David M Altshuler

List of Publications by Year in Descending Order

280 papers	160,064 citations	147 h-index	305 g-index
305 ext. papers	187,727 ext. citations	21.9 avg, IF	7.65 L-index

#	Paper	IF	Citations
280	CRISPR-Cas9 Gene Editing for Sickle Cell Disease and β-Thalassemia. *New England Journal of Medicine*, **2021**, 384, 252-260	59.2	292
279	Sex-dimorphic genetic effects and novel loci for fasting glucose and insulin variability. *Nature Communications*, **2021**, 12, 24	17.4	30
278	Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls. *Nature*, **2019**, 570, 71-76	50.4	129
277	Case-control analysis identifies shared properties of rare germline variation in cancer predisposing genes. *European Journal of Human Genetics*, **2019**, 27, 824-828	5.3	1
276	Evaluating the contribution of rare variants to type 2 diabetes and related traits using pedigrees. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 379-384	11.5	21
275	Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes. *Nature Communications*, **2018**, 9, 2252	17.4	71
274	Variation in Maturity-Onset Diabetes of the Young Genes Influence Response to Interventions for Diabetes Prevention. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 2678-2689	5.6	12
273	Functional Investigations of HNF1A Identify Rare Variants as Risk Factors for Type 2 Diabetes in the General Population. *Diabetes*, **2017**, 66, 335-346	0.9	35
272	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes*, **2017**, 66, 2888-2902	8.1	414
271	A Loss-of-Function Splice Acceptor Variant in Is Protective for Type 2 Diabetes. *Diabetes*, **2017**, 66, 2903-2914	8.1	32
270	Type 2 Diabetes Variants Disrupt Function of SLC16A11 through Two Distinct Mechanisms. *Cell*, **2017**, 170, 199-212.e20	56.2	94
269	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Scientific Data*, **2017**, 4, 170179	8.2	22
268	Genome Sequencing of Multiple Primary Tumors Reveals a Novel PALB2 Variant. *Journal of Clinical Oncology*, **2016**, 34, e61-7	2.2	6
267	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
266	Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, **2016**, 536, 285-91	50.4	6940
265	Association of Exome Sequences With Cardiovascular Traits Among Blacks in the Jackson Heart Study. *Circulation: Cardiovascular Genetics*, **2016**, 9, 368-74		7
264	Prospective functional classification of all possible missense variants in PPARG. *Nature Genetics*, **2016**, 48, 1570-1575	36.3	149

263	A null mutation in ANGPTL8 does not associate with either plasma glucose or type 2 diabetes in humans. *BMC Endocrine Disorders*, **2016**, 16, 7	3.3	8
262	Epilogue: What the Future Holds: Genomic Medicine at the Heart of Diabetes Management **2016**, 561-570		
261	Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. *European Journal of Human Genetics*, **2016**, 24, 1181-7	5.3	2
260	Transancestral fine-mapping of four type 2 diabetes susceptibility loci highlights potential causal regulatory mechanisms. *Human Molecular Genetics*, **2016**, 25, 2070-2081	5.6	20
259	Genetics of Endocrinology **2016**, 49-68		1
258	The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47	50.4	704
257	Guidelines for Large-Scale Sequence-Based Complex Trait Association Studies: Lessons Learned from the NHLBI Exome Sequencing Project. *American Journal of Human Genetics*, **2016**, 99, 791-801	11	67
256	Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. *Nature Genetics*, **2015**, 47, 921-5	36.3	92
255	The power of gene-based rare variant methods to detect disease-associated variation and test hypotheses about complex disease. *PLoS Genetics*, **2015**, 11, e1005165	6	98
254	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	6	76
253	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. *Science Translational Medicine*, **2015**, 7, 270ra6	17.5	267
252	IGF2BP2/IMP2-Deficient mice resist obesity through enhanced translation of Ucp1 mRNA and Other mRNAs encoding mitochondrial proteins. *Cell Metabolism*, **2015**, 21, 609-21	24.6	87
251	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
250	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. *Nature Genetics*, **2015**, 47, 1415-25	36.3	292
249	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. *Nature*, **2015**, 518, 102-6	50.4	463
248	Pathways targeted by antidiabetes drugs are enriched for multiple genes associated with type 2 diabetes risk. *Diabetes*, **2015**, 64, 1470-83	0.9	28
247	Genome-wide meta-analysis in alopecia areata resolves HLA associations and reveals two new susceptibility loci. *Nature Communications*, **2015**, 6, 5966	17.4	142
246	High-throughput luminescent reporter of insulin secretion for discovering regulators of pancreatic Beta-cell function. *Cell Metabolism*, **2015**, 21, 126-37	24.6	58

