

Aravinda Chakravarti

List of Publications by Citations

Source: <https://exaly.com/author-pdf/4437471/aravinda-chakravarti-publications-by-citations.pdf>

Version: 2024-04-20

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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 | A global reference for human genetic variation. <i>Nature</i> , 2015 , 526, 68-74 | 50.4 | 8599 |
| 303 | Finding the missing heritability of complex diseases. <i>Nature</i> , 2009 , 461, 747-53 | 50.4 | 6084 |
| 302 | A second generation human haplotype map of over 3.1 million SNPs. <i>Nature</i> , 2007 , 449, 851-61 | 50.4 | 3647 |
| 301 | Genetic studies of body mass index yield new insights for obesity biology. <i>Nature</i> , 2015 , 518, 197-206 | 50.4 | 2687 |
| 300 | Discovery and refinement of loci associated with lipid levels. <i>Nature Genetics</i> , 2013 , 45, 1274-1283 | 36.3 | 1904 |
| 299 | Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. <i>Nature Genetics</i> , 2013 , 45, 984-94 | 36.3 | 1628 |
| 298 | Genome-wide detection and characterization of positive selection in human populations. <i>Nature</i> , 2007 , 449, 913-8 | 50.4 | 1367 |
| 297 | Defining the role of common variation in the genomic and biological architecture of adult human height. <i>Nature Genetics</i> , 2014 , 46, 1173-86 | 36.3 | 1339 |
| 296 | Association between microdeletion and microduplication at 16p11.2 and autism. <i>New England Journal of Medicine</i> , 2008 , 358, 667-75 | 59.2 | 1249 |
| 295 | Genome-wide association scan shows genetic variants in the FTO gene are associated with obesity-related traits. <i>PLoS Genetics</i> , 2007 , 3, e115 | 6 | 1231 |
| 294 | DNA duplication associated with Charcot-Marie-Tooth disease type 1A. <i>Cell</i> , 1991 , 66, 219-32 | 56.2 | 1161 |
| 293 | Genome-wide association study of blood pressure and hypertension. <i>Nature Genetics</i> , 2009 , 41, 677-87 | 36.3 | 1065 |
| 292 | New genetic loci link adipose and insulin biology to body fat distribution. <i>Nature</i> , 2015 , 518, 187-196 | 50.4 | 920 |
| 291 | Patterns of single-nucleotide polymorphisms in candidate genes for blood-pressure homeostasis. <i>Nature Genetics</i> , 1999 , 22, 239-47 | 36.3 | 916 |
| 290 | Variations on a theme: cataloging human DNA sequence variation. <i>Science</i> , 1997 , 278, 1580-1 | 33.3 | 849 |
| 289 | Development of human protein reference database as an initial platform for approaching systems biology in humans. <i>Genome Research</i> , 2003 , 13, 2363-71 | 9.7 | 823 |
| 288 | A missense mutation of the endothelin-B receptor gene in multigenic Hirschsprung's disease. <i>Cell</i> , 1994 , 79, 1257-66 | 56.2 | 796 |

| | | | |
|-----|--|------|-----|
| 287 | A DNA polymorphism discovery resource for research on human genetic variation. <i>Genome Research</i> , 1998 , 8, 1229-31 | 9.7 | 627 |
| 286 | Common variants associated with plasma triglycerides and risk for coronary artery disease. <i>Nature Genetics</i> , 2013 , 45, 1345-52 | 36.3 | 597 |
| 285 | New goals for the U.S. Human Genome Project: 1998-2003. <i>Science</i> , 1998 , 282, 682-9 | 33.3 | 540 |
| 284 | Genomic alterations in cultured human embryonic stem cells. <i>Nature Genetics</i> , 2005 , 37, 1099-103 | 36.3 | 525 |
| 283 | Population genetics--making sense out of sequence. <i>Nature Genetics</i> , 1999 , 21, 56-60 | 36.3 | 487 |
| 282 | A common genetic variant in the neurexin superfamily member CNTNAP2 increases familial risk of autism. <i>American Journal of Human Genetics</i> , 2008 , 82, 160-4 | 11 | 482 |
| 281 | A genome-wide linkage and association scan reveals novel loci for autism. <i>Nature</i> , 2009 , 461, 802-8 | 50.4 | 474 |
| 280 | A common genetic variant in the NOS1 regulator NOS1AP modulates cardiac repolarization. <i>Nature Genetics</i> , 2006 , 38, 644-51 | 36.3 | 438 |
| 279 | Schizophrenia susceptibility loci on chromosomes 13q32 and 8p21. <i>Nature Genetics</i> , 1998 , 20, 70-3 | 36.3 | 435 |
| 278 | The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. <i>American Journal of Human Genetics</i> , 2015 , 97, 199-215 | 11 | 432 |
| 277 | Meta-analysis identifies six new susceptibility loci for atrial fibrillation. <i>Nature Genetics</i> , 2012 , 44, 670-5 | 36.3 | 429 |
| 276 | Automated construction of genetic linkage maps using an expert system (MultiMap): a human genome linkage map. <i>Nature Genetics</i> , 1994 , 6, 384-90 | 36.3 | 408 |
| 275 | Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. <i>Nature Genetics</i> , 2018 , 50, 1412-1425 | 36.3 | 386 |
| 274 | A common sex-dependent mutation in a RET enhancer underlies Hirschsprung disease risk. <i>Nature</i> , 2005 , 434, 857-63 | 50.4 | 377 |
| 273 | Haploinsufficiency of telomerase reverse transcriptase leads to anticipation in autosomal dominant dyskeratosis congenita. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005 , 102, 15960-4 | 11.5 | 365 |
| 272 | Common variants in KCNN3 are associated with lone atrial fibrillation. <i>Nature Genetics</i> , 2010 , 42, 240-4 | 36.3 | 362 |
| 271 | Genome-wide association study of PR interval. <i>Nature Genetics</i> , 2010 , 42, 153-9 | 36.3 | 340 |
| 270 | Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. <i>Nature Genetics</i> , 2011 , 43, 1005-11 | 36.3 | 338 |

