David M Altshuler

List of Publications by Year
in descending order

284	204,193	179	373
papers	citations	152 h-index	281 g-index

305	305	305	168491
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The Genome Analysis Toolkit: A MapReduce framework for analyzing next-generation DNA sequencing data. Genome Research, 2010, 20, 1297-1303.	2.4	21,358
2	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
3	A framework for variation discovery and genotyping using next-generation DNA sequencing data. Nature Genetics, 2011, 43, 491-498.	9.4	10,018
4	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	13.7	9,051
5	PGC-1Î±-responsive genes involved in oxidative phosphorylation are coordinately downregulated in human diabetes. Nature Genetics, 2003, 34, 267-273.	9.4	8,185
6	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	13.7	7,209
7	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
8	The Structure of Haplotype Blocks in the Human Genome. Science, 2002, 296, 2225-2229.	6.0	5,300
9	From FastQ Data to Highâ€Confidence Variant Calls: The Genome Analysis Toolkit Best Practices Pipeline. Current Protocols in Bioinformatics, 2013, 43, 11.10.1-11.10.33.	25.8	4,796
10	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	13.7	4,137
11	Age-Related Clonal Hematopoiesis Associated with Adverse Outcomes. New England Journal of Medicine, 2014, 371, 2488-2498.	13.9	3,474
12	Biological, clinical and population relevance of 95 loci for blood lipids. Nature, 2010, 466, 707-713.	13.7	3,249
13	A map of human genome sequence variation containing 1.42 million single nucleotide polymorphisms. Nature, 2001, 409, 928-933.	13.7	2,794
14	Integrating common and rare genetic variation in diverse human populations. Nature, 2010, 467, 52-58.	13.7	2,625
15	Genome-Wide Association Analysis Identifies Loci for Type 2 Diabetes and Triglyceride Levels. Science, 2007, 316, 1331-1336.	6.0	2,623
16	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	9.4	2,421
17	New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. Nature Genetics, 2010, 42, 105-116.	9.4	1,982
18	Plasma HDL cholesterol and risk of myocardial infarction: a mendelian randomisation study. Lancet, The, 2012, 380, 572-580.	6.3	1,937

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19	Detecting recent positive selection in the human genome from haplotype structure. Nature, 2002, 419, 832-837.	13.7	1,881
20	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. Nature, 2011, 478, 103-109.	13.7	1,855
21	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	13.7	1,788
22	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. Nature Genetics, 2012, 44, 981-990.	9.4	1,748
23	Characterization of single-nucleotide polymorphisms in coding regions of human genes. Nature Genetics, 1999, 22, 231-238.	9.4	1,746
24	Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease. Nature Genetics, 2011, 43, 333-338.	9.4	1,685
25	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. Nature Genetics, 2008, 40, 638-645.	9.4	1,683
26	The common PPARÎ³ Pro12Ala polymorphism is associated with decreased risk of type 2 diabetes. Nature Genetics, 2000, 26, 76-80.	9.4	1,672
27	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. Nature Genetics, 2010, 42, 579-589.	9.4	1,631
28	Efficiency and power in genetic association studies. Nature Genetics, 2005, 37, 1217-1223.	9.4	1,597
29	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. Nature Genetics, 2009, 41, 25-34.	9.4	1,572
30	Evolution and Functional Impact of Rare Coding Variation from Deep Sequencing of Human Exomes. Science, 2012, 337, 64-69.	6.0	1,535
31	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. Nature Genetics, 2009, 41, 703-707.	9.4	1,513
32	Replicating genotypeâ€“phenotype associations. Nature, 2007, 447, 655-660.	13.7	1,509
33	Association between Microdeletion and Microduplication at 16p11.2 and Autism. New England Journal of Medicine, 2008, 358, 667-675.	13.9	1,476
34	Large-scale association analysis identifies new risk loci for coronary artery disease. Nature Genetics, 2013, 45, 25-33.	9.4	1,439
35	Genetic Mapping in Human Disease. Science, 2008, 322, 881-888.	6.0	1,289
36	Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol or triglycerides in humans. Nature Genetics, 2008, 40, 189-197.	9.4	1,286