245	Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol. *American Journal of Human Genetics*, **2014**, 94, 233-45	11	170
244	Sequence variants in SLC16A11 are a common risk factor for type 2 diabetes in Mexico. *Nature*, **2014**, 506, 97-101	50.4	323
243	Leveraging cross-species transcription factor binding site patterns: from diabetes risk loci to disease mechanisms. *Cell*, **2014**, 156, 343-58	56.2	96
242	Inactivating mutations in NPC1L1 and protection from coronary heart disease. *New England Journal of Medicine*, **2014**, 371, 2072-82	59.2	307
241	A novel test for recessive contributions to complex diseases implicates Bardet-Biedl syndrome gene BBS10 in idiopathic type 2 diabetes and obesity. *American Journal of Human Genetics*, **2014**, 95, 509-20	11	26
240	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
239	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. *Nature Genetics*, **2014**, 46, 234-44	36.3	784
238	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. *Nature Genetics*, **2014**, 46, 357-63	36.3	351
237	Loss-of-function mutations in APOC3, triglycerides, and coronary disease. *New England Journal of Medicine*, **2014**, 371, 22-31	59.2	721
236	Simulation of Finnish population history, guided by empirical genetic data, to assess power of rare-variant tests in Finland. *American Journal of Human Genetics*, **2014**, 94, 710-20	11	19
235	Genetic modifiers of EGFR dependence in non-small cell lung cancer. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 18661-6	11.5	37
234	Increased burden of cardiovascular disease in carriers of APOL1 genetic variants. *Circulation Research*, **2014**, 114, 845-50	15.7	119
233	Whole-exome imputation of sequence variants identified two novel alleles associated with adult body height in African Americans. *Human Molecular Genetics*, **2014**, 23, 6607-15	5.6	11
232	Distribution and medical impact of loss-of-function variants in the Finnish founder population. *PLoS Genetics*, **2014**, 10, e1004494	6	243
231	Age-related clonal hematopoiesis associated with adverse outcomes. *New England Journal of Medicine*, **2014**, 371, 2488-98	59.2	2314
230	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, 13127-32	11.5	121
229	Association of a low-frequency variant in HNF1A with type 2 diabetes in a Latino population. *JAMA - Journal of the American Medical Association*, **2014**, 311, 2305-14	27.4	164
228	Clonal Hematopoiesis with Somatic Mutations Is a Common, Age-Related Condition Associated with Adverse Outcomes. *Blood*, **2014**, 124, 840-840	2.2	0

227	Validating therapeutic targets through human genetics. *Nature Reviews Drug Discovery*, **2013**, 12, 581-94	64.1	405
226	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. *Nature Genetics*, **2013**, 45, 1226-1231	36.3	205
225	Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nature Genetics*, **2013**, 45, 1345-52	36.3	597
224	Evaluating empirical bounds on complex disease genetic architecture. *Nature Genetics*, **2013**, 45, 1418-27	36.3	120
223	Assessing the phenotypic effects in the general population of rare variants in genes for a dominant Mendelian form of diabetes. *Nature Genetics*, **2013**, 45, 1380-5	36.3	103
222	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, 144-53		81
221	Large-scale association analysis identifies new risk loci for coronary artery disease. *Nature Genetics*, **2013**, 45, 25-33	36.3	1172
220	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. *Nature*, **2013**, 493, 216-20	50.4	723
219	Confirming glycemic status in the Diabetes Prevention Program: implications for diagnosing diabetes in high risk adults. *Journal of Diabetes and Its Complications*, **2013**, 27, 150-7	3.2	14
218	From FastQ data to high confidence variant calls: the Genome Analysis Toolkit best practices pipeline. *Current Protocols in Bioinformatics*, **2013**, 43, 11.10.1-11.10.33	24.2	2907
217	Branched chain and aromatic amino acids change acutely following two medical therapies for type 2 diabetes mellitus. *Metabolism: Clinical and Experimental*, **2013**, 62, 1772-8	12.7	48
216	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190
215	Long-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	3.5	136
214	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
213	Exome sequencing and directed clinical phenotyping diagnose cholesterol ester storage disease presenting as autosomal recessive hypercholesterolemia. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2013**, 33, 2909-14	9.4	76
212	Genetic loci for retinal arteriolar microcirculation. *PLoS ONE*, **2013**, 8, e65804	3.7	19
211	Fine-scale patterns of population stratification confound rare variant association tests. *PLoS ONE*, **2013**, 8, e65834	3.7	35
210	Plasma HDL cholesterol and risk of myocardial infarction: a mendelian randomisation study. *Lancet, The*, **2012**, 380, 572-80	40	1523