| | | | |
|-----|---|------|-----|
| 269 | Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. <i>Science</i> , 2016 , 351, 1166-71 | 33.3 | 325 |
| 268 | Genome-wide association analysis identifies novel blood pressure loci and offers biological insights into cardiovascular risk. <i>Nature Genetics</i> , 2017 , 49, 403-415 | 36.3 | 313 |
| 267 | Common variants at ten loci modulate the QT interval duration in the QTSCD Study. <i>Nature Genetics</i> , 2009 , 41, 407-14 | 36.3 | 308 |
| 266 | Variants in ZFHX3 are associated with atrial fibrillation in individuals of European ancestry. <i>Nature Genetics</i> , 2009 , 41, 879-81 | 36.3 | 307 |
| 265 | A BDNF coding variant is associated with the NEO personality inventory domain neuroticism, a risk factor for depression. <i>Neuropsychopharmacology</i> , 2003 , 28, 397-401 | 8.7 | 287 |
| 264 | Multiple loci influence erythrocyte phenotypes in the CHARGE Consortium. <i>Nature Genetics</i> , 2009 , 41, 1191-8 | 36.3 | 285 |
| 263 | A homozygous mutation in the endothelin-3 gene associated with a combined Waardenburg type 2 and Hirschsprung phenotype (Shah-Waardenburg syndrome). <i>Nature Genetics</i> , 1996 , 12, 445-7 | 36.3 | 277 |
| 262 | Germline mutations in glial cell line-derived neurotrophic factor (GDNF) and RET in a Hirschsprung disease patient. <i>Nature Genetics</i> , 1996 , 14, 341-4 | 36.3 | 252 |
| 261 | The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. <i>Nature Genetics</i> , 2016 , 48, 1171-1184 | 36.3 | 251 |
| 260 | Common variants in 22 loci are associated with QRS duration and cardiac ventricular conduction. <i>Nature Genetics</i> , 2010 , 42, 1068-76 | 36.3 | 249 |
| 259 | Genome-wide association study of coronary heart disease and its risk factors in 8,090 African Americans: the NHLBI CARE Project. <i>PLoS Genetics</i> , 2011 , 7, e1001300 | 6 | 249 |
| 258 | From the Cover: Whole-genome association study identifies STK39 as a hypertension susceptibility gene. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009 , 106, 226-31 | 11.5 | 240 |
| 257 | High-throughput variation detection and genotyping using microarrays. <i>Genome Research</i> , 2001 , 11, 1913-25 | 9.7 | 239 |
| 256 | Parallel genotyping of human SNPs using generic high-density oligonucleotide tag arrays. <i>Genome Research</i> , 2000 , 10, 853-60 | 9.7 | 232 |
| 255 | Genome-wide association study and mouse model identify interaction between RET and EDNRB pathways in Hirschsprung disease. <i>Nature Genetics</i> , 2002 , 32, 237-44 | 36.3 | 229 |
| 254 | Identity-by-descent and association mapping of a recessive gene for Hirschsprung disease on human chromosome 13q22. <i>Human Molecular Genetics</i> , 1994 , 3, 1217-25 | 5.6 | 228 |
| 253 | Human embryonic stem cells have a unique epigenetic signature. <i>Genome Research</i> , 2006 , 16, 1075-83 | 9.7 | 224 |
| 252 | Segregation at three loci explains familial and population risk in Hirschsprung disease. <i>Nature Genetics</i> , 2002 , 31, 89-93 | 36.3 | 223 |

| | | | |
|-----|--|------|-----|
| 251 | The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. <i>PLoS Genetics</i> , 2015 , 11, e1005378 | 6 | 220 |
| 250 | Identifying disease alleles by genome sharing. <i>Nature Genetics</i> , 1999 , 23, 25-25 | 36.3 | 213 |
| 249 | Parental origin and phenotype of triploidy in spontaneous abortions: predominance of diandry and association with the partial hydatidiform mole. <i>American Journal of Human Genetics</i> , 2000 , 66, 1807-20 | 11 | 208 |
| 248 | The Human MitoChip: a high-throughput sequencing microarray for mitochondrial mutation detection. <i>Genome Research</i> , 2004 , 14, 812-9 | 9.7 | 201 |
| 247 | Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. <i>Nature Genetics</i> , 2014 , 46, 826-36 | 36.3 | 199 |
| 246 | Differential contributions of rare and common, coding and noncoding Ret mutations to multifactorial Hirschsprung disease liability. <i>American Journal of Human Genetics</i> , 2010 , 87, 60-74 | 11 | 198 |
| 245 | A bivariate genome-wide approach to metabolic syndrome: STAMPEED consortium. <i>Diabetes</i> , 2011 , 60, 1329-39 | 0.9 | 194 |
| 244 | Differential susceptibility to hypertension is due to selection during the out-of-Africa expansion. <i>PLoS Genetics</i> , 2005 , 1, e82 | 6 | 175 |
| 243 | Haplotype inference in random population samples. <i>American Journal of Human Genetics</i> , 2002 , 71, 1129-37 | 37 | 164 |
| 242 | Joint analysis of psychiatric disorders increases accuracy of risk prediction for schizophrenia, bipolar disorder, and major depressive disorder. <i>American Journal of Human Genetics</i> , 2015 , 96, 283-94 | 11 | 161 |
| 241 | Genome-wide association analyses using electronic health records identify new loci influencing blood pressure variation. <i>Nature Genetics</i> , 2017 , 49, 54-64 | 36.3 | 157 |
| 240 | Association of hypertension drug target genes with blood pressure and hypertension in 86,588 individuals. <i>Hypertension</i> , 2011 , 57, 903-10 | 8.5 | 154 |
| 239 | Multiplex PCR of three dinucleotide repeats in the Prader-Willi/Angelman critical region (15q11-q13): molecular diagnosis and mechanism of uniparental disomy. <i>Human Molecular Genetics</i> , 1993 , 2, 143-51 | 5.6 | 153 |
| 238 | Meta-analysis identifies common and rare variants influencing blood pressure and overlapping with metabolic trait loci. <i>Nature Genetics</i> , 2016 , 48, 1162-70 | 36.3 | 152 |
| 237 | Undetected genotyping errors cause apparent overtransmission of common alleles in the transmission/disequilibrium test. <i>American Journal of Human Genetics</i> , 2003 , 72, 598-610 | 11 | 149 |
| 236 | Association of genetic variation with systolic and diastolic blood pressure among African Americans: the Candidate Gene Association Resource study. <i>Human Molecular Genetics</i> , 2011 , 20, 2273-84 | 5.6 | 146 |
| 235 | Genome-wide association analysis of blood-pressure traits in African-ancestry individuals reveals common associated genes in African and non-African populations. <i>American Journal of Human Genetics</i> , 2013 , 93, 545-54 | 11 | 145 |
| 234 | Genome-wide association studies of serum magnesium, potassium, and sodium concentrations identify six Loci influencing serum magnesium levels. <i>PLoS Genetics</i> , 2010 , 6, e1001045 | 6 | 144 |

