Goncalo Abecasis

List of Publications by Citations

386 papers	157,913 citations	144 h-index	397 g-index
424 ext. papers	198,271 ext. citations	17.7 avg, IF	7.91 L-index

#	Paper	IF	Citations
386	The Sequence Alignment/Map format and SAMtools. *Bioinformatics*, **2009**, 25, 2078-9	7.2	30805
385	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
384	The variant call format and VCFtools. *Bioinformatics*, **2011**, 27, 2156-8	7.2	6200
383	A map of human genome variation from population-scale sequencing. *Nature*, **2010**, 467, 1061-73	50.4	6142
382	An integrated map of genetic variation from 1,092 human genomes. *Nature*, **2012**, 491, 56-65	50.4	6049
381	A second generation human haplotype map of over 3.1 million SNPs. *Nature*, **2007**, 449, 851-61	50.4	3647
380	Merlin--rapid analysis of dense genetic maps using sparse gene flow trees. *Nature Genetics*, **2002**, 30, 97-101	36.3	2878
379	Biological, clinical and population relevance of 95 loci for blood lipids. *Nature*, **2010**, 466, 707-13	50.4	2742
378	METAL: fast and efficient meta-analysis of genomewide association scans. *Bioinformatics*, **2010**, 26, 2190-1	7.2	2697
377	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
376	A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants. *Science*, **2007**, 316, 1341-5	33.3	2269
375	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nature Genetics*, **2010**, 42, 937-48	36.3	2267
374	Genome-wide association studies for complex traits: consensus, uncertainty and challenges. *Nature Reviews Genetics*, **2008**, 9, 356-69	30.1	2126
373	LocusZoom: regional visualization of genome-wide association scan results. *Bioinformatics*, **2010**, 26, 2336-7	7.2	1751
372	MaCH: using sequence and genotype data to estimate haplotypes and unobserved genotypes. *Genetic Epidemiology*, **2010**, 34, 816-34	2.6	1535
371	Plasma HDL cholesterol and risk of myocardial infarction: a mendelian randomisation study. *Lancet, The*, **2012**, 380, 572-80	40	1523
370	Hundreds of variants clustered in genomic loci and biological pathways affect human height. *Nature*, **2010**, 467, 832-8	50.4	1514

369	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
368	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. *Nature Genetics*, **2010**, 42, 579-89	36.3	1449
367	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
366	Next-generation genotype imputation service and methods. *Nature Genetics*, **2016**, 48, 1284-1287	36.3	1369
365	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
364	Genome-wide detection and characterization of positive selection in human populations. *Nature*, **2007**, 449, 913-8	50.4	1367
363	Replicating genotype-phenotype associations. *Nature*, **2007**, 447, 655-60	50.4	1363
362	Defining the role of common variation in the genomic and biological architecture of adult human height. *Nature Genetics*, **2014**, 46, 1173-86	36.3	1339
361	Newly identified loci that influence lipid concentrations and risk of coronary artery disease. *Nature Genetics*, **2008**, 40, 161-9	36.3	1304
360	Fast and accurate genotype imputation in genome-wide association studies through pre-phasing. *Nature Genetics*, **2012**, 44, 955-9	36.3	1292
359	Evolution and functional impact of rare coding variation from deep sequencing of human exomes. *Science*, **2012**, 337, 64-9	33.3	1280
358	Genome-wide association scan shows genetic variants in the FTO gene are associated with obesity-related traits. *PLoS Genetics*, **2007**, 3, e115	6	1231
357	Genetic variants regulating ORMDL3 expression contribute to the risk of childhood asthma. *Nature*, **2007**, 448, 470-3	50.4	1201
356	Common variants at 30 loci contribute to polygenic dyslipidemia. *Nature Genetics*, **2009**, 41, 56-65	36.3	1095
355	A note on exact tests of Hardy-Weinberg equilibrium. *American Journal of Human Genetics*, **2005**, 76, 887-93	11	1064
354	Common variants near MC4R are associated with fat mass, weight and risk of obesity. *Nature Genetics*, **2008**, 40, 768-75	36.3	1048
353	Joint analysis is more efficient than replication-based analysis for two-stage genome-wide association studies. *Nature Genetics*, **2006**, 38, 209-13	36.3	1040
352	Genome-wide scan reveals association of psoriasis with IL-23 and NF-kappaB pathways. *Nature Genetics*, **2009**, 41, 199-204	36.3	1038

