Aravinda Chakravarti

List of Publications by Year in Descending Order

304 papers	61,039 citations	92 h-index	246 g-index
330 ext. papers	72,375 ext. citations	13.4 avg, IF	6.58 L-index

#	Paper	IF	Citations
304	Rare coding variants in RCN3 are associated with blood pressure.. *BMC Genomics*, **2022**, 23, 148	4.5	
303	A multi-enhancer regulatory code is disrupted in Hirschsprung disease. *Genome Research*, **2021**,	9.7	2
302	Pathogenic alleles in microtubule, secretory granule and extracellular matrix-related genes in familial keratoconus. *Human Molecular Genetics*, **2021**, 30, 658-671	5.6	1
301	Multiple, independent, common variants at RET, SEMA3 and NRG1 gut enhancers specify Hirschsprung disease risk in European ancestry subjects. *Journal of Pediatric Surgery*, **2021**, 56, 2286-2294	2.6	1
300	Sequence-based correction of barcode bias in massively parallel reporter assays. *Genome Research*, **2021**, 31, 1638-1645	9.7	0
299	Magnitude of Mendelian versus complex inheritance of rare disorders. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3287-3293	2.5	2
298	Analysis of putative cis-regulatory elements regulating blood pressure variation. *Human Molecular Genetics*, **2020**, 29, 1922-1932	5.6	0
297	MicroRNA-4516-mediated regulation of relies on 3RUTR -acting variants and contributes to the altered risk of Hirschsprung disease. *Journal of Medical Genetics*, **2020**, 57, 634-642	5.8	2
296	The road ahead in genetics and genomics. *Nature Reviews Genetics*, **2020**, 21, 581-596	30.1	43
295	Leveraging linkage evidence to identify low-frequency and rare variants on 16p13 associated with blood pressure using TOPMed whole genome sequencing data. *Human Genetics*, **2019**, 138, 199-210	6.3	14
294	Multiple variant enhancers modulate its cardiac gene expression and the QT interval. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 10636-10645	11.5	13
293	Molecular Genetic Anatomy and Risk Profile of HirschsprungR Disease. *New England Journal of Medicine*, **2019**, 380, 1421-1432	59.2	71
292	A multi-ancestry genome-wide study incorporating gene-smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. *Human Molecular Genetics*, **2019**, 28, 2615-2633	5.6	14
291	A gene regulatory network explains RET-EDNRB epistasis in Hirschsprung disease. *Human Molecular Genetics*, **2019**, 28, 3137-3147	5.6	8
290	Sequence characterization of RET in 117 Chinese Hirschsprung disease families identifies a large burden of de novo and parental mosaic mutations. *Orphanet Journal of Rare Diseases*, **2019**, 14, 237	4.2	5
289	High Levels of Interest in Reproductive Genetic Information in Parents of Children and Adults With Hirschsprung Disease. *Journal of Pediatric Gastroenterology and Nutrition*, **2019**, 69, 299-305	2.8	0
288	Gene- and tissue-level interactions in normal gastrointestinal development and Hirschsprung disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**,	11.5	5