| | | | |
|-----|---|------|-----|
| 233 | Polymorphisms in the NOS1AP gene modulate QT interval duration and risk of arrhythmias in the long QT syndrome. <i>Journal of the American College of Cardiology</i> , 2010 , 55, 2745-52 | 15.1 | 143 |
| 232 | Genetic variations in nitric oxide synthase 1 adaptor protein are associated with sudden cardiac death in US white community-based populations. <i>Circulation</i> , 2009 , 119, 940-51 | 16.7 | 141 |
| 231 | GENOME ENGINEERING. The Genome Project-Write. <i>Science</i> , 2016 , 353, 126-7 | 33.3 | 138 |
| 230 | Exhaustive allelic transmission disequilibrium tests as a new approach to genome-wide association studies. <i>Nature Genetics</i> , 2004 , 36, 1181-8 | 36.3 | 138 |
| 229 | A gene for Hirschsprung disease (megacolon) in the pericentromeric region of human chromosome 10. <i>Nature Genetics</i> , 1993 , 4, 351-6 | 36.3 | 136 |
| 228 | Gene-centric meta-analysis in 87,736 individuals of European ancestry identifies multiple blood-pressure-related loci. <i>American Journal of Human Genetics</i> , 2014 , 94, 349-60 | 11 | 131 |
| 227 | Phenotype variation in two-locus mouse models of Hirschsprung disease: tissue-specific interaction between Ret and Ednrb. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003 , 100, 1826-31 | 11.5 | 120 |
| 226 | Directional dominance on stature and cognition in diverse human populations. <i>Nature</i> , 2015 , 523, 459-463 | 50.4 | 119 |
| 225 | Loci influencing blood pressure identified using a cardiovascular gene-centric array. <i>Human Molecular Genetics</i> , 2013 , 22, 1663-78 | 5.6 | 119 |
| 224 | Global implementation of genomic medicine: We are not alone. <i>Science Translational Medicine</i> , 2015 , 7, 290ps13 | 17.5 | 112 |
| 223 | Nature, nurture and human disease. <i>Nature</i> , 2003 , 421, 412-4 | 50.4 | 110 |
| 222 | Future of genetics of mood disorders research. <i>Biological Psychiatry</i> , 2002 , 52, 457-77 | 7.9 | 108 |
| 221 | Associations between hypertension and genes in the renin-angiotensin system. <i>Hypertension</i> , 2003 , 41, 1027-34 | 8.5 | 105 |
| 220 | Allele frequency distributions in pooled DNA samples: applications to mapping complex disease genes. <i>Genome Research</i> , 1998 , 8, 111-23 | 9.7 | 104 |
| 219 | Loss of Ectenin function in severe autism. <i>Nature</i> , 2015 , 520, 51-6 | 50.4 | 97 |
| 218 | Identification of a sudden cardiac death susceptibility locus at 2q24.2 through genome-wide association in European ancestry individuals. <i>PLoS Genetics</i> , 2011 , 7, e1002158 | 6 | 95 |
| 217 | Trans-ethnic fine-mapping of lipid loci identifies population-specific signals and allelic heterogeneity that increases the trait variance explained. <i>PLoS Genetics</i> , 2013 , 9, e1003379 | 6 | 94 |
| 216 | Multiple genes for essential-hypertension susceptibility on chromosome 1q. <i>American Journal of Human Genetics</i> , 2007 , 80, 253-64 | 11 | 93 |

| | | | |
|-----|---|------|----|
| 215 | Multiple loci are associated with white blood cell phenotypes. <i>PLoS Genetics</i> , 2011 , 7, e1002113 | 6 | 92 |
| 214 | Sequence variations in the public human genome data reflect a bottlenecked population history. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003 , 100, 376-81 | 11.5 | 92 |
| 213 | Contribution of rare and common variants determine complex diseases-Hirschsprung disease as a model. <i>Developmental Biology</i> , 2013 , 382, 320-9 | 3.1 | 90 |
| 212 | Pleiotropic skeletal and ocular phenotypes of the mouse mutation congenital hydrocephalus (ch/Mf1) arise from a winged helix/forkhead transcriptionfactor gene. <i>Human Molecular Genetics</i> , 1999 , 8, 625-37 | 5.6 | 88 |
| 211 | EDNRB/EDN3 and Hirschsprung disease type II. <i>Pigment Cell & Melanoma Research</i> , 2001 , 14, 161-9 | | 86 |
| 210 | Drug-sensitized zebrafish screen identifies multiple genes, including GINS3, as regulators of myocardial repolarization. <i>Circulation</i> , 2009 , 120, 553-9 | 16.7 | 84 |
| 209 | An integrated genetic linkage map of the laboratory rat. <i>Mammalian Genome</i> , 1998 , 9, 521-30 | 3.2 | 83 |
| 208 | Functional loss of semaphorin 3C and/or semaphorin 3D and their epistatic interaction with ret are critical to Hirschsprung disease liability. <i>American Journal of Human Genetics</i> , 2015 , 96, 581-96 | 11 | 82 |
| 207 | Endothelin-3 frameshift mutation in congenital central hypoventilation syndrome. <i>Nature Genetics</i> , 1996 , 13, 395-6 | 36.3 | 82 |
| 206 | Two-locus models of disease. <i>Genetic Epidemiology</i> , 1992 , 9, 347-65 | 2.6 | 82 |
| 205 | Linkage disequilibrium analysis of biallelic DNA markers, human quantitative trait loci, and threshold-defined case and control subjects. <i>American Journal of Human Genetics</i> , 2000 , 67, 1208-18 | 11 | 81 |
| 204 | Gene-age interactions in blood pressure regulation: a large-scale investigation with the CHARGE, Global BPgen, and ICBP Consortia. <i>American Journal of Human Genetics</i> , 2014 , 95, 24-38 | 11 | 80 |
| 203 | Genomics in sudden cardiac death. <i>Circulation Research</i> , 2004 , 94, 712-23 | 15.7 | 80 |
| 202 | Enhancer Variants Synergistically Drive Dysfunction of a Gene Regulatory Network In Hirschsprung Disease. <i>Cell</i> , 2016 , 167, 355-368.e10 | 56.2 | 80 |
| 201 | Positional identification of hypertension susceptibility genes on chromosome 2. <i>Hypertension</i> , 2004 , 43, 477-82 | 8.5 | 79 |
| 200 | A genetic linkage map of 27 markers on human chromosome 21. <i>Genomics</i> , 1991 , 9, 407-19 | 4.3 | 78 |
| 199 | 52 Genetic Loci Influencing Myocardial Mass. <i>Journal of the American College of Cardiology</i> , 2016 , 68, 1435-1448 | 15.1 | 76 |
| 198 | Genetics and genomics for the prevention and treatment of cardiovascular disease: update: a scientific statement from the American Heart Association. <i>Circulation</i> , 2013 , 128, 2813-51 | 16.7 | 76 |

| | | | |
|-----|--|------|----|
| 197 | A genetic linkage map of 17 markers on human chromosome 21. <i>Genomics</i> , 1989 , 4, 579-91 | 4.3 | 75 |
| 196 | Drift variances of F_{ST} and G_{ST} statistics obtained from a finite number of isolated populations. <i>Theoretical Population Biology</i> , 1977 , 11, 307-25 | 1.2 | 75 |
| 195 | Mitochondrial DNA variants of respiratory complex I that uniquely characterize haplogroup T2 are associated with increased risk of age-related macular degeneration. <i>PLoS ONE</i> , 2009 , 4, e5508 | 3.7 | 74 |
| 194 | Identifying allelic loss and homozygous deletions in pancreatic cancer without matched normals using high-density single-nucleotide polymorphism arrays. <i>Cancer Research</i> , 2006 , 66, 7920-8 | 10.1 | 74 |
| 193 | Molecular Genetic Anatomy and Risk Profile of Hirschsprung Disease. <i>New England Journal of Medicine</i> , 2019 , 380, 1421-1432 | 59.2 | 71 |
| 192 | Combined admixture mapping and association analysis identifies a novel blood pressure genetic locus on 5p13: contributions from the CARE consortium. <i>Human Molecular Genetics</i> , 2011 , 20, 2285-95 | 5.6 | 70 |
| 191 | Mean and variance of F_{ST} in a finite number of incompletely isolated populations. <i>Theoretical Population Biology</i> , 1977 , 11, 291-306 | 1.2 | 70 |
| 190 | On consanguineous marriages and the genetic load. <i>Human Genetics</i> , 1977 , 36, 47-54 | 6.3 | 70 |
| 189 | Evaluation of the RET regulatory landscape reveals the biological relevance of a HSCR-implicated enhancer. <i>Human Molecular Genetics</i> , 2005 , 14, 3837-45 | 5.6 | 67 |
| 188 | Patterns of genetic variation in Mendelian and complex traits. <i>Annual Review of Genomics and Human Genetics</i> , 2000 , 1, 387-407 | 9.7 | 65 |
| 187 | DNA polymorphism haplotypes of the human apolipoprotein APOA1-APOC3-APOA4 gene cluster. <i>Human Genetics</i> , 1988 , 80, 265-73 | 6.3 | 65 |
| 186 | Associations between genetic variants in the NOS1AP (CAPON) gene and cardiac repolarization in the old order Amish. <i>Human Heredity</i> , 2007 , 64, 214-9 | 1.1 | 64 |
| 185 | Distilling pathophysiology from complex disease genetics. <i>Cell</i> , 2013 , 155, 21-6 | 56.2 | 63 |
| 184 | A linkage map of human chromosome 21:43 PCR markers at average intervals of 2.5 cM. <i>Genomics</i> , 1993 , 16, 562-71 | 4.3 | 62 |
| 183 | Mendelian disorders and multifactorial traits: the big divide or one for all?. <i>Nature Reviews Genetics</i> , 2010 , 11, 380-4 | 30.1 | 60 |
| 182 | Phylogeny of human beta-globin haplotypes and its implications for recent human evolution. <i>American Journal of Physical Anthropology</i> , 1990 , 81, 113-30 | 2.5 | 60 |
| 181 | A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure. <i>American Journal of Human Genetics</i> , 2018 , 102, 375-400 | 11 | 59 |
| 180 | Linkage disequilibrium and haplotype diversity in the genes of the renin-angiotensin system: findings from the family blood pressure program. <i>Genome Research</i> , 2003 , 13, 173-81 | 9.7 | 59 |