351	Genome-wide association study identifies eight loci associated with blood pressure. *Nature Genetics*, **2009**, 41, 666-76	36.3	970
350	New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196	50.4	920
349	Genotype imputation. *Annual Review of Genomics and Human Genetics*, **2009**, 10, 387-406	9.7	812
348	A genome-wide association study of global gene expression. *Nature Genetics*, **2007**, 39, 1202-7	36.3	801
347	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
346	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. *Nature Genetics*, **2016**, 48, 134-43	36.3	769
345	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. *Nature Genetics*, **2010**, 42, 949-60	36.3	724
344	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. *Nature*, **2013**, 493, 216-20	50.4	723
343	Loss-of-function mutations in APOC3, triglycerides, and coronary disease. *New England Journal of Medicine*, **2014**, 371, 22-31	59.2	721
342	The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47	50.4	704
341	Reference-based phasing using the Haplotype Reference Consortium panel. *Nature Genetics*, **2016**, 48, 1443-1448	36.3	699
340	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. *Nature Genetics*, **2012**, 44, 1341-8	36.3	681
339	Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. *Nature Genetics*, **2018**, 50, 1505-1513	36.3	675
338	Mapping complex disease traits with global gene expression. *Nature Reviews Genetics*, **2009**, 10, 184-94	30.1	658
337	Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nature Genetics*, **2013**, 45, 1345-52	36.3	597
336	Rare-variant association analysis: study designs and statistical tests. *American Journal of Human Genetics*, **2014**, 95, 5-23	11	596
335	Variants in MTNR1B influence fasting glucose levels. *Nature Genetics*, **2009**, 41, 77-81	36.3	584
334	Seven new loci associated with age-related macular degeneration. *Nature Genetics*, **2013**, 45, 433-9, 439e1-2	36.3	577

333	An abundance of rare functional variants in 202 drug target genes sequenced in 14,002 people. *Science*, **2012**, 337, 100-4	33.3	523
332	Meta-analysis and imputation refines the association of 15q25 with smoking quantity. *Nature Genetics*, **2010**, 42, 436-40	36.3	521
331	Association studies of up to 1.2 million individuals yield new insights into the genetic etiology of tobacco and alcohol use. *Nature Genetics*, **2019**, 51, 237-244	36.3	516
330	Identification of ten loci associated with height highlights new biological pathways in human growth. *Nature Genetics*, **2008**, 40, 584-91	36.3	482
329	Genome-wide association study shows BCL11A associated with persistent fetal hemoglobin and amelioration of the phenotype of beta-thalassemia. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 1620-5	11.5	469
328	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. *Nature*, **2015**, 518, 102-6	50.4	463
327	Sequence and haplotype analysis supports HLA-C as the psoriasis susceptibility 1 gene. *American Journal of Human Genetics*, **2006**, 78, 827-851	11	441
326	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. *Nature Genetics*, **2013**, 45, 501-12	36.3	437
325	Genetic variants near TIMP3 and high-density lipoprotein-associated loci influence susceptibility to age-related macular degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 7401-6	11.5	417
324	Genome-wide association study identifies loci influencing concentrations of liver enzymes in plasma. *Nature Genetics*, **2011**, 43, 1131-8	36.3	415
323	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes*, **2017**, 66, 2888-2902	8.9	414
322	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
321	Genome-wide association study in individuals of South Asian ancestry identifies six new type 2 diabetes susceptibility loci. *Nature Genetics*, **2011**, 43, 984-9	36.3	406
320	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. *Nature Genetics*, **2009**, 41, 211-5	36.3	405
319	Heritability of cardiovascular and personality traits in 6,148 Sardinians. *PLoS Genetics*, **2006**, 2, e132	6	401
318	The metabochip, a custom genotyping array for genetic studies of metabolic, cardiovascular, and anthropometric traits. *PLoS Genetics*, **2012**, 8, e1002793	6	395
317	Genome-wide association scan meta-analysis identifies three Loci influencing adiposity and fat distribution. *PLoS Genetics*, **2009**, 5, e1000508	6	393
316	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. *Nature Genetics*, **2018**, 50, 1412-1425	36.3	386