287	Associations of Mitochondrial and Nuclear Mitochondrial Variants and Genes with Seven Metabolic Traits. *American Journal of Human Genetics*, **2019**, 104, 112-138	11	54
286	Combined linkage and association analysis identifies rare and low frequency variants for blood pressure at 1q31. *European Journal of Human Genetics*, **2019**, 27, 269-277	5.3	3
285	Cardiomyocytes have mosaic patterns of protein expression. *Cardiovascular Pathology*, **2018**, 34, 50-57	3.8	9
284	Response to Brosens et al. *Genetics in Medicine*, **2018**, 20, 1479-1480	8.1	
283	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	11	59
282	Genome-wide association study of Hirschsprung disease detects a novel low-frequency variant at the RET locus. *European Journal of Human Genetics*, **2018**, 26, 561-569	5.3	11
281	The genetic underpinnings of variation in ages at menarche and natural menopause among women from the multi-ethnic Population Architecture using Genomics and Epidemiology (PAGE) Study: A trans-ethnic meta-analysis. *PLoS ONE*, **2018**, 13, e0200486	3.7	14
280	Human cardiac -regulatory elements, their cognate transcription factors, and regulatory DNA sequence variants. *Genome Research*, **2018**, 28, 1577-1588	9.7	11
279	Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries. *PLoS ONE*, **2018**, 13, e0198166	3.7	31
278	RET somatic mutations are underrecognized in Hirschsprung disease. *Genetics in Medicine*, **2018**, 20, 770-777	8.1	20
277	A comprehensive evaluation of the genetic architecture of sudden cardiac arrest. *European Heart Journal*, **2018**, 39, 3961-3969	9.5	31
276	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. *Nature Genetics*, **2018**, 50, 1412-1425	36.3	386
275	Newton E. Morton (1929-2018). *American Journal of Human Genetics*, **2018**, 102, 1011-1017	11	
274	Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. *Nature Genetics*, **2017**, 49, 403-415	36.3	313
273	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. *Genome Biology*, **2017**, 18, 48	18.3	55
272	Testing the Ret and Sema3d genetic interaction in mouse enteric nervous system development. *Human Molecular Genetics*, **2017**, 26, 1811-1820	5.6	3
271	Genome-wide association analyses using electronic health records identify new loci influencing blood pressure variation. *Nature Genetics*, **2017**, 49, 54-64	36.3	157
270	Gene-by-Psychosocial Factor Interactions Influence Diastolic Blood Pressure in European and African Ancestry Populations: Meta-Analysis of Four Cohort Studies. *International Journal of Environmental Research and Public Health*, **2017**, 14,	4.6	3

269	Rare variants in fox-1 homolog A (RBFOX1) are associated with lower blood pressure. *PLoS Genetics* , **2017**, 13, e1006678	6	11
268	Single-trait and multi-trait genome-wide association analyses identify novel loci for blood pressure in African-ancestry populations. *PLoS Genetics*, **2017**, 13, e1006728	6	58
267	MicroRNAs in the miR-17 and miR-15 families are downregulated in chronic kidney disease with hypertension. *PLoS ONE*, **2017**, 12, e0176734	3.7	27
266	52 Genetic Loci Influencing Myocardial Mass. *Journal of the American College of Cardiology*, **2016**, 68, 1435-1448	15.1	76
265	Meta-analysis identifies common and rare variants influencing blood pressure and overlapping with metabolic trait loci. *Nature Genetics*, **2016**, 48, 1162-70	36.3	152
264	The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. *Nature Genetics*, **2016**, 48, 1171-1184	36.3	251
263	Transcriptional regulation of PRPF31 gene expression by MSR1 repeat elements causes incomplete penetrance in retinitis pigmentosa. *Scientific Reports*, **2016**, 6, 19450	4.9	31
262	Rare coding TTN variants are associated with electrocardiographic QT interval in the general population. *Scientific Reports*, **2016**, 6, 28356	4.9	5
261	GENOME ENGINEERING. The Genome Project-Write. *Science*, **2016**, 353, 126-7	33.3	138
260	Rare Exome Sequence Variants in CLCN6 Reduce Blood Pressure Levels and Hypertension Risk. *Circulation: Cardiovascular Genetics*, **2016**, 9, 64-70		35
259	Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. *Science*, **2016**, 351, 1166-71	33.3	325
258	Variant Discovery and Fine Mapping of Genetic Loci Associated with Blood Pressure Traits in Hispanics and African Americans. *PLoS ONE*, **2016**, 11, e0164132	3.7	19
257	Revealing rate-limiting steps in complex disease biology: The crucial importance of studying rare, extreme-phenotype families. *BioEssays*, **2016**, 38, 578-86	4.1	32
256	Commentary: The central questions of human genetics: Richard Lewontin's 1968 senior lecture in Victor McKusick's Bar Harbor short course. *International Journal of Epidemiology*, **2016**, 45, 668-72	7.8	1
255	Enhancer Variants Synergistically Drive Dysfunction of a Gene Regulatory Network In Hirschsprung Disease. *Cell*, **2016**, 167, 355-368.e10	56.2	80
254	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. *Human Molecular Genetics*, **2016**, 25, 5265-5275	5.6	23
253	A PIGN mutation responsible for multiple congenital anomalies-hypotonia-seizures syndrome 1 (MCAHS1) in an Israeli-Arab family. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 176-82	2.5	22
252	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	30.4	119