| | | | |
|-----|--|------|----|
| 179 | Linkage analysis of the human HMG14 gene on chromosome 21 using a GT dinucleotide repeat as polymorphic marker. <i>Genomics</i> , 1990 , 7, 136-8 | 4.3 | 59 |
| 178 | Safety issues in cell-based intervention trials. <i>Fertility and Sterility</i> , 2003 , 80, 1077-85 | 4.8 | 58 |
| 177 | A genome-wide scan for obesity in African-Americans. <i>Diabetes</i> , 2002 , 51, 541-4 | 0.9 | 58 |
| 176 | Single-trait and multi-trait genome-wide association analyses identify novel loci for blood pressure in African-ancestry populations. <i>PLoS Genetics</i> , 2017 , 13, e1006728 | 6 | 58 |
| 175 | Replication of the Wellcome Trust genome-wide association study of essential hypertension: the Family Blood Pressure Program. <i>European Journal of Human Genetics</i> , 2008 , 16, 1507-11 | 5.3 | 57 |
| 174 | An enhancer polymorphism at the cardiomyocyte intercalated disc protein NOS1AP locus is a major regulator of the QT interval. <i>American Journal of Human Genetics</i> , 2014 , 94, 854-69 | 11 | 56 |
| 173 | Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. <i>Genome Biology</i> , 2017 , 18, 48 | 18.3 | 55 |
| 172 | Understanding cardiovascular disease through the lens of genome-wide association studies. <i>Trends in Genetics</i> , 2009 , 25, 387-94 | 8.5 | 55 |
| 171 | Next-generation sequencing of human mitochondrial reference genomes uncovers high heteroplasmy frequency. <i>PLoS Computational Biology</i> , 2012 , 8, e1002737 | 5 | 54 |
| 170 | The gene for soluble N-ethylmaleimide sensitive factor attachment protein alpha is mutated in hydrocephaly with hop gait (hyh) mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004 , 101, 1748-53 | 11.5 | 54 |
| 169 | Associations of Mitochondrial and Nuclear Mitochondrial Variants and Genes with Seven Metabolic Traits. <i>American Journal of Human Genetics</i> , 2019 , 104, 112-138 | 11 | 54 |
| 168 | Copy number variants in candidate genes are genetic modifiers of Hirschsprung disease. <i>PLoS ONE</i> , 2011 , 6, e21219 | 3.7 | 53 |
| 167 | Effects of long-term averaging of quantitative blood pressure traits on the detection of genetic associations. <i>American Journal of Human Genetics</i> , 2014 , 95, 49-65 | 11 | 52 |
| 166 | Positional identification of variants of Adamts16 linked to inherited hypertension. <i>Human Molecular Genetics</i> , 2009 , 18, 2825-38 | 5.6 | 52 |
| 165 | Elevated frequency and allelic heterogeneity of congenital nephrotic syndrome, Finnish type, in the old order Mennonites. <i>American Journal of Human Genetics</i> , 1999 , 65, 1785-90 | 11 | 52 |
| 164 | Interaction between a chromosome 10 RET enhancer and chromosome 21 in the Down syndrome-Hirschsprung disease association. <i>Human Mutation</i> , 2009 , 30, 771-5 | 4.7 | 50 |
| 163 | Follow-up of a major linkage peak on chromosome 1 reveals suggestive QTLs associated with essential hypertension: GenNet study. <i>European Journal of Human Genetics</i> , 2009 , 17, 1650-7 | 5.3 | 50 |
| 162 | A genome-wide linkage analysis investigating the determinants of blood pressure in whites and African Americans. <i>American Journal of Hypertension</i> , 2003 , 16, 151-3 | 2.3 | 50 |

| | | | |
|-----|--|------|----|
| 161 | Human GFRA1: cloning, mapping, genomic structure, and evaluation as a candidate gene for Hirschsprung disease susceptibility. <i>Genomics</i> , 1998 , 48, 354-62 | 4.3 | 50 |
| 160 | Population variation in total genetic risk of Hirschsprung disease from common RET, SEMA3 and NRG1 susceptibility polymorphisms. <i>Human Molecular Genetics</i> , 2015 , 24, 2997-3003 | 5.6 | 49 |
| 159 | Microsatellite polymorphism linkage map of human chromosome 13q. <i>Genomics</i> , 1993 , 15, 376-86 | 4.3 | 49 |
| 158 | Genome-wide association study identifies GPC5 as a novel genetic locus protective against sudden cardiac arrest. <i>PLoS ONE</i> , 2010 , 5, e9879 | 3.7 | 48 |
| 157 | Genetic variation associated with circulating monocyte count in the eMERGE Network. <i>Human Molecular Genetics</i> , 2013 , 22, 2119-27 | 5.6 | 46 |
| 156 | Population bottlenecks as a potential major shaping force of human genome architecture. <i>PLoS Genetics</i> , 2007 , 3, e119 | 6 | 46 |
| 155 | Novel approach identifies SNPs in SLC2A10 and KCNK9 with evidence for parent-of-origin effect on body mass index. <i>PLoS Genetics</i> , 2014 , 10, e1004508 | 6 | 45 |
| 154 | Down syndrome consequent to a cryptic maternal 12p;21q chromosome translocation. <i>American Journal of Medical Genetics Part A</i> , 1995 , 56, 67-71 | | 45 |
| 153 | A graphical representation of genetic and physical maps: the Marey map. <i>Genomics</i> , 1991 , 11, 219-22 | 4.3 | 45 |
| 152 | Cytogenetics and origins of pediatric germ cell tumors. <i>Cancer Genetics and Cytogenetics</i> , 1994 , 74, 54-8 | | 44 |
| 151 | Waardenburg syndrome and Hirschsprung disease: evidence for pleiotropic effects of a single dominant gene. <i>American Journal of Medical Genetics Part A</i> , 1990 , 35, 100-4 | | 44 |
| 150 | Methods for studying recombination on chromosomes that undergo nondisjunction. <i>Genomics</i> , 1987 , 1, 35-42 | 4.3 | 43 |
| 149 | The road ahead in genetics and genomics. <i>Nature Reviews Genetics</i> , 2020 , 21, 581-596 | 30.1 | 43 |
| 148 | Mining gold dust under the genome wide significance level: a two-stage approach to analysis of GWAS. <i>Genetic Epidemiology</i> , 2011 , 35, 111-8 | 2.6 | 41 |
| 147 | Cloning of a novel homeobox-containing gene, PKNOX1, and mapping to human chromosome 21q22.3. <i>Genomics</i> , 1997 , 41, 193-200 | 4.3 | 39 |
| 146 | A multilevel model to address batch effects in copy number estimation using SNP arrays. <i>Biostatistics</i> , 2011 , 12, 33-50 | 3.7 | 38 |
| 145 | Polymorphisms in the mitochondrial DNA control region and frailty in older adults. <i>PLoS ONE</i> , 2010 , 5, e11069 | 3.7 | 37 |
| 144 | Hemostasis, inflammation, and fatal and nonfatal coronary heart disease: long-term follow-up of the atherosclerosis risk in communities (ARIC) cohort. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2009 , 29, 2182-90 | 9.4 | 37 |