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37	Common variants at 30 loci contribute to polygenic dyslipidemia. Nature Genetics, 2009, 41, 56-65.	9.4	1,234
38	Genome-wide association study identifies eight loci associated with blood pressure. Nature Genetics, 2009, 41, 666-676.	9.4	1,104
39	Genome-wide meta-analyses identify multiple loci associated with smoking behavior. Nature Genetics, 2010, 42, 441-447.	9.4	1,083
40	Positive Natural Selection in the Human Lineage. Science, 2006, 312, 1614-1620.	6.0	1,037
41	Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. Nature Genetics, 2009, 41, 334-341.	9.4	990
42	Mapping and sequencing of structural variation from eight human genomes. Nature, 2008, 453, 56-64.	13.7	983
43	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. Nature Genetics, 2014, 46, 234-244.	9.4	959
44	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	13.7	952
45	CRISPR-Cas9 Gene Editing for Sickle Cell Disease and Î²-Thalassemia. New England Journal of Medicine, 2021, 384, 252-260.	13.9	939
46	Loss-of-Function Mutations inAPOC3,Triglycerides, and Coronary Disease. New England Journal of Medicine, 2014, 371, 22-31.	13.9	936
47	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. Nature, 2013, 493, 216-220.	13.7	898
48	Integrated detection and population-genetic analysis of SNPs and copy number variation. Nature Genetics, 2008, 40, 1166-1174.	9.4	838
49	Clinical Risk Factors, DNA Variants, and the Development of Type 2 Diabetes. New England Journal of Medicine, 2008, 359, 2220-2232.	13.9	812
50	The Lin28/let-7 Axis Regulates Glucose Metabolism. Cell, 2011, 147, 81-94.	13.5	812
51	TCF7L2Polymorphisms and Progression to Diabetes in the Diabetes Prevention Program. New England Journal of Medicine, 2006, 355, 241-250.	13.9	762
52	A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance. Nature Genetics, 2012, 44, 659-669.	9.4	762
53	Common variants associated with plasma triglycerides and risk for coronary artery disease. Nature Genetics, 2013, 45, 1345-1352.	9.4	754
54	Assessing the impact of population stratification on genetic association studies. Nature Genetics, 2004, 36, 388-393.	9.4	734

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55	TRAF1â€“C5as a Risk Locus for Rheumatoid Arthritis â€” A Genomewide Study. New England Journal of Medicine, 2007, 357, 1199-1209.	13.9	729
56	Integrated genotype calling and association analysis of SNPs, common copy number polymorphisms and rare CNVs. Nature Genetics, 2008, 40, 1253-1260.	9.4	712
57	Estimation of the multiple testing burden for genomewide association studies of nearly all common variants. Genetic Epidemiology, 2008, 32, 381-385.	0.6	699
58	Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. Nature Genetics, 2011, 43, 1066-1073.	9.4	698
59	Copy number variation: New insights in genome diversity. Genome Research, 2006, 16, 949-961.	2.4	697
60	Variants in MTNR1B influence fasting glucose levels. Nature Genetics, 2009, 41, 77-81.	9.4	662
61	An SNP map of the human genome generated by reduced representation shotgun sequencing. Nature, 2000, 407, 513-516.	13.7	658
62	Common deletion polymorphisms in the human genome. Nature Genetics, 2006, 38, 86-92.	9.4	656
63	Common variant in MTNR1B associated with increased risk of type 2 diabetes and impaired early insulin secretion. Nature Genetics, 2009, 41, 82-88.	9.4	642
64	Exome Sequencing,ANGPTL3Mutations, and Familial Combined Hypolipidemia. New England Journal of Medicine, 2010, 363, 2220-2227.	13.9	640
65	ErrÂ and Gabpa/b specify PGC-1Â-dependent oxidative phosphorylation gene expression that is altered in diabetic muscle. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 6570-6575.	3.3	627
66	Multiple regions within 8q24 independently affect risk for prostate cancer. Nature Genetics, 2007, 39, 638-644.	9.4	621
67	Polymorphisms Associated with Cholesterol and Risk of Cardiovascular Events. New England Journal of Medicine, 2008, 358, 1240-1249.	13.9	618
68	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. Diabetes, 2017, 66, 2888-2902.	0.3	615
69	Deletion polymorphism upstream of IRGM associated with altered IRGM expression and Crohn's disease. Nature Genetics, 2008, 40, 1107-1112.	9.4	604
70	A common haplotype of interferon regulatory factor 5 (IRF5) regulates splicing and expression and is associated with increased risk of systemic lupus erythematosus. Nature Genetics, 2006, 38, 550-555.	9.4	593
71	Genetic variation in GIPR influences the glucose and insulin responses to an oral glucose challenge. Nature Genetics, 2010, 42, 142-148.	9.4	591
72	Calibrating a coalescent simulation of human genome sequence variation. Genome Research, 2005, 15, 1576-1583.	2.4	581