251	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics*, **2015**, 97, 199-215	11	432
250	Loss of β-catenin function in severe autism. *Nature*, **2015**, 520, 51-6	50.4	97
249	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
248	Proteins linked to autosomal dominant and autosomal recessive disorders harbor characteristic rare missense mutation distribution patterns. *Human Molecular Genetics*, **2015**, 24, 5995-6002	5.6	24
247	Perspectives on Human Variation through the Lens of Diversity and Race. *Cold Spring Harbor Perspectives in Biology*, **2015**, 7, a023358	10.2	11
246	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	6	220
245	Population variation in total genetic risk of Hirschsprung disease from common RET, SEMA3 and NRG1 susceptibility polymorphisms. *Human Molecular Genetics*, **2015**, 24, 2997-3003	5.6	49
244	Global implementation of genomic medicine: We are not alone. *Science Translational Medicine*, **2015**, 7, 290ps13	17.5	112
243	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, 581-96	11	82
242	Intestinal Neuronal Dysplasia-Like Submucosal Ganglion Cell Hyperplasia at the Proximal Margins of Hirschsprung Disease Resections. *Pediatric and Developmental Pathology*, **2015**, 18, 466-76	2.2	18
241	The role of rare variants in systolic blood pressure: analysis of ExomeChip data in HyperGEN African Americans. *Human Heredity*, **2015**, 79, 20-7	1.1	5
240	New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196	50.4	920
239	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
238	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, 283-94	11	161
237	Direct Estimates of the Genomic Contributions to Blood Pressure Heritability within a Population-Based Cohort (ARIC). *PLoS ONE*, **2015**, 10, e0133031	3.7	32
236	HPASubC: A suite of tools for user subclassification of human protein atlas tissue images. *Journal of Pathology Informatics*, **2015**, 6, 36	4.4	9
235	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	11	80
234	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	11	52

233	Defining the role of common variation in the genomic and biological architecture of adult human height. *Nature Genetics*, **2014**, 46, 1173-86	36.3	1339
232	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. *Nature Genetics*, **2014**, 46, 826-36	36.3	199
231	2013 William Allan Award: My multifactorial journey. *American Journal of Human Genetics*, **2014**, 94, 326-33	11	2
230	Common variation in fatty acid metabolic genes and risk of incident sudden cardiac arrest. *Heart Rhythm*, **2014**, 11, 471-7	6.7	12
229	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, 854-69	11	56
228	Novel approach identifies SNPs in SLC2A10 and KCNK9 with evidence for parent-of-origin effect on body mass index. *PLoS Genetics*, **2014**, 10, e1004508	6	45
227	Effects of RET and NRG1 polymorphisms in Indonesian patients with Hirschsprung disease. *Journal of Pediatric Surgery*, **2014**, 49, 1614-8	2.6	33
226	Profile of Mary-Claire King, 2014 Lasker-Koshland Special Achievement in Medical Science awardee. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 17690-2	11.5	
225	A population-based study of KCNH7 p.Arg394His and bipolar spectrum disorder. *Human Molecular Genetics*, **2014**, 23, 6395-406	5.6	30
224	Gene-centric meta-analysis in 87,736 individuals of European ancestry identifies multiple blood-pressure-related loci. *American Journal of Human Genetics*, **2014**, 94, 349-60	11	131
223	Linkage analysis incorporating gene-age interactions identifies seven novel lipid loci: the Family Blood Pressure Program. *Atherosclerosis*, **2014**, 235, 84-93	3.1	9
222	Sequence analysis of six blood pressure candidate regions in 4,178 individuals: the Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) targeted sequencing study. *PLoS ONE*, **2014**, 9, e109155	3.7	15
221	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, 545-54	11	145
220	Discovery and refinement of loci associated with lipid levels. *Nature Genetics*, **2013**, 45, 1274-1283	36.3	1904
219	Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nature Genetics*, **2013**, 45, 1345-52	36.3	597
218	Distilling pathophysiology from complex disease genetics. *Cell*, **2013**, 155, 21-6	56.2	63
217	Contribution of rare and common variants determine complex diseases-Hirschsprung disease as a model. *Developmental Biology*, **2013**, 382, 320-9	3.1	90
216	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628