315	Efficiently controlling for case-control imbalance and sample relatedness in large-scale genetic association studies. *Nature Genetics*, **2018**, 50, 1335-1341	36.3	375
314	A variant of mitochondrial protein LOC387715/ARMS2, not HTRA1, is strongly associated with age-related macular degeneration. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 16227-32	11.5	356
313	PEDSTATS: descriptive statistics, graphics and quality assessment for gene mapping data. *Bioinformatics*, **2005**, 21, 3445-7	7.2	356
312	Family-based association tests for genomewide association scans. *American Journal of Human Genetics*, **2007**, 81, 913-26	11	353
311	A first-generation linkage disequilibrium map of human chromosome 22. *Nature*, **2002**, 418, 544-8	50.4	342
310	Genome-wide association study of PR interval. *Nature Genetics*, **2010**, 42, 153-9	36.3	340
309	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nature Genetics*, **2011**, 43, 1005-11	36.3	338
308	Gene polymorphism in Netherton and common atopic disease. *Nature Genetics*, **2001**, 29, 175-8	36.3	327
307	Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. *Science*, **2016**, 351, 1166-71	33.3	325
306	Genetic variants influencing circulating lipid levels and risk of coronary artery disease. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2010**, 30, 2264-76	9.4	318
305	Common variants in the GDF5-UQCC region are associated with variation in human height. *Nature Genetics*, **2008**, 40, 198-203	36.3	315
304	Common variants at 10 genomic loci influence hemoglobin A(1C) levels via glycemic and nonglycemic pathways. *Diabetes*, **2010**, 59, 3229-39	0.9	314
303	Exome-wide association study of plasma lipids in >300,000 individuals. *Nature Genetics*, **2017**, 49, 1758-1766	36.3	310
302	Common variants at ten loci modulate the QT interval duration in the QTSCD Study. *Nature Genetics*, **2009**, 41, 407-14	36.3	308
301	Inactivating mutations in NPC1L1 and protection from coronary heart disease. *New England Journal of Medicine*, **2014**, 371, 2072-82	59.2	307
300	FTO genotype is associated with phenotypic variability of body mass index. *Nature*, **2012**, 490, 267-72	50.4	304
299	Genetic linkage of childhood atopic dermatitis to psoriasis susceptibility loci. *Nature Genetics*, **2001**, 27, 372-3	36.3	303
298	Genome-wide association study of plasma polyunsaturated fatty acids in the InCHIANTI Study. *PLoS Genetics*, **2009**, 5, e1000338	6	300