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73	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. Nature, 2015, 518, 102-106.	13.7	581
74	Admixture mapping identifies 8q24 as a prostate cancer risk locus in African-American men. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 14068-14073.	3.3	575
75	Common variation in three genes, including a noncoding variant in CFH, strongly influences risk of age-related macular degeneration. Nature Genetics, 2006, 38, 1055-1059.	9.4	570
76	Guilt by association. Nature Genetics, 2000, 26, 135-137.	9.4	569
77	Validating therapeutic targets through human genetics. Nature Reviews Drug Discovery, 2013, 12, 581-594.	21.5	548
78	Genetic variants near TNFAIP3 on 6q23 are associated with systemic lupus erythematosus. Nature Genetics, 2008, 40, 1059-1061.	9.4	534
79	Copy-number variation and association studies of human disease. Nature Genetics, 2007, 39, S37-S42.	9.4	531
80	Testing for an Unusual Distribution of Rare Variants. PLoS Genetics, 2011, 7, e1001322.	1.5	530
81	Parental origin of sequence variants associated with complex diseases. Nature, 2009, 462, 868-874.	13.7	521
82	Two independent alleles at 6q23 associated with risk of rheumatoid arthritis. Nature Genetics, 2007, 39, 1477-1482.	9.4	497
83	Replication of Putative Candidate-Gene Associations with Rheumatoid Arthritis in >4,000 Samples from North America and Sweden: Association of Susceptibility with PTPN22, CTLA4, and PADI4. American Journal of Human Genetics, 2005, 77, 1044-1060.	2.6	494
84	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. Nature Genetics, 2009, 41, 1182-1190.	9.4	481
85	The role of PPAR-Î³ in macrophage differentiation and cholesterol uptake. Nature Medicine, 2001, 7, 41-47.	15.2	476
86	Common variants at CD40 and other loci confer risk of rheumatoid arthritis. Nature Genetics, 2008, 40, 1216-1223.	9.4	476
87	The Metabochip, a Custom Genotyping Array for Genetic Studies of Metabolic, Cardiovascular, and Anthropometric Traits. PLoS Genetics, 2012, 8, e1002793.	1.5	448
88	New susceptibility locus for coronary artery disease on chromosome 3q22.3. Nature Genetics, 2009, 41, 280-282.	9.4	440
89	Sequence variants in SLC16A11 are a common risk factor for type 2 diabetes in Mexico. Nature, 2014, 506, 97-101.	13.7	439
90	Methods for High-Density Admixture Mapping of Disease Genes. American Journal of Human Genetics, 2004, 74, 979-1000.	2.6	437

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91	TXNIP Regulates Peripheral Glucose Metabolism in Humans. PLoS Medicine, 2007, 4, e158.	3.9	435
92	Common Inherited Variation in Mitochondrial Genes Is Not Enriched for Associations with Type 2 Diabetes or Related Glycemic Traits. PLoS Genetics, 2010, 6, e1001058.	1.5	429
93	Three functional variants of IFN regulatory factor 5 (IRF5) define risk and protective haplotypes for human lupus. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 6758-6763.	3.3	428
94	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. Nature Genetics, 2014, 46, 357-363.	9.4	428
95	Demonstrating stratification in a European American population. Nature Genetics, 2005, 37, 868-872.	9.4	424
96	A High-Density Admixture Map for Disease Gene Discovery in African Americans. American Journal of Human Genetics, 2004, 74, 1001-1013.	2.6	416
97	Whole population, genome-wide mapping of hidden relatedness. Genome Research, 2009, 19, 318-326.	2.4	411
98	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	9.4	403
99	Common Variants at 10 Genomic Loci Influence Hemoglobin A1C Levels via Glycemic and Nonglycemic Pathways. Diabetes, 2010, 59, 3229-3239.	0.3	387
100	Choosing Haplotype-Tagging SNPS Based on Unphased Genotype Data Using a Preliminary Sample of Unrelated Subjects with an Example from the Multiethnic Cohort Study. Human Heredity, 2003, 55, 27-36.	0.4	386
101	Inactivating Mutations in NPC1L1 and Protection from Coronary Heart Disease. New England Journal of Medicine, 2014, 371, 2072-2082.	13.9	386
102	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. Science Translational Medicine, 2015, 7, 270ra6.	5.8	375
103	De novo copy number variants identify new genes and loci in isolated sporadic tetralogy of Fallot. Nature Genetics, 2009, 41, 931-935.	9.4	373
104	Identifying Relationships among Genomic Disease Regions: Predicting Genes at Pathogenic SNP Associations and Rare Deletions. PLoS Genetics, 2009, 5, e1000534.	1.5	371
105	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. Nature Genetics, 2015, 47, 1415-1425.	9.4	365
106	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. PLoS Genetics, 2014, 10, e1004494.	1.5	351
107	Detection of regulatory variation in mouse genes. Nature Genetics, 2002, 32, 432-437.	9.4	348
108	Common Single Nucleotide Polymorphisms in TCF7L2 Are Reproducibly Associated With Type 2 Diabetes and Reduce the Insulin Response to Glucose in Nondiabetic Individuals. Diabetes, 2006, 55, 2890-2895.	0.3	346