215	Loci influencing blood pressure identified using a cardiovascular gene-centric array. *Human Molecular Genetics*, **2013**, 22, 1663-78	5.6	119
214	Genetics and genomics for the prevention and treatment of cardiovascular disease: update: a scientific statement from the American Heart Association. *Circulation*, **2013**, 128, 2813-51	16.7	76
213	Associations between NOS1AP single nucleotide polymorphisms (SNPs) and QT interval duration in four racial/ethnic groups in the Multi-Ethnic Study of Atherosclerosis (MESA). *Annals of Noninvasive Electrocardiology*, **2013**, 18, 29-40	1.5	9
212	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	6	94
211	Effects of rare and common blood pressure gene variants on essential hypertension: results from the Family Blood Pressure Program, CLUE, and Atherosclerosis Risk in Communities studies. *Circulation Research*, **2013**, 112, 318-26	15.7	20
210	Genetic variation associated with circulating monocyte count in the eMERGE Network. *Human Molecular Genetics*, **2013**, 22, 2119-27	5.6	46
209	Chromosome 21 scan in Down syndrome reveals DSCAM as a predisposing locus in Hirschsprung disease. *PLoS ONE*, **2013**, 8, e62519	3.7	18
208	A polymorphic 3′UTR element in ATP1B1 regulates alternative polyadenylation and is associated with blood pressure. *PLoS ONE*, **2013**, 8, e76290	3.7	12
207	Defining the contribution of CNTNAP2 to autism susceptibility. *PLoS ONE*, **2013**, 8, e77906	3.7	25
206	Rapid and efficient human mutation detection using a bench-top next-generation DNA sequencer. *Human Mutation*, **2012**, 33, 281-9	4.7	31
205	Meta-analysis identifies six new susceptibility loci for atrial fibrillation. *Nature Genetics*, **2012**, 44, 670-5	36.3	429
204	Genetics. Mendelian puzzles. *Science*, **2012**, 335, 930-1	33.3	16
203	2011 introduction to Curt Stern Award. *American Journal of Human Genetics*, **2012**, 90, 405-6	11	
202	Next-generation sequencing of human mitochondrial reference genomes uncovers high heteroplasmy frequency. *PLoS Computational Biology*, **2012**, 8, e1002737	5	54
201	Male and female differential reproductive rate could explain parental transmission asymmetry of mutation origin in Hirschsprung disease. *European Journal of Human Genetics*, **2012**, 20, 917-20	5.3	7
200	Quantifying and modeling birth order effects in autism. *PLoS ONE*, **2011**, 6, e26418	3.7	16
199	SNPs and other features as they predispose to complex disease: genome-wide predictive analysis of a quantitative phenotype for hypertension. *PLoS ONE*, **2011**, 6, e27891	3.7	3
198	Mining gold dust under the genome wide significance level: a two-stage approach to analysis of GWAS. *Genetic Epidemiology*, **2011**, 35, 111-8	2.6	41

197	A multilevel model to address batch effects in copy number estimation using SNP arrays. *Biostatistics*, **2011**, 12, 33-50	3.7	38
196	Association of hypertension drug target genes with blood pressure and hypertension in 86,588 individuals. *Hypertension*, **2011**, 57, 903-10	8.5	154
195	Widespread promiscuous genetic information transfer from DNA to RNA. *Circulation Research*, **2011**, 109, 1202-3	15.7	4
194	A bivariate genome-wide approach to metabolic syndrome: STAMPEED consortium. *Diabetes*, **2011**, 60, 1329-39	0.9	194
193	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, 2285-95	5.6	70
192	Genomic contributions to Mendelian disease. *Genome Research*, **2011**, 21, 643-4	9.7	14
191	Association of genetic variation with systolic and diastolic blood pressure among African Americans: the Candidate Gene Association Resource study. *Human Molecular Genetics*, **2011**, 20, 2273-84	5.6	146
190	Five blood pressure loci identified by an updated genome-wide linkage scan: meta-analysis of the Family Blood Pressure Program. *American Journal of Hypertension*, **2011**, 24, 347-54	2.3	15
189	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
188	Multiple loci are associated with white blood cell phenotypes. *PLoS Genetics*, **2011**, 7, e1002113	6	92
187	Identification of a sudden cardiac death susceptibility locus at 2q24.2 through genome-wide association in European ancestry individuals. *PLoS Genetics*, **2011**, 7, e1002158	6	95
186	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	6	249
185	Copy number variants in candidate genes are genetic modifiers of Hirschsprung disease. *PLoS ONE*, **2011**, 6, e21219	3.7	53
184	Genome-wide association study of PR interval. *Nature Genetics*, **2010**, 42, 153-9	36.3	340
183	Common variants in KCNN3 are associated with lone atrial fibrillation. *Nature Genetics*, **2010**, 42, 240-4	36.3	362
182	Common variants in 22 loci are associated with QRS duration and cardiac ventricular conduction. *Nature Genetics*, **2010**, 42, 1068-76	36.3	249
181	Mendelian disorders and multifactorial traits: the big divide or one for all?. *Nature Reviews Genetics*, **2010**, 11, 380-4	30.1	60
180	Genome-wide association study identifies GPC5 as a novel genetic locus protective against sudden cardiac arrest. *PLoS ONE*, **2010**, 5, e9879	3.7	48