| | | | |
|-----|--|------|----|
| 143 | Allele-specific expression in the germline of patients with familial pancreatic cancer: an unbiased approach to cancer gene discovery. <i>Cancer Biology and Therapy</i> , 2008 , 7, 135-44 | 4.6 | 37 |
| 142 | Patterns of meiotic recombination on the long arm of human chromosome 21. <i>Genome Research</i> , 2000 , 10, 1319-32 | 9.7 | 37 |
| 141 | Haplotype and missing data inference in nuclear families. <i>Genome Research</i> , 2004 , 14, 1624-32 | 9.7 | 36 |
| 140 | Elementary methods for the analysis of dichotomous outcomes in unselected samples of twins. <i>Genetic Epidemiology</i> , 1992 , 9, 273-87 | 2.6 | 36 |
| 139 | Rare Exome Sequence Variants in CLCN6 Reduce Blood Pressure Levels and Hypertension Risk. <i>Circulation: Cardiovascular Genetics</i> , 2016 , 9, 64-70 | | 35 |
| 138 | Genetics and biology of human ovarian teratomas. III. Cytogenetics and origins of malignant ovarian germ cell tumors. <i>Cancer Genetics and Cytogenetics</i> , 1992 , 62, 58-65 | | 34 |
| 137 | Effects of RET and NRG1 polymorphisms in Indonesian patients with Hirschsprung disease. <i>Journal of Pediatric Surgery</i> , 2014 , 49, 1614-8 | 2.6 | 33 |
| 136 | Phenotype-genotype correlation in Hirschsprung disease is illuminated by comparative analysis of the RET protein sequence. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005 , 102, 8949-54 | 11.5 | 32 |
| 135 | Direct Estimates of the Genomic Contributions to Blood Pressure Heritability within a Population-Based Cohort (ARIC). <i>PLoS ONE</i> , 2015 , 10, e0133031 | 3.7 | 32 |
| 134 | Revealing rate-limiting steps in complex disease biology: The crucial importance of studying rare, extreme-phenotype families. <i>BioEssays</i> , 2016 , 38, 578-86 | 4.1 | 32 |
| 133 | Transcriptional regulation of PRPF31 gene expression by MSR1 repeat elements causes incomplete penetrance in retinitis pigmentosa. <i>Scientific Reports</i> , 2016 , 6, 19450 | 4.9 | 31 |
| 132 | Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries. <i>PLoS ONE</i> , 2018 , 13, e0198166 | 3.7 | 31 |
| 131 | Rapid and efficient human mutation detection using a bench-top next-generation DNA sequencer. <i>Human Mutation</i> , 2012 , 33, 281-9 | 4.7 | 31 |
| 130 | A comprehensive evaluation of the genetic architecture of sudden cardiac arrest. <i>European Heart Journal</i> , 2018 , 39, 3961-3969 | 9.5 | 31 |
| 129 | A population-based study of KCNH7 p.Arg394His and bipolar spectrum disorder. <i>Human Molecular Genetics</i> , 2014 , 23, 6395-406 | 5.6 | 30 |
| 128 | Variation in the ciliary neurotrophic factor gene and muscle strength in older Caucasian women. <i>Journal of the American Geriatrics Society</i> , 2006 , 54, 823-6 | 5.6 | 30 |
| 127 | Linkage mapping of highly informative DNA polymorphisms within the human interferon-alpha receptor gene on chromosome 21. <i>Genomics</i> , 1991 , 11, 573-6 | 4.3 | 30 |
| 126 | A compelling genetic hypothesis for a complex disease: PRODH2/DGCR6 variation leads to schizophrenia susceptibility. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002 , 99, 4755-6 | 11.5 | 29 |

| | | | |
|-----|---|------|----|
| 125 | Parent-of-origin effects in autism identified through genome-wide linkage analysis of 16,000 SNPs. <i>PLoS ONE</i> , 2010 , 5, e12513 | 3.7 | 28 |
| 124 | Linkage mapping of the highly informative DNA marker D21S156 to human chromosome 21 using a polymorphic GT dinucleotide repeat. <i>Genomics</i> , 1990 , 8, 400-2 | 4.3 | 28 |
| 123 | MicroRNAs in the miR-17 and miR-15 families are downregulated in chronic kidney disease with hypertension. <i>PLoS ONE</i> , 2017 , 12, e0176734 | 3.7 | 27 |
| 122 | Lack of association between a biallelic polymorphism in the adducin gene and blood pressure in whites and African Americans. <i>American Journal of Hypertension</i> , 2000 , 13, 693-8 | 2.3 | 26 |
| 121 | A radiation hybrid map of 48 loci including the clouston hidrotic ectodermal dysplasia locus in the pericentromeric region of chromosome 13q. <i>Genomics</i> , 1999 , 56, 127-30 | 4.3 | 26 |
| 120 | Guidelines for human linkage maps. An International System for Human Linkage Maps (ISLM, 1990). <i>Annals of Human Genetics</i> , 1991 , 55, 1-6 | 2.2 | 26 |
| 119 | Multiple independent genetic factors at NOS1AP modulate the QT interval in a multi-ethnic population. <i>PLoS ONE</i> , 2009 , 4, e4333 | 3.7 | 25 |
| 118 | Defining the contribution of CNTNAP2 to autism susceptibility. <i>PLoS ONE</i> , 2013 , 8, e77906 | 3.7 | 25 |
| 117 | Proteins linked to autosomal dominant and autosomal recessive disorders harbor characteristic rare missense mutation distribution patterns. <i>Human Molecular Genetics</i> , 2015 , 24, 5995-6002 | 5.6 | 24 |
| 116 | An ancestral variant of Secretogranin II confers regulation by PHOX2 transcription factors and association with hypertension. <i>Human Molecular Genetics</i> , 2007 , 16, 1752-64 | 5.6 | 24 |
| 115 | Preliminary ordering of multiple linked loci using pairwise linkage data. <i>Genetic Epidemiology</i> , 1992 , 9, 367-75 | 2.6 | 24 |
| 114 | Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. <i>Human Molecular Genetics</i> , 2016 , 25, 5265-5275 | 5.6 | 23 |
| 113 | Age, sex, and the familial risk of rheumatoid arthritis. <i>American Journal of Epidemiology</i> , 1996 , 144, 15-24 | 3.8 | 22 |
| 112 | A PIGN mutation responsible for multiple congenital anomalies-hypotonia-seizures syndrome 1 (MCAHS1) in an Israeli-Arab family. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 176-82 | 2.5 | 22 |
| 111 | An investigation of genome-wide associations of hypertension with microsatellite markers in the family blood pressure program (FBPP). <i>Human Genetics</i> , 2007 , 121, 577-90 | 6.3 | 21 |
| 110 | Haplotype association analysis of AGT variants with hypertension-related traits: the HyperGEN study. <i>Human Heredity</i> , 2005 , 60, 164-76 | 1.1 | 21 |
| 109 | On the probability that a novel variant is a disease-causing mutation. <i>Genome Research</i> , 2005 , 15, 960-6 | 9.7 | 21 |
| 108 | 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. <i>Circulation Research</i> , 2013 , 112, 318-26 | 15.7 | 20 |