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109	Comparison of Fine-Scale Recombination Rates in Humans and Chimpanzees. Science, 2005, 308, 107-111.	6.0	335
110	High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. Nature Genetics, 2010, 42, 851-858.	9.4	332
111	Challenges and standards in integrating surveys of structural variation. Nature Genetics, 2007, 39, S7-S15.	9.4	331
112	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. Nature Genetics, 2012, 44, 916-921.	9.4	319
113	Corticosteroid pharmacogenetics: association of sequence variants in CRHR1 with improved lung function in asthmatics treated with inhaled corticosteroids. Human Molecular Genetics, 2004, 13, 1353-1359.	1.4	315
114	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptorâ€“negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	9.4	309
115	The 1000 Genomes Project: data management and community access. Nature Methods, 2012, 9, 459-462.	9.0	308
116	Genetic variants at CD28, PRDM1 and CD2/CD58 are associated with rheumatoid arthritis risk. Nature Genetics, 2009, 41, 1313-1318.	9.4	306
117	Genome-wide association study in Han Chinese identifies four new susceptibility loci for coronary artery disease. Nature Genetics, 2012, 44, 890-894.	9.4	295
118	The multiethnic cohort study: exploring genes, lifestyle and cancer risk. Nature Reviews Cancer, 2004, 4, 519-527.	12.8	290
119	Haplotype Structure and Genotype-Phenotype Correlations of the Sulfonylurea Receptor and the Islet ATP-Sensitive Potassium Channel Gene Region. Diabetes, 2004, 53, 1360-1368.	0.3	284
120	Linkage Disequilibrium and Heritability of Copy-Number Polymorphisms within Duplicated Regions of the Human Genome. American Journal of Human Genetics, 2006, 79, 275-290.	2.6	283
121	THEINHERITEDBASIS OFDIABETESMELLITUS: Implications for the Genetic Analysis of Complex Traits. Annual Review of Genomics and Human Genetics, 2003, 4, 257-291.	2.5	281
122	Human genome sequence variation and the influence of gene history, mutation and recombination. Nature Genetics, 2002, 32, 135-142.	9.4	278
123	Evaluating and improving power in whole-genome association studies using fixed marker sets. Nature Genetics, 2006, 38, 663-667.	9.4	274
124	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 1226-1231.	9.4	270
125	Genome-Wide Association Study for Coronary Artery Calcification With Follow-Up in Myocardial Infarction. Circulation, 2011, 124, 2855-2864.	1.6	269
126	Genome coverage and sequence fidelity of Â29 polymerase-based multiple strand displacement whole genome amplification. Nucleic Acids Research, 2004, 32, e71-e71.	6.5	266