209	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. *Nature Genetics*, **2012**, 44, 981-90	36.3	1482
208	The 1000 Genomes Project: data management and community access. *Nature Methods*, **2012**, 9, 459-62	21.6	202
207	Exome sequencing of extreme phenotypes identifies DCTN4 as a modifier of chronic Pseudomonas aeruginosa infection in cystic fibrosis. *Nature Genetics*, **2012**, 44, 886-9	36.3	170
206	An integrated map of genetic variation from 1,092 human genomes. *Nature*, **2012**, 491, 56-65	50.4	6049
205	Burden of rare sarcomere gene variants in the Framingham and Jackson Heart Study cohorts. *American Journal of Human Genetics*, **2012**, 91, 513-9	11	88
204	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. *Nature Genetics*, **2012**, 44, 916-21	36.3	257
203	African ancestry and its correlation to type 2 diabetes in African Americans: a genetic admixture analysis in three U.S. population cohorts. *PLoS ONE*, **2012**, 7, e32840	3.7	56
202	A genome-wide association search for type 2 diabetes genes in African Americans. *PLoS ONE*, **2012**, 7, e29202	3.7	138
201	The Inherited Basis of Common Diseases **2012**, 195-198		
200	A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance. *Nature Genetics*, **2012**, 44, 659-69	36.3	615
199	Evolution and functional impact of rare coding variation from deep sequencing of human exomes. *Science*, **2012**, 337, 64-9	33.3	1280
198	Genome-wide association study in Han Chinese identifies four new susceptibility loci for coronary artery disease. *Nature Genetics*, **2012**, 44, 890-4	36.3	243
197	Large-scale gene-centric meta-analysis across 39 studies identifies type 2 diabetes loci. *American Journal of Human Genetics*, **2012**, 90, 410-25	11	214
196	2011 Curt Stern Award address. *American Journal of Human Genetics*, **2012**, 90, 407-9	11	2
195	Large-Scale Gene-Centric Meta-Analysis across 39 Studies Identifies Type 2 Diabetes Loci. *American Journal of Human Genetics*, **2012**, 90, 753	11	4
194	Informed conditioning on clinical covariates increases power in case-control association studies. *PLoS Genetics*, **2012**, 8, e1003032	6	58
193	The metabochip, a custom genotyping array for genetic studies of metabolic, cardiovascular, and anthropometric traits. *PLoS Genetics*, **2012**, 8, e1002793	6	395
192	Analysis of case-control association studies with known risk variants. *Bioinformatics*, **2012**, 28, 1729-37	7.2	25

