003.	1.4	66
141	From The Cover: The gene for soluble N-ethylmaleimide sensitive factor attachment protein Â is mutated in hydrocephaly with hop gait (hyh) mice. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 1748-1753.	3.3	65
142	Replication of the Wellcome Trust genome-wide association study of essential hypertension: the Family Blood Pressure Program. European Journal of Human Genetics, 2008, 16, 1507-1511.	1.4	64
143	Next-Generation Sequencing of Human Mitochondrial Reference Genomes Uncovers High Heteroplasmy Frequency. PLoS Computational Biology, 2012, 8, e1002737.	1.5	61
144	Linkage analysis of the human HMG14 gene on chromosome 21 using a GT dinucleotide repeat as polymorphic marker. Genomics, 1990, 7, 136-138.	1.3	60

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145	A Genome-Wide Scan for Obesity in African-Americans. Diabetes, 2002, 51, 541-544.	0.3	60
146	A Genome-Wide Linkage Analysis Investigating the Determinants of Blood Pressure in Whites and African Americans. American Journal of Hypertension, 2003, 16, 151-153.	1.0	60
147	A comprehensive evaluation of the genetic architecture of sudden cardiac arrest. European Heart Journal, 2018, 39, 3961-3969.	1.0	59
148	Cloning of a Novel Homeobox-Containing Gene,PKNOX1,and Mapping to Human Chromosome 21q22.3. Genomics, 1997, 41, 193-200.	1.3	58
149	HumanGFRA1: Cloning, Mapping, Genomic Structure, and Evaluation as a Candidate Gene for Hirschsprung Disease Susceptibility. Genomics, 1998, 48, 354-362.	1.3	58
150	Positional identification of variants of Adamts16 linked to inherited hypertension. Human Molecular Genetics, 2009, 18, 2825-2838.	1.4	57
151	Interaction between a chromosome 10RETenhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. Human Mutation, 2009, 30, 771-775.	1.1	57
152	Genetic variation associated with circulating monocyte count in the eMERGE Network. Human Molecular Genetics, 2013, 22, 2119-2127.	1.4	56
153	Copy Number Variants in Candidate Genes Are Genetic Modifiers of Hirschsprung Disease. PLoS ONE, 2011, 6, e21219.	1.1	56
154	Microsatellite Polymorphism Linkage Map of Human Chromosome 13q. Genomics, 1993, 15, 376-386.	1.3	55
155	Elevated Frequency and Allelic Heterogeneity of Congenital Nephrotic Syndrome, Finnish Type, in the Old Order Mennonites. American Journal of Human Genetics, 1999, 65, 1785-1790.	2.6	55
156	Population Bottlenecks as a Potential Major Shaping Force of Human Genome Architecture. PLoS Genetics, 2007, 3, e119.	1.5	55
157	Genome-Wide Association Study Identifies GPC5 as a Novel Genetic Locus Protective against Sudden Cardiac Arrest. PLoS ONE, 2010, 5, e9879.	1.1	54
158	Follow-up of a major linkage peak on chromosome 1 reveals suggestive QTLs associated with essential hypertension: GenNet study. European Journal of Human Genetics, 2009, 17, 1650-1657.	1.4	52
159	Waardenburg syndrome and Hirschsprung disease: Evidence for pleiotropic effects of a single dominant gene. American Journal of Medical Genetics Part A, 1990, 35, 100-104.	2.4	48
160	Cytogenetics and origins of pediatric germ cell tumors. Cancer Genetics and Cytogenetics, 1994, 74, 54-58.	1.0	48
161	A population-based study of KCNH7 p.Arg394His and bipolar spectrum disorder. Human Molecular Genetics, 2014, 23, 6395-6406.	1.4	48
162	Methods for studying recombination on chromosomes that undergo nondisjunction. Genomics, 1987, 1, 35-42.	1.3	47

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163	Patterns of Meiotic Recombination on the Long Arm of Human Chromosome 21. Genome Research, 2000, 10, 1319-1332.	2.4	47
164	Revealing rateâ€limiting steps in complex disease biology: The crucial importance of studying rare, extremeâ€phenotype families. BioEssays, 2016, 38, 578-586.	1.2	47
165	Direct Estimates of the Genomic Contributions to Blood Pressure Heritability within a Population-Based Cohort (ARIC). PLoS ONE, 2015, 10, e0133031.	1.1	47
166	Mining gold dust under the genome wide significance level: a twoâ€stage approach to analysis of GWAS. Genetic Epidemiology, 2011, 35, 111-118.	0.6	46
167	Elementary methods for the analysis of dichotomous outcomes in unselected samples of Twins. Genetic Epidemiology, 1992, 9, 273-287.	0.6	45
168	Polymorphisms in the Mitochondrial DNA Control Region and Frailty in Older Adults. PLoS ONE, 2010, 5, e11069.	1.1	44
169	Rare Exome Sequence Variants in CLCN6 Reduce Blood Pressure Levels and Hypertension Risk. Circulation: Cardiovascular Genetics, 2016, 9, 64-70.	5.1	44
170	Genetics and biology of human ovarian teratomas. Cancer Genetics and Cytogenetics, 1992, 62, 58-65.	1.0	43
171	A multilevel model to address batch effects in copy number estimation using SNP arrays. Biostatistics, 2011, 12, 33-50.	0.9	43
172	Fragile X founder effect?. Nature Genetics, 1992, 1, 237-238.	9.4	42
173	Haplotype and Missing Data Inference in Nuclear Families. Genome Research, 2004, 14, 1624-1632.	2.4	42
174	Allele-specific expression in the germline of patients with familial pancreatic cancer: An unbiased approach to cancer gene discovery. Cancer Biology and Therapy, 2008, 7, 135-144.	1.5	42
175	Transcriptional regulation of PRPF31 gene expression by MSR1 repeat elements causes incomplete penetrance in retinitis pigmentosa. Scientific Reports, 2016, 6, 19450.	1.6	42
176	Hemostasis, Inflammation, and Fatal and Nonfatal Coronary Heart Disease. Arteriosclerosis, Thrombosis, and Vascular Biology, 2009, 29, 2182-2190.	1.1	41
177	Proteins linked to autosomal dominant and autosomal recessive disorders harbor characteristic rare missense mutation distribution patterns. Human Molecular Genetics, 2015, 24, 5995-6002.	1.4	40
178	Genomics Is Not Enough. Science, 2011, 334, 15-15.	6.0	39
179	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	1.4	38
180	MicroRNAs in the miR-17 and miR-15 families are downregulated in chronic kidney disease with hypertension. PLoS ONE, 2017, 12, e0176734.	1.1	38

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