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127	Common Missense Variant in the Glucokinase Regulatory Protein Gene Is Associated With Increased Plasma Triglyceride and C-Reactive Protein but Lower Fasting Glucose Concentrations. Diabetes, 2008, 57, 3112-3121.	0.3	264
128	Modeling and E-M Estimation of Haplotype-Specific Relative Risks from Genotype Data for a Case-Control Study of Unrelated Individuals. Human Heredity, 2003, 55, 179-190.	0.4	249
129	Exome sequencing of 20,791Âcases of type 2 diabetes and 24,440Âcontrols. Nature, 2019, 570, 71-76.	13.7	248
130	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	3.8	242
131	Large-Scale Gene-Centric Meta-Analysis across 39 Studies Identifies Type 2 Diabetes Loci. American Journal of Human Genetics, 2012, 90, 410-425.	2.6	239
132	Common Variants in 40 Genes Assessed for Diabetes Incidence and Response to Metformin and Lifestyle Intervention in the Diabetes Prevention Program. Diabetes, 2010, 59, 2672-2681.	0.3	234
133	Association of a Low-Frequency Variant inHNF1AWith Type 2 Diabetes in a Latino Population. JAMA - Journal of the American Medical Association, 2014, 311, 2305.	3.8	230
134	Transferability of tag SNPs in genetic association studies in multiple populations. Nature Genetics, 2006, 38, 1298-1303.	9.4	224
135	Genome-wide meta-analysis in alopecia areata resolves HLA associations and reveals two new susceptibility loci. Nature Communications, 2015, 6, 5966.	5.8	213
136	Exome sequencing of extreme phenotypes identifies DCTN4 as a modifier of chronic Pseudomonas aeruginosa infection in cystic fibrosis. Nature Genetics, 2012, 44, 886-889.	9.4	211
137	Prospective functional classification of all possible missense variants in PPARG. Nature Genetics, 2016, 48, 1570-1575.	9.4	210
138	Genetic Analysis of Human Traits In Vitro: Drug Response and Gene Expression in Lymphoblastoid Cell Lines. PLoS Genetics, 2008, 4, e1000287.	1.5	200
139	A Genome-Wide Association Search for Type 2 Diabetes Genes in African Americans. PLoS ONE, 2012, 7, e29202.	1.1	197
140	Polymorphism at the TNF superfamily gene TNFSF4 confers susceptibility to systemic lupus erythematosus. Nature Genetics, 2008, 40, 83-89.	9.4	193
141	Whole-Exome Sequencing Identifies Rare and Low-Frequency Coding Variants Associated with LDL Cholesterol. American Journal of Human Genetics, 2014, 94, 233-245.	2.6	193
142	The Case for Selection at CCR5-Î”32. PLoS Biology, 2005, 3, e378.	2.6	190
143	Quality and completeness of SNP databases. Nature Genetics, 2003, 33, 457-458.	9.4	182
144	Evaluation of Common Variants in the Six Known Maturity-Onset Diabetes of the Young (MODY) Genes for Association With Type 2 Diabetes. Diabetes, 2007, 56, 685-693.	0.3	178

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145	Completing the map of human genetic variation. Nature, 2007, 447, 161-165.	13.7	178
146	Genomewide Linkage Analysis of Stature in Multiple Populations Reveals Several Regions with Evidence of Linkage to Adult Height. American Journal of Human Genetics, 2001, 69, 106-116.	2.6	177
147	Comprehensive Association Testing of Common Mitochondrial DNA Variation in Metabolic Disease. American Journal of Human Genetics, 2006, 79, 54-61.	2.6	173
148	The functional spectrum of low-frequency coding variation. Genome Biology, 2011, 12, R84.	13.9	173
149	Updated Genetic Score Based on 34 Confirmed Type 2 Diabetes Loci Is Associated With Diabetes Incidence and Regression to Normoglycemia in the Diabetes Prevention Program. Diabetes, 2011, 60, 1340-1348.	0.3	172
150	Consistent Association of Type 2 Diabetes Risk Variants Found in Europeans in Diverse Racial and Ethnic Groups. PLoS Genetics, 2010, 6, e1001078.	1.5	168
151	Once and Againâ€”Issues Surrounding Replication in Genetic Association Studies. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 4438-4441.	1.8	166
152	Longâ€term effects of the Diabetes Prevention Program interventions on cardiovascular risk factors: a report from the DPP Outcomes Study. Diabetic Medicine, 2013, 30, 46-55.	1.2	166
153	5' Flanking Variants of Resistin Are Associated With Obesity. Diabetes, 2002, 51, 1629-1634.	0.3	158
154	A candidate gene approach to searching for low-penetrance breast and prostate cancer genes. Nature Reviews Cancer, 2005, 5, 977-985.	12.8	152
155	Rare variants in PPARG with decreased activity in adipocyte differentiation are associated with increased risk of type 2 diabetes. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 13127-13132.	3.3	152
156	IGF2BP2/IMP2-Deficient Mice Resist Obesity through Enhanced Translation of Ucp1 mRNA and Other mRNAs Encoding Mitochondrial Proteins. Cell Metabolism, 2015, 21, 609-621.	7.2	148
157	Evaluating empirical bounds on complex disease genetic architecture. Nature Genetics, 2013, 45, 1418-1427.	9.4	147
158	Variations in the G6PC2/ABCB11 genomic region are associated with fasting glucose levels. Journal of Clinical Investigation, 2008, 118, 2620-8.	3.9	146
159	A comprehensive haplotype analysis of CYP19 and breast cancer risk: the Multiethnic Cohort. Human Molecular Genetics, 2003, 12, 2679-2692.	1.4	144
160	Role for Msh5 in the regulation of Ig class switch recombination. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 7193-7198.	3.3	142
161	Increased Burden of Cardiovascular Disease in Carriers of APOL1 Genetic Variants. Circulation Research, 2014, 114, 845-850.	2.0	141
162	Genetic Variation at the CYP19A1 Locus Predicts Circulating Estrogen Levels but not Breast Cancer Risk in Postmenopausal Women. Cancer Research, 2007, 67, 1893-1897.	0.4	140