191	Efficiency and power as a function of sequence coverage, SNP array density, and imputation. *PLoS Computational Biology*, **2012**, 8, e1002604	5	17
190	Genetic modulation of lipid profiles following lifestyle modification or metformin treatment: the Diabetes Prevention Program. *PLoS Genetics*, **2012**, 8, e1002895	6	27
189	Risk and return for the clinician-investigator. *Science Translational Medicine*, **2012**, 4, 135cm6	17.5	3
188	Hypothesis-based analysis of gene-gene interactions and risk of myocardial infarction. *PLoS ONE*, **2012**, 7, e41730	3.7	15
187	Influence of 9p21.3 genetic variants on clinical and angiographic outcomes in early-onset myocardial infarction. *Journal of the American College of Cardiology*, **2011**, 58, 426-34	15.1	54
186	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. *Nature* , **2011**, 478, 103-9	50.4	1564
185	Comparing strategies to fine-map the association of common SNPs at chromosome 9p21 with type 2 diabetes and myocardial infarction. *Nature Genetics*, **2011**, 43, 801-5	36.3	75
184	Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. *Nature Genetics*, **2011**, 43, 1066-73	36.3	584
183	The Lin28/let-7 axis regulates glucose metabolism. *Cell*, **2011**, 147, 81-94	56.2	649
182	The functional spectrum of low-frequency coding variation. *Genome Biology*, **2011**, 12, R84	18.3	161
181	Strong association of the APOA5-1131T>C gene variant and early-onset acute myocardial infarction. *Atherosclerosis*, **2011**, 214, 397-403	3.1	36
180	Testing for an unusual distribution of rare variants. *PLoS Genetics*, **2011**, 7, e1001322	6	465
179	Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease. *Nature Genetics*, **2011**, 43, 333-8	36.3	1394
178	A framework for variation discovery and genotyping using next-generation DNA sequencing data. *Nature Genetics*, **2011**, 43, 491-8	36.3	7264
177	Genome-wide association study for coronary artery calcification with follow-up in myocardial infarction. *Circulation*, **2011**, 124, 2855-64	16.7	213
176	A genome-wide association study identifies LIPA as a susceptibility gene for coronary artery disease. *Circulation: Cardiovascular Genetics*, **2011**, 4, 403-12		98
175	DASH: a method for identical-by-descent haplotype mapping uncovers association with recent variation. *American Journal of Human Genetics*, **2011**, 88, 706-717	11	60
174	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15

173	Power in the phenotypic extremes: a simulation study of power in discovery and replication of rare variants. *Genetic Epidemiology*, **2011**, 35, 236-46	2.6	83
172	Triglyceride response to an intensive lifestyle intervention is enhanced in carriers of the GCKR Pro446Leu polymorphism. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E1142-7	5.6	32
171	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, 1340-8	0.9	153
170	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
169	Increased power of mixed models facilitates association mapping of 10 loci for metabolic traits in an isolated population. *Human Molecular Genetics*, **2011**, 20, 827-39	5.6	22
168	European admixture on the Micronesian island of Kosrae: lessons from complete genetic information. *European Journal of Human Genetics*, **2010**, 18, 309-16	5.3	10
167	Biological, clinical and population relevance of 95 loci for blood lipids. *Nature*, **2010**, 466, 707-13	50.4	2742
166	Integrating common and rare genetic variation in diverse human populations. *Nature*, **2010**, 467, 52-8	50.4	2135
165	A map of human genome variation from population-scale sequencing. *Nature*, **2010**, 467, 1061-73	50.4	6142
164	Genetic variation in GIPR influences the glucose and insulin responses to an oral glucose challenge. *Nature Genetics*, **2010**, 42, 142-8	36.3	527
163	Genome-wide meta-analyses identify multiple loci associated with smoking behavior. *Nature Genetics*, **2010**, 42, 441-7	36.3	927
162	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. *Nature Genetics*, **2010**, 42, 579-89	36.3	1449
161	High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. *Nature Genetics*, **2010**, 42, 851-8	36.3	292
160	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. *Nature Genetics*, **2010**, 42, 885-92	36.3	276
159	Common variants at 10 genomic loci influence hemoglobin A(C) levels via glycemic and nonglycemic pathways. *Diabetes*, **2010**, 59, 3229-39	0.9	314
158	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
157	Common inherited variation in mitochondrial genes is not enriched for associations with type 2 diabetes or related glycemic traits. *PLoS Genetics*, **2010**, 6, e1001058	6	366
156	Consistent association of type 2 diabetes risk variants found in europeans in diverse racial and ethnic groups. *PLoS Genetics*, **2010**, 6, e1001078	6	142