297	Strong association of the Y402H variant in complement factor H at 1q32 with susceptibility to age-related macular degeneration. *American Journal of Human Genetics*, **2005**, 77, 149-53	11	298
296	Detecting and estimating contamination of human DNA samples in sequencing and array-based genotype data. *American Journal of Human Genetics*, **2012**, 91, 839-48	11	295
295	Using haplotype blocks to map human complex trait loci. *Trends in Genetics*, **2003**, 19, 135-40	8.5	293
294	CFH haplotypes without the Y402H coding variant show strong association with susceptibility to age-related macular degeneration. *Nature Genetics*, **2006**, 38, 1049-54	36.3	291
293	Extent and distribution of linkage disequilibrium in three genomic regions. *American Journal of Human Genetics*, **2001**, 68, 191-197	11	285
292	Genome-wide association study identifies a psoriasis susceptibility locus at TRAF3IP2. *Nature Genetics*, **2010**, 42, 991-5	36.3	283
291	Positional cloning of a novel gene influencing asthma from chromosome 2q14. *Nature Genetics*, **2003**, 35, 258-63	36.3	283
290	Sex-stratified genome-wide association studies including 270,000 individuals show sexual dimorphism in genetic loci for anthropometric traits. *PLoS Genetics*, **2013**, 9, e1003500	6	277
289	Pedigree tests of transmission disequilibrium. *European Journal of Human Genetics*, **2000**, 8, 545-51	5.3	274
288	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. *Nature*, **2021**, 590, 290-299	50.4	268
287	A comparison of phasing algorithms for trios and unrelated individuals. *American Journal of Human Genetics*, **2006**, 78, 437-50	11	267
286	Positional cloning of a quantitative trait locus on chromosome 13q14 that influences immunoglobulin E levels and asthma. *Nature Genetics*, **2003**, 34, 181-6	36.3	263
285	Genetics of blood lipids among ~300,000 multi-ethnic participants of the Million Veteran Program. *Nature Genetics*, **2018**, 50, 1514-1523	36.3	260
284	New models of collaboration in genome-wide association studies: the Genetic Association Information Network. *Nature Genetics*, **2007**, 39, 1045-51	36.3	258
283	Shared genetic origin of asthma, hay fever and eczema elucidates allergic disease biology. *Nature Genetics*, **2017**, 49, 1752-1757	36.3	256
282	Biobank-driven genomic discovery yields new insight into atrial fibrillation biology. *Nature Genetics*, **2018**, 50, 1234-1239	36.3	254
281	Genome-wide association analysis identifies three psoriasis susceptibility loci. *Nature Genetics*, **2010**, 42, 1000-4	36.3	251
280	Powerful regression-based quantitative-trait linkage analysis of general pedigrees. *American Journal of Human Genetics*, **2002**, 71, 238-53	11	250

279	Low-coverage sequencing: implications for design of complex trait association studies. *Genome Research*, **2011**, 21, 940-51	9.7	231
278	Handling marker-marker linkage disequilibrium: pedigree analysis with clustered markers. *American Journal of Human Genetics*, **2005**, 77, 754-67	11	228
277	Unified representation of genetic variants. *Bioinformatics*, **2015**, 31, 2202-4	7.2	226
276	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
275	Combined analysis of genome-wide association studies for Crohn disease and psoriasis identifies seven shared susceptibility loci. *American Journal of Human Genetics*, **2012**, 90, 636-47	11	224
274	Impact of common genetic determinants of Hemoglobin A1c on type 2 diabetes risk and diagnosis in ancestrally diverse populations: A transethnic genome-wide meta-analysis. *PLoS Medicine*, **2017**, 14, e1002383	11.6	223
273	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
272	Meta-analysis of Genome-wide Association Studies for Neuroticism, and the Polygenic Association With Major Depressive Disorder. *JAMA Psychiatry*, **2015**, 72, 642-50	14.5	222
271	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. *PLoS Genetics*, **2015**, 11, e1005378	6	220
270	Unraveling a multifactorial late-onset disease: from genetic susceptibility to disease mechanisms for age-related macular degeneration. *Annual Review of Genomics and Human Genetics*, **2009**, 10, 19-43	9.7	217
269	The GLUT9 gene is associated with serum uric acid levels in Sardinia and Chianti cohorts. *PLoS Genetics*, **2007**, 3, e194	6	217
268	Systematic evaluation of coding variation identifies a candidate causal variant in TM6SF2 influencing total cholesterol and myocardial infarction risk. *Nature Genetics*, **2014**, 46, 345-51	36.3	213
267	Exome array analysis identifies new loci and low-frequency variants influencing insulin processing and secretion. *Nature Genetics*, **2013**, 45, 197-201	36.3	212
266	Molecular dissection of psoriasis: integrating genetics and biology. *Journal of Investigative Dermatology*, **2010**, 130, 1213-26	4.3	206
265	Genetic loci influencing kidney function and chronic kidney disease. *Nature Genetics*, **2010**, 42, 373-5	36.3	205
264	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. *Nature Genetics*, **2016**, 48, 593-9	36.3	204
263	RVTESTS: an efficient and comprehensive tool for rare variant association analysis using sequence data. *Bioinformatics*, **2016**, 32, 1423-6	7.2	199
262	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. *Nature Genetics*, **2014**, 46, 826-36	36.3	199