179	Parent-of-origin effects in autism identified through genome-wide linkage analysis of 16,000 SNPs. *PLoS ONE*, **2010**, 5, e12513	3.7	28
178	Polymorphisms in the mitochondrial DNA control region and frailty in older adults. *PLoS ONE*, **2010**, 5, e11069	3.7	37
177	Genome-wide association studies of serum magnesium, potassium, and sodium concentrations identify six Loci influencing serum magnesium levels. *PLoS Genetics*, **2010**, 6, e1001045	6	144
176	Polymorphisms in the NOS1AP gene modulate QT interval duration and risk of arrhythmias in the long QT syndrome. *Journal of the American College of Cardiology*, **2010**, 55, 2745-52	15.1	143
175	2008 Presidential Address: Principia Genetica: Our Future Science. *American Journal of Human Genetics*, **2010**, 86, 302-308	11	2
174	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	11	198
173	Multiple independent genetic factors at NOS1AP modulate the QT interval in a multi-ethnic population. *PLoS ONE*, **2009**, 4, e4333	3.7	25
172	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	3.7	74
171	Genetic variations in nitric oxide synthase 1 adaptor protein are associated with sudden cardiac death in US white community-based populations. *Circulation*, **2009**, 119, 940-51	16.7	141
170	Positional identification of variants of Adamts16 linked to inherited hypertension. *Human Molecular Genetics*, **2009**, 18, 2825-38	5.6	52
169	From the Cover: 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, 226-31	11.5	240
168	Hybrids of aneuploid human cancer cells permit complementation of simple and complex cancer defects. *Cancer Biology and Therapy*, **2009**, 8, 347-55	4.6	2
167	The association of cell cycle checkpoint 2 variants and kidney function: findings of the Family Blood Pressure Program and the Atherosclerosis Risk In Communities study. *American Journal of Hypertension*, **2009**, 22, 552-8	2.3	1
166	Hemostasis, inflammation, and fatal and nonfatal coronary heart disease: long-term follow-up of the atherosclerosis risk in communities (ARIC) cohort. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2009**, 29, 2182-90	9.4	37
165	Drug-sensitized zebrafish screen identifies multiple genes, including GINS3, as regulators of myocardial repolarization. *Circulation*, **2009**, 120, 553-9	16.7	84
164	Understanding cardiovascular disease through the lens of genome-wide association studies. *Trends in Genetics*, **2009**, 25, 387-94	8.5	55
163	Interaction between a chromosome 10 RET enhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. *Human Mutation*, **2009**, 30, 771-5	4.7	50
162	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, 1650-7	5.3	50