155	A large study of androgen receptor germline variants and their relation to sex hormone levels and prostate cancer risk. Results from the National Cancer Institute Breast and Prostate Cancer Cohort Consortium. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, E121-7	5.6	42
154	The HapMap Project and Low-Penetrance Cancer Susceptibility Alleles **2010**, 195-204		
153	Lack of association between the Trp719Arg polymorphism in kinesin-like protein-6 and coronary artery disease in 19 case-control studies. *Journal of the American College of Cardiology*, **2010**, 56, 1552-63	15.1	75
152	The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. *Genome Research*, **2010**, 20, 1297-303	9.7	14079
151	New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. *Nature Genetics*, **2010**, 42, 105-16	36.3	1673
150	Common variants in 40 genes assessed for diabetes incidence and response to metformin and lifestyle intervention in the diabetes prevention program. *Diabetes*, **2010**, 59, 2672-81	0.9	200
149	Exome sequencing, ANGPTL3 mutations, and familial combined hypolipidemia. *New England Journal of Medicine*, **2010**, 363, 2220-7	59.2	485
148	Accurately assessing the risk of schizophrenia conferred by rare copy-number variation affecting genes with brain function. *PLoS Genetics*, **2010**, 6, e1001097	6	118
147	The T-381C SNP in BNP gene may be modestly associated with type 2 diabetes: an updated meta-analysis in 49 279 subjects. *Human Molecular Genetics*, **2009**, 18, 2495-501	5.6	26
146	Underlying genetic models of inheritance in established type 2 diabetes associations. *American Journal of Epidemiology*, **2009**, 170, 537-45	3.8	60
145	Systematic haplotype analysis resolves a complex plasma plant sterol locus on the Micronesian Island of Kosrae. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 13886-91	11.5	20
144	Identifying relationships among genomic disease regions: predicting genes at pathogenic SNP associations and rare deletions. *PLoS Genetics*, **2009**, 5, e1000534	6	337
143	Genome-wide association studies in an isolated founder population from the Pacific Island of Kosrae. *PLoS Genetics*, **2009**, 5, e1000365	6	75
142	Parental origin of sequence variants associated with complex diseases. *Nature*, **2009**, 462, 868-74	50.4	459
141	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. *Nature Genetics*, **2009**, 41, 25-34	36.3	1368
140	Common variant in MTNR1B associated with increased risk of type 2 diabetes and impaired early insulin secretion. *Nature Genetics*, **2009**, 41, 82-8	36.3	550
139	Variants in MTNR1B influence fasting glucose levels. *Nature Genetics*, **2009**, 41, 77-81	36.3	584
138	Common variants at 30 loci contribute to polygenic dyslipidemia. *Nature Genetics*, **2009**, 41, 56-65	36.3	1095

137	New susceptibility locus for coronary artery disease on chromosome 3q22.3. *Nature Genetics*, **2009**, 41, 280-2	36.3	389
136	Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. *Nature Genetics*, **2009**, 41, 334-41	36.3	884
135	Genome-wide association study identifies eight loci associated with blood pressure. *Nature Genetics*, **2009**, 41, 666-76	36.3	970
134	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. *Nature Genetics*, **2009**, 41, 703-7	36.3	1298
133	De novo copy number variants identify new genes and loci in isolated sporadic tetralogy of Fallot. *Nature Genetics*, **2009**, 41, 931-5	36.3	325
132	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. *Nature Genetics*, **2009**, 41, 1182-90	36.3	433
131	Genetic variants at CD28, PRDM1 and CD2/CD58 are associated with rheumatoid arthritis risk. *Nature Genetics*, **2009**, 41, 1313-8	36.3	272
130	Donor-recipient mismatch for common gene deletion polymorphisms in graft-versus-host disease. *Nature Genetics*, **2009**, 41, 1341-4	36.3	75
129	Tissue-specific alternative splicing of TCF7L2. *Human Molecular Genetics*, **2009**, 18, 3795-804	5.6	89
128	Genome-wide association study of electrocardiographic conduction measures in an isolated founder population: Kosrae. *Heart Rhythm*, **2009**, 6, 634-41	6.7	56
127	Whole population, genome-wide mapping of hidden relatedness. *Genome Research*, **2009**, 19, 318-26	9.7	326
126	Mapping and sequencing of structural variation from eight human genomes. *Nature*, **2008**, 453, 56-64	50.4	878
125	Genetic variants near TNFAIP3 on 6q23 are associated with systemic lupus erythematosus. *Nature Genetics*, **2008**, 40, 1059-61	36.3	459
124	Polymorphism at the TNF superfamily gene TNFSF4 confers susceptibility to systemic lupus erythematosus. *Nature Genetics*, **2008**, 40, 83-9	36.3	167
123	Deletion polymorphism upstream of IRGM associated with altered IRGM expression and Crohn© disease. *Nature Genetics*, **2008**, 40, 1107-12	36.3	527
122	Common variants at CD40 and other loci confer risk of rheumatoid arthritis. *Nature Genetics*, **2008**, 40, 1216-23	36.3	416
121	Integrated genotype calling and association analysis of SNPs, common copy number polymorphisms and rare CNVs. *Nature Genetics*, **2008**, 40, 1253-60	36.3	656
120	Integrated detection and population-genetic analysis of SNPs and copy number variation. *Nature Genetics*, **2008**, 40, 1166-74	36.3	773