261	Overexpression of the Cytokine BAFF and Autoimmunity Risk. *New England Journal of Medicine*, **2017**, 376, 1615-1626	59.2	198
260	E2-2 protein and Fuchs corneal dystrophy. *New England Journal of Medicine*, **2010**, 363, 1016-24	59.2	197
259	Meta-analysis of genome scans of age-related macular degeneration. *Human Molecular Genetics*, **2005**, 14, 2257-64	5.6	197
258	Genome-wide association study of vitamin B6, vitamin B12, folate, and homocysteine blood concentrations. *American Journal of Human Genetics*, **2009**, 84, 477-82	11	193
257	Genotype-imputation accuracy across worldwide human populations. *American Journal of Human Genetics*, **2009**, 84, 235-50	11	191
256	Population genetic differentiation of height and body mass index across Europe. *Nature Genetics*, **2015**, 47, 1357-62	36.3	186
255	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. *Nature Genetics*, **2018**, 50, 26-41	36.3	186
254	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. *American Journal of Human Genetics*, **2015**, 97, 816-36	11	185
253	A genome-wide association study on African-ancestry populations for asthma. *Journal of Allergy and Clinical Immunology*, **2010**, 125, 336-346.e4	11.5	179
252	Genome-wide association study identifies variants in TMPRSS6 associated with hemoglobin levels. *Nature Genetics*, **2009**, 41, 1170-2	36.3	179
251	Discovery of common and rare genetic risk variants for colorectal cancer. *Nature Genetics*, **2019**, 51, 76-87	36.3	177
250	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
249	Meta-analysis of genome-wide association studies from the CHARGE consortium identifies common variants associated with carotid intima media thickness and plaque. *Nature Genetics*, **2011**, 43, 940-7	36.3	168
248	Genetic variation in the 22q11 locus and susceptibility to schizophrenia. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 16859-64	11.5	168
247	Toll-like receptor 4 variant D299G is associated with susceptibility to age-related macular degeneration. *Human Molecular Genetics*, **2005**, 14, 1449-55	5.6	158
246	An efficient and scalable analysis framework for variant extraction and refinement from population-scale DNA sequence data. *Genome Research*, **2015**, 25, 918-25	9.7	155
245	Assessment of the psoriatic transcriptome in a large sample: additional regulated genes and comparisons with in vitro models. *Journal of Investigative Dermatology*, **2010**, 130, 1829-40	4.3	155
244	Fine mapping major histocompatibility complex associations in psoriasis and its clinical subtypes. *American Journal of Human Genetics*, **2014**, 95, 162-72	11	151

243	Genetic susceptibility to age-related macular degeneration: a paradigm for dissecting complex disease traits. *Human Molecular Genetics*, **2007**, 16 Spec No. 2, R174-82	5.6	146
242	Gene expression in skin and lymphoblastoid cells: Refined statistical method reveals extensive overlap in cis-eQTL signals. *American Journal of Human Genetics*, **2010**, 87, 779-89	11	144
241	Meta-analysis of gene-level tests for rare variant association. *Nature Genetics*, **2014**, 46, 200-4	36.3	142
240	Age-related macular degeneration: a high-resolution genome scan for susceptibility loci in a population enriched for late-stage disease. *American Journal of Human Genetics*, **2004**, 74, 482-94	11	141
239	Blood pressure loci identified with a gene-centric array. *American Journal of Human Genetics*, **2011**, 89, 688-700	11	137
238	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. *Nature Communications*, **2017**, 8, 15382	17.4	136
237	Variants within the immunoregulatory CBLB gene are associated with multiple sclerosis. *Nature Genetics*, **2010**, 42, 495-7	36.3	136
236	Comparison of methods that use whole genome data to estimate the heritability and genetic architecture of complex traits. *Nature Genetics*, **2018**, 50, 737-745	36.3	131
235	Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls. *Nature*, **2019**, 570, 71-76	50.4	129
234	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. *Nature Genetics*, **2015**, 47, 1272-1281	36.3	129
233	Inherited causes of clonal haematopoiesis in 97,691 whole genomes. *Nature*, **2020**, 586, 763-768	50.4	127
232	Phenotypic Characterization of Genetically Lowered Human Lipoprotein(a) Levels. *Journal of the American College of Cardiology*, **2016**, 68, 2761-2772	15.1	127
231	Global gene expression analysis reveals evidence for decreased lipid biosynthesis and increased innate immunity in uninvolved psoriatic skin. *Journal of Investigative Dermatology*, **2009**, 129, 2795-804	4.3	123
230	Meta-analysis of Genome-Wide Association Studies for Extraversion: Findings from the Genetics of Personality Consortium. *Behavior Genetics*, **2016**, 46, 170-82	3.2	122
229	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. *Nature Communications*, **2015**, 6, 7001	17.4	122
228	Identification of novel genetic Loci associated with thyroid peroxidase antibodies and clinical thyroid disease. *PLoS Genetics*, **2014**, 10, e1004123	6	122
227	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. *Nature*, **2020**, 586, 749-756	50.4	122
226	Fine mapping of five loci associated with low-density lipoprotein cholesterol detects variants that double the explained heritability. *PLoS Genetics*, **2011**, 7, e1002198	6	118