161	A genome-wide linkage and association scan reveals novel loci for autism. *Nature*, **2009**, 461, 802-8	50.4	474
160	Finding the missing heritability of complex diseases. *Nature*, **2009**, 461, 747-53	50.4	6084
159	Being human: kinship: race relations. *Nature*, **2009**, 457, 380-1	50.4	15
158	Common variants at ten loci modulate the QT interval duration in the QTSCD Study. *Nature Genetics*, **2009**, 41, 407-14	36.3	308
157	Genome-wide association study of blood pressure and hypertension. *Nature Genetics*, **2009**, 41, 677-87	36.3	1065
156	Variants in ZFHX3 are associated with atrial fibrillation in individuals of European ancestry. *Nature Genetics*, **2009**, 41, 879-81	36.3	307
155	Multiple loci influence erythrocyte phenotypes in the CHARGE Consortium. *Nature Genetics*, **2009**, 41, 1191-8	36.3	285
154	Replication of the Wellcome Trust genome-wide association study of essential hypertension: the Family Blood Pressure Program. *European Journal of Human Genetics*, **2008**, 16, 1507-11	5.3	57
153	A common genetic variant in the neurexin superfamily member CNTNAP2 increases familial risk of autism. *American Journal of Human Genetics*, **2008**, 82, 160-4	11	482
152	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, 135-44	4.6	37
151	Estimating genome-wide copy number using allele-specific mixture models. *Journal of Computational Biology*, **2008**, 15, 857-66	1.7	17
150	Association between microdeletion and microduplication at 16p11.2 and autism. *New England Journal of Medicine*, **2008**, 358, 667-75	59.2	1249
149	Genome-wide association scan shows genetic variants in the FTO gene are associated with obesity-related traits. *PLoS Genetics*, **2007**, 3, e115	6	1231
148	Genome-wide detection and characterization of positive selection in human populations. *Nature*, **2007**, 449, 913-8	50.4	1367
147	A second generation human haplotype map of over 3.1 million SNPs. *Nature*, **2007**, 449, 851-61	50.4	3647
146	An investigation of genome-wide associations of hypertension with microsatellite markers in the family blood pressure program (FBPP). *Human Genetics*, **2007**, 121, 577-90	6.3	21
145	Population bottlenecks as a potential major shaping force of human genome architecture. *PLoS Genetics*, **2007**, 3, e119	6	46
144	Associations between genetic variants in the NOS1AP (CAPON) gene and cardiac repolarization in the old order Amish. *Human Heredity*, **2007**, 64, 214-9	1.1	64

143	An ancestral variant of Secretogranin II confers regulation by PHOX2 transcription factors and association with hypertension. *Human Molecular Genetics*, **2007**, 16, 1752-64	5.6	24
142	Multiple genes for essential-hypertension susceptibility on chromosome 1q. *American Journal of Human Genetics*, **2007**, 80, 253-64	11	93
141	Identifying allelic loss and homozygous deletions in pancreatic cancer without matched normals using high-density single-nucleotide polymorphism arrays. *Cancer Research*, **2006**, 66, 7920-8	10.1	74
140	High incidence of deafness from three frequent connexin 26 mutations in an isolated community. *Genetic Testing and Molecular Biomarkers*, **2006**, 10, 40-3		10
139	Human embryonic stem cells have a unique epigenetic signature. *Genome Research*, **2006**, 16, 1075-83	9.7	224
138	Variation in the ciliary neurotrophic factor gene and muscle strength in older Caucasian women. *Journal of the American Geriatrics Society*, **2006**, 54, 823-6	5.6	30
137	A common genetic variant in the NOS1 regulator NOS1AP modulates cardiac repolarization. *Nature Genetics*, **2006**, 38, 644-51	36.3	438
136	The impact of data quality on the identification of complex disease genes: experience from the Family Blood Pressure Program. *European Journal of Human Genetics*, **2006**, 14, 469-77	5.3	18
135	Haplotype association analysis of AGT variants with hypertension-related traits: the HyperGEN study. *Human Heredity*, **2005**, 60, 164-76	1.1	21
134	Differential susceptibility to hypertension is due to selection during the out-of-Africa expansion. *PLoS Genetics*, **2005**, 1, e82	6	175
133	Genomic alterations in cultured human embryonic stem cells. *Nature Genetics*, **2005**, 37, 1099-103	36.3	525
132	A common sex-dependent mutation in a RET enhancer underlies Hirschsprung disease risk. *Nature*, **2005**, 434, 857-63	50.4	377
131	A population association study of angiotensinogen polymorphisms and haplotypes with left ventricular phenotypes. *Hypertension*, **2005**, 46, 1294-9	8.5	12
130	Evaluation of the RET regulatory landscape reveals the biological relevance of a HSCR-implicated enhancer. *Human Molecular Genetics*, **2005**, 14, 3837-45	5.6	67
129	On the probability that a novel variant is a disease-causing mutation. *Genome Research*, **2005**, 15, 960-6	9.7	21
128	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, 15960-4	11.5	365
127	Phenotype-genotype correlation in Hirschsprung disease is illuminated by comparative analysis of the RET protein sequence. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 8949-54	11.5	32
126	Finding needles in haystacks--IRF6 gene variants in isolated cleft lip or cleft palate. *New England Journal of Medicine*, **2004**, 351, 822-4	59.2	10

125	Haplotype and missing data inference in nuclear families. *Genome Research*, **2004**, 14, 1624-32	9.7	36
124	The Human MitoChip: a high-throughput sequencing microarray for mitochondrial mutation detection. *Genome Research*, **2004**, 14, 812-9	9.7	201
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