| | | | |
|-----|---|------|----|
| 107 | Genomic structure of the gene for the SH2 and pleckstrin homology domain-containing protein GRB10 and evaluation of its role in Hirschsprung disease. <i>Oncogene</i> , 1998 , 17, 3065-70 | 9.2 | 20 |
| 106 | Erythrocyte sodium-lithium countertransport and blood pressure: a genome-wide linkage study. <i>Hypertension</i> , 2003 , 41, 842-6 | 8.5 | 20 |
| 105 | RET somatic mutations are underrecognized in Hirschsprung disease. <i>Genetics in Medicine</i> , 2018 , 20, 770-77 | 8.7 | 20 |
| 104 | Public stem cell banks: considerations of justice in stem cell research and therapy. <i>Hastings Center Report</i> , 2003 , 33, 13-27 | 3.3 | 20 |
| 103 | Linkage mapping of the cystathionine beta-synthase (CBS) gene on human chromosome 21 using a DNA polymorphism in the 3' untranslated region. <i>Human Genetics</i> , 1993 , 90, 566-8 | 6.3 | 19 |
| 102 | Evidence against close linkage of unipolar affective illness to human chromosome 11p markers HRAS1 and INS and chromosome Xq marker DXS52. <i>Biological Psychiatry</i> , 1990 , 28, 63-72 | 7.9 | 19 |
| 101 | Etiological heterogeneity in Hodgkin's disease: HLA linked and unlinked determinants of susceptibility independent of histological concordance. <i>Genetic Epidemiology</i> , 1986 , 3, 407-15 | 2.6 | 19 |
| 100 | Variant Discovery and Fine Mapping of Genetic Loci Associated with Blood Pressure Traits in Hispanics and African Americans. <i>PLoS ONE</i> , 2016 , 11, e0164132 | 3.7 | 19 |
| 99 | Intestinal Neuronal Dysplasia-Like Submucosal Ganglion Cell Hyperplasia at the Proximal Margins of Hirschsprung Disease Resections. <i>Pediatric and Developmental Pathology</i> , 2015 , 18, 466-76 | 2.2 | 18 |
| 98 | Chromosome 21 scan in Down syndrome reveals DSCAM as a predisposing locus in Hirschsprung disease. <i>PLoS ONE</i> , 2013 , 8, e62519 | 3.7 | 18 |
| 97 | The impact of data quality on the identification of complex disease genes: experience from the Family Blood Pressure Program. <i>European Journal of Human Genetics</i> , 2006 , 14, 469-77 | 5.3 | 18 |
| 96 | An evaluation of the assembly of an approximately 15-Mb region on human chromosome 13q32-q33 linked to bipolar disorder and schizophrenia. <i>Genomics</i> , 2002 , 79, 635-56 | 4.3 | 18 |
| 95 | Estimating genome-wide copy number using allele-specific mixture models. <i>Journal of Computational Biology</i> , 2008 , 15, 857-66 | 1.7 | 17 |
| 94 | Estimation of the frequency of isoform-genotype discrepancies at the apolipoprotein E locus in heterozygotes for the isoforms. <i>Genetic Epidemiology</i> , 1992 , 9, 239-48 | 2.6 | 17 |
| 93 | Genetics. Mendelian puzzles. <i>Science</i> , 2012 , 335, 930-1 | 33.3 | 16 |
| 92 | Quantifying and modeling birth order effects in autism. <i>PLoS ONE</i> , 2011 , 6, e26418 | 3.7 | 16 |
| 91 | Being human: kinship: race relations. <i>Nature</i> , 2009 , 457, 380-1 | 50.4 | 15 |
| 90 | Five blood pressure loci identified by an updated genome-wide linkage scan: meta-analysis of the Family Blood Pressure Program. <i>American Journal of Hypertension</i> , 2011 , 24, 347-54 | 2.3 | 15 |

| | | | |
|----|---|------|----|
| 89 | Dietary intake and gene variation influence the response of plasma lipids to dietary intervention. <i>Genetic Epidemiology</i> , 1992 , 9, 249-60 | 2.6 | 15 |
| 88 | Tests of linkage and heterogeneity in Mendelian diseases using identity by descent scores. <i>Genetic Epidemiology</i> , 1987 , 4, 255-66 | 2.6 | 15 |
| 87 | Variation in allele frequencies among caste groups of the Dhangars of Maharashtra, India: an analysis with Wright's F_{st} statistic. <i>Annals of Human Biology</i> , 1977 , 4, 275-80 | 1.7 | 15 |
| 86 | 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. <i>PLoS ONE</i> , 2014 , 9, e109155 | 3.7 | 15 |
| 85 | Leveraging linkage evidence to identify low-frequency and rare variants on 16p13 associated with blood pressure using TOPMed whole genome sequencing data. <i>Human Genetics</i> , 2019 , 138, 199-210 | 6.3 | 14 |
| 84 | A multi-ancestry genome-wide study incorporating gene-smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. <i>Human Molecular Genetics</i> , 2019 , 28, 2615-2633 | 5.6 | 14 |
| 83 | 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. <i>PLoS ONE</i> , 2018 , 13, e0200486 | 3.7 | 14 |
| 82 | Genomic contributions to Mendelian disease. <i>Genome Research</i> , 2011 , 21, 643-4 | 9.7 | 14 |
| 81 | D21S215 is a (GT) $_n$ polymorphic marker close to centromeric alphoid sequences on chromosome 21. <i>Genomics</i> , 1992 , 13, 1365-7 | 4.3 | 14 |
| 80 | Multiple variant enhancers modulate its cardiac gene expression and the QT interval. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019 , 116, 10636-10645 | 11.5 | 13 |
| 79 | The CD4/CD8 ratio: message in a bottle?. <i>Nature Medicine</i> , 1995 , 1, 1240-1 | 50.5 | 13 |
| 78 | Aggregation of colon cancer in family data. <i>Genetic Epidemiology</i> , 1984 , 1, 53-61 | 2.6 | 13 |
| 77 | Common variation in fatty acid metabolic genes and risk of incident sudden cardiac arrest. <i>Heart Rhythm</i> , 2014 , 11, 471-7 | 6.7 | 12 |
| 76 | A polymorphic 3'UTR element in ATP1B1 regulates alternative polyadenylation and is associated with blood pressure. <i>PLoS ONE</i> , 2013 , 8, e76290 | 3.7 | 12 |
| 75 | A population association study of angiotensinogen polymorphisms and haplotypes with left ventricular phenotypes. <i>Hypertension</i> , 2005 , 46, 1294-9 | 8.5 | 12 |
| 74 | Linkage mapping of D21S171 to the distal long arm of human chromosome 21 using a polymorphic (AC) $_n$ dinucleotide repeat. <i>Human Genetics</i> , 1991 , 87, 401-4 | 6.3 | 12 |
| 73 | Perspectives on Human Variation through the Lens of Diversity and Race. <i>Cold Spring Harbor Perspectives in Biology</i> , 2015 , 7, a023358 | 10.2 | 11 |
| 72 | Genome-wide association study of Hirschsprung disease detects a novel low-frequency variant at the RET locus. <i>European Journal of Human Genetics</i> , 2018 , 26, 561-569 | 5.3 | 11 |