119	Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol or triglycerides in humans. *Nature Genetics*, **2008**, 40, 189-97	36.3	1108
118	Polymorphisms associated with cholesterol and risk of cardiovascular events. *New England Journal of Medicine*, **2008**, 358, 1240-9	59.2	534
117	Clinical risk factors, DNA variants, and the development of type 2 diabetes. *New England Journal of Medicine*, **2008**, 359, 2220-32	59.2	698
116	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. *Nature Genetics*, **2008**, 40, 638-45	36.3	1496
115	From Darwin's finches to canaries in the coal mine--mining the genome for new biology. *New England Journal of Medicine*, **2008**, 358, 2760-3	59.2	63
114	Genetic mapping in human disease. *Science*, **2008**, 322, 881-8	33.3	1086
113	Extension of type 2 diabetes genome-wide association scan results in the diabetes prevention program. *Diabetes*, **2008**, 57, 2503-10	0.9	86
112	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, 3112-21	0.9	223
111	Genetic analysis of human traits in vitro: drug response and gene expression in lymphoblastoid cell lines. *PLoS Genetics*, **2008**, 4, e1000287	6	182
110	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, 2078-84	9.4	106
109	A novel polymorphism of the human CD40 receptor with enhanced function. *Blood*, **2008**, 112, 1863-71	2.2	25
108	Haplotypes of the estrogen receptor beta gene and breast cancer risk. *International Journal of Cancer*, **2008**, 122, 387-92	7.5	36
107	Estimation of the multiple testing burden for genomewide association studies of nearly all common variants. *Genetic Epidemiology*, **2008**, 32, 381-5	2.6	584
106	Association between microdeletion and microduplication at 16p11.2 and autism. *New England Journal of Medicine*, **2008**, 358, 667-75	59.2	1249
105	Variations in the G6PC2/ABCB11 genomic region are associated with fasting glucose levels. *Journal of Clinical Investigation*, **2008**, 118, 2620-8	15.9	127
104	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, 685-93	0.9	160
103	CYP17 genetic variation and risk of breast and prostate cancer from the National Cancer Institute Breast and Prostate Cancer Cohort Consortium (BPC3). *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 2237-46	4	49
102	Type 2 diabetes-associated missense polymorphisms KCNJ11 E23K and ABCC8 A1369S influence progression to diabetes and response to interventions in the Diabetes Prevention Program. *Diabetes*, **2007**, 56, 531-6	0.9	105

101	Estimation of the Multiple Testing Burden for Genomewide Association Studies of Common Variants. *Nature Precedings*, **2007**,		2
100	Genetic and functional analysis of CHEK2 (CHK2) variants in multiethnic cohorts. *International Journal of Cancer*, **2007**, 121, 2661-7	7.5	62
99	A comprehensive analysis of common genetic variation in prolactin (PRL) and PRL receptor (PRLR) genes in relation to plasma prolactin levels and breast cancer risk: the multiethnic cohort. *BMC Medical Genetics*, **2007**, 8, 72	2.1	32
98	Completing the map of human genetic variation. *Nature*, **2007**, 447, 161-5	50.4	153
97	Replicating genotype-phenotype associations. *Nature*, **2007**, 447, 655-60	50.4	1363
96	Two independent alleles at 6q23 associated with risk of rheumatoid arthritis. *Nature Genetics*, **2007**, 39, 1477-82	36.3	449
95	Multiple regions within 8q24 independently affect risk for prostate cancer. *Nature Genetics*, **2007**, 39, 638-44	36.3	563
94	Copy-number variation and association studies of human disease. *Nature Genetics*, **2007**, 39, S37-42	36.3	464
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