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163	Guilt beyond a reasonable doubt. Nature Genetics, 2007, 39, 813-815.	9.4	140
164	Accurately Assessing the Risk of Schizophrenia Conferred by Rare Copy-Number Variation Affecting Genes with Brain Function. PLoS Genetics, 2010, 6, e1001097.	1.5	134
165	A Genome-Wide Association Study Identifies LIPA as a Susceptibility Gene for Coronary Artery Disease. Circulation: Cardiovascular Genetics, 2011, 4, 403-412.	5.1	130
166	Assessing the phenotypic effects in the general population of rare variants in genes for a dominant Mendelian form of diabetes. Nature Genetics, 2013, 45, 1380-1385.	9.4	129
167	The Power of Gene-Based Rare Variant Methods to Detect Disease-Associated Variation and Test Hypotheses About Complex Disease. PLoS Genetics, 2015, 11, e1005165.	1.5	124
168	Effects of the Type 2 Diabetes-AssociatedPPARGP12A Polymorphism on Progression to Diabetes and Response to Troglitazone. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 1502-1509.	1.8	122
169	Type 2 Diabetes Variants Disrupt Function of SLC16A11 through Two Distinct Mechanisms. Cell, 2017, 170, 199-212.e20.	13.5	121
170	Common SNPs in HMGCR in Micronesians and Whites Associated With LDL-Cholesterol Levels Affect Alternative Splicing of Exon13. Arteriosclerosis, Thrombosis, and Vascular Biology, 2008, 28, 2078-2084.	1.1	120
171	Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. Nature Genetics, 2015, 47, 921-925.	9.4	120
172	Burden of Rare Sarcomere Gene Variants in the Framingham and Jackson Heart Study Cohorts. American Journal of Human Genetics, 2012, 91, 513-519.	2.6	116
173	Type 2 Diabetesâ€“Associated Missense Polymorphisms KCNJ11 E23K and ABCC8 A1369S Influence Progression to Diabetes and Response to Interventions in the Diabetes Prevention Program. Diabetes, 2007, 56, 531-536.	0.3	115
174	Leveraging Cross-Species Transcription Factor Binding Site Patterns: From Diabetes Risk Loci to Disease Mechanisms. Cell, 2014, 156, 343-358.	13.5	113
175	Common Genetic Variation in IGF1 and Prostate Cancer Risk in the Multiethnic Cohort. Journal of the National Cancer Institute, 2006, 98, 123-134.	3.0	107
176	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	1.5	105
177	Genetic Polymorphisms and Disease. New England Journal of Medicine, 1998, 338, 1626-1626.	13.9	100
178	Tissue-specific alternative splicing of TCF7L2. Human Molecular Genetics, 2009, 18, 3795-3804.	1.4	100
179	Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes. Nature Communications, 2018, 9, 2252.	5.8	99
180	Power in the phenotypic extremes: a simulation study of power in discovery and replication of rare variants. Genetic Epidemiology, 2011, 35, 236-246.	0.6	97

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181	High-Throughput Luminescent Reporter of Insulin Secretion for Discovering Regulators of Pancreatic Beta-Cell Function. Cell Metabolism, 2015, 21, 126-137.	7.2	97
182	Exome Sequencing and Genome-Wide Linkage Analysis in 17 Families Illustrate the Complex Contribution of TTN Truncating Variants to Dilated Cardiomyopathy. Circulation: Cardiovascular Genetics, 2013, 6, 144-153.	5.1	95
183	Identification and Functional Characterization of G6PC2 Coding Variants Influencing Glycemic Traits Define an Effector Transcript at the G6PC2-ABCB11 Locus. PLoS Genetics, 2015, 11, e1004876.	1.5	95
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