| | | | |
|----|--|------|----|
| 71 | Human cardiac -regulatory elements, their cognate transcription factors, and regulatory DNA sequence variants. <i>Genome Research</i> , 2018 , 28, 1577-1588 | 9.7 | 11 |
| 70 | Testing for colon neoplasia susceptibility variants at the human COX2 locus. <i>Journal of the National Cancer Institute</i> , 2001 , 93, 635-9 | 9.7 | 11 |
| 69 | ViewGene: a graphical tool for polymorphism visualization and characterization. <i>Genome Research</i> , 2002 , 12, 333-8 | 9.7 | 11 |
| 68 | Efficient construction of high-resolution physical maps from yeast artificial chromosomes using radiation hybrids: inner product mapping. <i>Genomics</i> , 1993 , 18, 283-9 | 4.3 | 11 |
| 67 | Segregation analysis of 159 soft tissue sarcoma kindreds: comparison of fixed and sequential sampling schemes. <i>Genetic Epidemiology</i> , 1992 , 9, 291-304 | 2.6 | 11 |
| 66 | Rare variants in fox-1 homolog A (RFX1) are associated with lower blood pressure. <i>PLoS Genetics</i> , 2017 , 13, e1006678 | 6 | 11 |
| 65 | Sequence variation within the fragile X locus. <i>Genome Research</i> , 2001 , 11, 1382-91 | 9.7 | 11 |
| 64 | High incidence of deafness from three frequent connexin 26 mutations in an isolated community. <i>Genetic Testing and Molecular Biomarkers</i> , 2006 , 10, 40-3 | | 10 |
| 63 | Finding needles in haystacks--IRF6 gene variants in isolated cleft lip or cleft palate. <i>New England Journal of Medicine</i> , 2004 , 351, 822-4 | 59.2 | 10 |
| 62 | Dinucleotide repeat (GT) _n markers on chromosome 21. <i>Genomics</i> , 1992 , 14, 818-9 | 4.3 | 10 |
| 61 | Commingling analysis of memory performance in offspring of Alzheimer patients. <i>Genetic Epidemiology</i> , 1992 , 9, 333-45 | 2.6 | 10 |
| 60 | Cardiomyocytes have mosaic patterns of protein expression. <i>Cardiovascular Pathology</i> , 2018 , 34, 50-57 | 3.8 | 9 |
| 59 | Associations between NOS1AP single nucleotide polymorphisms (SNPs) and QT interval duration in four racial/ethnic groups in the Multi-Ethnic Study of Atherosclerosis (MESA). <i>Annals of Noninvasive Electrocardiology</i> , 2013 , 18, 29-40 | 1.5 | 9 |
| 58 | Linkage analysis incorporating gene-age interactions identifies seven novel lipid loci: the Family Blood Pressure Program. <i>Atherosclerosis</i> , 2014 , 235, 84-93 | 3.1 | 9 |
| 57 | GIST: A web tool for collecting gene information. <i>Physiological Genomics</i> , 1999 , 1, 75-81 | 3.6 | 9 |
| 56 | Chromosomal localization of the mouse Src-like adapter protein (Slap) gene and its putative human homolog SLA. <i>Genomics</i> , 1995 , 30, 623-5 | 4.3 | 9 |
| 55 | DNA profile similarity in a subdivided population. <i>Human Heredity</i> , 1994 , 44, 100-9 | 1.1 | 9 |
| 54 | Schizophrenia and porphobilinogen deaminase gene polymorphisms: an association study. <i>Schizophrenia Research</i> , 1992 , 8, 51-8 | 3.6 | 9 |

| | | | |
|----|--|-----|---|
| 53 | Pedigree analysis of blood pressure in subjects from rural Greece and relatives who migrated to Melbourne, Australia. <i>Genetic Epidemiology</i> , 1992 , 9, 225-38 | 2.6 | 9 |
| 52 | A test of nonrandom segregation. <i>Genetic Epidemiology</i> , 1984 , 1, 329-40 | 2.6 | 9 |
| 51 | HPASubC: A suite of tools for user subclassification of human protein atlas tissue images. <i>Journal of Pathology Informatics</i> , 2015 , 6, 36 | 4.4 | 9 |
| 50 | A gene regulatory network explains RET-EDNRB epistasis in Hirschsprung disease. <i>Human Molecular Genetics</i> , 2019 , 28, 3137-3147 | 5.6 | 8 |
| 49 | Information content of the Centre d'Etude du Polymorphisme Humain (CEPH) family structures for linkage studies. <i>Human Genetics</i> , 1991 , 87, 721-4 | 6.3 | 8 |
| 48 | Male and female differential reproductive rate could explain parental transmission asymmetry of mutation origin in Hirschsprung disease. <i>European Journal of Human Genetics</i> , 2012 , 20, 917-20 | 5.3 | 7 |
| 47 | Cloning of rat thymic stromal lymphopoietin receptor (TSLPR) and characterization of genomic structure of murine Tslpr gene. <i>Gene</i> , 2002 , 284, 161-8 | 3.8 | 7 |
| 46 | Linkage mapping of the carbonyl reductase (CBR) gene on human chromosome 21 using a DNA polymorphism in the 3' untranslated region. <i>Genomics</i> , 1992 , 13, 447-8 | 4.3 | 7 |
| 45 | DNA polymorphisms in the 3' untranslated region of genes on human chromosome 21. <i>Genomics</i> , 1993 , 15, 98-102 | 4.3 | 6 |
| 44 | Detection of genetic heterogeneity for complex quantitative phenotypes. <i>Genetic Epidemiology</i> , 1992 , 9, 207-23 | 2.6 | 6 |
| 43 | D21S210: a highly polymorphic (GT) _n marker closely linked to the beta-amyloid protein precursor (APP) gene. <i>Human Genetics</i> , 1993 , 91, 87-8 | 6.3 | 6 |
| 42 | Estimation of segregation and ascertainment probabilities by discarding the single probands. <i>Genetic Epidemiology</i> , 1987 , 4, 185-91 | 2.6 | 6 |
| 41 | Genetic differentiation in the colonising lizard <i>Anolis grahami</i> . <i>Heredity</i> , 1977 , 38, 121-123 | 3.6 | 6 |
| 40 | Rare coding TTN variants are associated with electrocardiographic QT interval in the general population. <i>Scientific Reports</i> , 2016 , 6, 28356 | 4.9 | 5 |
| 39 | Sequence characterization of RET in 117 Chinese Hirschsprung disease families identifies a large burden of de novo and parental mosaic mutations. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 237 | 4.2 | 5 |
| 38 | The role of rare variants in systolic blood pressure: analysis of ExomeChip data in HyperGEN African Americans. <i>Human Heredity</i> , 2015 , 79, 20-7 | 1.1 | 5 |
| 37 | Chronic constipation due to Hirschsprung's disease and desmosis coli in a family. <i>Pediatric Surgery International</i> , 2002 , 18, 110-4 | 2.1 | 5 |
| 36 | Cloning and linkage mapping of three polymorphic tetranucleotide (TAAA) _n repeats on human chromosome 21. <i>Genomics</i> , 1992 , 14, 1071-5 | 4.3 | 5 |

| | | | |
|----|--|------|---|
| 35 | Linkage map on chromosome 21q and the association of a DNA haplotype with a propensity to nondisjunction and trisomy 21. <i>Annals of the New York Academy of Sciences</i> , 1985 , 450, 95-107 | 6.5 | 5 |
| 34 | Gene- and tissue-level interactions in normal gastrointestinal development and Hirschsprung disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019 , | 11.5 | 5 |
| 33 | Widespread promiscuous genetic information transfer from DNA to RNA. <i>Circulation Research</i> , 2011 , 109, 1202-3 | 15.7 | 4 |
| 32 | Ching Chun Li (1912-2003): A Personal Remembrance of a Hero of Genetics. <i>American Journal of Human Genetics</i> , 2004 , 74, 789-792 | 11 | 4 |
| 31 | A somatic cell hybrid map of human chromosome 13. <i>Genomics</i> , 1993 , 18, 486-95 | 4.3 | 4 |
| 30 | Linkage mapping of the AML1 gene on human chromosome 21 using a DNA polymorphism in the 3R untranslated region. <i>Genomics</i> , 1992 , 14, 506-7 | 4.3 | 4 |
| 29 | Testing the Ret and Sema3d genetic interaction in mouse enteric nervous system development. <i>Human Molecular Genetics</i> , 2017 , 26, 1811-1820 | 5.6 | 3 |
| 28 | Gene-by-Psychosocial Factor Interactions Influence Diastolic Blood Pressure in European and African Ancestry Populations: Meta-Analysis of Four Cohort Studies. <i>International Journal of Environmental Research and Public Health</i> , 2017 , 14, | 4.6 | 3 |
| 27 | SNPs and other features as they predispose to complex disease: genome-wide predictive analysis of a quantitative phenotype for hypertension. <i>PLoS ONE</i> , 2011 , 6, e27891 | 3.7 | 3 |
| 26 | 1998 ASHG Award for Excellence in Education. Professor Ching Chun Li, courageous scholar and educator. <i>American Journal of Human Genetics</i> , 1999 , 64, 14-5 | 11 | 3 |
| 25 | Dinucleotide repeat polymorphisms at the D13S192 and D13S193 loci. <i>Human Molecular Genetics</i> , 1993 , 2, 86 | 5.6 | 3 |
| 24 | Combined linkage and association analysis identifies rare and low frequency variants for blood pressure at 1q31. <i>European Journal of Human Genetics</i> , 2019 , 27, 269-277 | 5.3 | 3 |
| 23 | MicroRNA-4516-mediated regulation of relies on 3RUTR-acting variants and contributes to the altered risk of Hirschsprung disease. <i>Journal of Medical Genetics</i> , 2020 , 57, 634-642 | 5.8 | 2 |
| 22 | 2013 William Allan Award: My multifactorial journey. <i>American Journal of Human Genetics</i> , 2014 , 94, 326-333 | | 2 |
| 21 | Hybrids of aneuploid human cancer cells permit complementation of simple and complex cancer defects. <i>Cancer Biology and Therapy</i> , 2009 , 8, 347-55 | 4.6 | 2 |
| 20 | 2008 Presidential Address: Principia Genetica: Our Future Science. <i>American Journal of Human Genetics</i> , 2010 , 86, 302-308 | 11 | 2 |
| 19 | Dinucleotide repeat polymorphism at the DXS1146 locus. <i>Human Molecular Genetics</i> , 1993 , 2, 1078 | 5.6 | 2 |
| 18 | Identity of different mutations for deleterious genes (reply). <i>Nature</i> , 1983 , 301, 176-177 | 50.4 | 2 |

| | | | |
|----|---|------|---|
| 17 | A multi-enhancer regulatory code is disrupted in Hirschsprung disease. <i>Genome Research</i> , 2021 , | 9.7 | 2 |
| 16 | Magnitude of Mendelian versus complex inheritance of rare disorders. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3287-3293 | 2.5 | 2 |
| 15 | 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. <i>American Journal of Hypertension</i> , 2009 , 22, 552-8 | 2.3 | 1 |
| 14 | A PvuII polymorphism detected by a cDNA clone of the gene encoding the human spasmodic protein protein (SML1 gene), one of three members of the trefoil peptide gene family clustered on chromosome 21q22.3. <i>Clinical Genetics</i> , 1997 , 52, 247-8 | 4 | 1 |
| 13 | A genetic map of human chromosome 11p. <i>Genetic Epidemiology</i> , 1986 , 1, 135-40 | 2.6 | 1 |
| 12 | Pathogenic alleles in microtubule, secretory granule and extracellular matrix-related genes in familial keratoconus. <i>Human Molecular Genetics</i> , 2021 , 30, 658-671 | 5.6 | 1 |
| 11 | Multiple, independent, common variants at RET, SEMA3 and NRG1 gut enhancers specify Hirschsprung disease risk in European ancestry subjects. <i>Journal of Pediatric Surgery</i> , 2021 , 56, 2286-2294 | 2.6 | 1 |
| 10 | Commentary: The central questions of human genetics: Richard Lewontin's 1968 senior lecture in Victor McKusick's Bar Harbor short course. <i>International Journal of Epidemiology</i> , 2016 , 45, 668-72 | 7.8 | 1 |
| 9 | Analysis of putative cis-regulatory elements regulating blood pressure variation. <i>Human Molecular Genetics</i> , 2020 , 29, 1922-1932 | 5.6 | 0 |
| 8 | Sequence-based correction of barcode bias in massively parallel reporter assays. <i>Genome Research</i> , 2021 , 31, 1638-1645 | 9.7 | 0 |
| 7 | High Levels of Interest in Reproductive Genetic Information in Parents of Children and Adults With Hirschsprung Disease. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2019 , 69, 299-305 | 2.8 | 0 |
| 6 | Response to Brosens et al. <i>Genetics in Medicine</i> , 2018 , 20, 1479-1480 | 8.1 | |
| 5 | Profile of Mary-Claire King, 2014 Lasker-Koshland Special Achievement in Medical Science awardee. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014 , 111, 17690-2 | 11.5 | |
| 4 | 2011 introduction to Curt Stern Award. <i>American Journal of Human Genetics</i> , 2012 , 90, 405-6 | 11 | |
| 3 | Linkage analysis between Huntington disease and the G8 marker locus. <i>Genetic Epidemiology</i> , 1986 , 1, 211-6 | 2.6 | |
| 2 | Newton E. Morton (1929-2018). <i>American Journal of Human Genetics</i> , 2018 , 102, 1011-1017 | 11 | |
| 1 | Rare coding variants in RCN3 are associated with blood pressure.. <i>BMC Genomics</i> , 2022 , 23, 148 | 4.5 | |