225	A cross-platform analysis of 14,177 expression quantitative trait loci derived from lymphoblastoid cell lines. *Genome Research*, **2013**, 23, 716-26	9.7	117
224	A genome-wide association analysis of serum iron concentrations. *Blood*, **2010**, 115, 94-6	2.2	117
223	Elucidating the genetic architecture of familial schizophrenia using rare copy number variant and linkage scans. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 16746-51	11.5	115
222	Amelioration of Sardinian beta0 thalassemia by genetic modifiers. *Blood*, **2009**, 114, 3935-7	2.2	115
221	In silico method for inferring genotypes in pedigrees. *Nature Genetics*, **2006**, 38, 1002-4	36.3	115
220	Genome-wide association scan of trait depression. *Biological Psychiatry*, **2010**, 68, 811-7	7.9	114
219	Common variants in the SLCO1B3 locus are associated with bilirubin levels and unconjugated hyperbilirubinemia. *Human Molecular Genetics*, **2009**, 18, 2711-8	5.6	113
218	A genome-wide association scan on the levels of markers of inflammation in Sardinians reveals associations that underpin its complex regulation. *PLoS Genetics*, **2012**, 8, e1002480	6	112
217	The impact of genotyping error on family-based analysis of quantitative traits. *European Journal of Human Genetics*, **2001**, 9, 130-4	5.3	112
216	Multiple Loci within the major histocompatibility complex confer risk of psoriasis. *PLoS Genetics*, **2009**, 5, e1000606	6	110
215	Association of Rare and Common Variation in the Lipoprotein Lipase Gene With Coronary Artery Disease. *JAMA - Journal of the American Medical Association*, **2017**, 317, 937-946	27.4	109
214	Optimal designs for two-stage genome-wide association studies. *Genetic Epidemiology*, **2007**, 31, 776-88	2.6	109
213	Genome-wide meta-analysis of 241,258 adults accounting for smoking behaviour identifies novel loci for obesity traits. *Nature Communications*, **2017**, 8, 14977	17.4	105
212	Phosphodiesterase 8B gene variants are associated with serum TSH levels and thyroid function. *American Journal of Human Genetics*, **2008**, 82, 1270-80	11	105
211	Exome sequencing and complex disease: practical aspects of rare variant association studies. *Human Molecular Genetics*, **2012**, 21, R1-9	5.6	104
210	Joint modeling of linkage and association: identifying SNPs responsible for a linkage signal. *American Journal of Human Genetics*, **2005**, 76, 934-49	11	104
209	Genome-wide physical activity interactions in adiposity - A meta-analysis of 200,452 adults. *PLoS Genetics*, **2017**, 13, e1006528	6	103
208	Evidence of association of APOE with age-related macular degeneration: a pooled analysis of 15 studies. *Human Mutation*, **2011**, 32, 1407-16	4.7	99

207	GENOME: a rapid coalescent-based whole genome simulator. *Bioinformatics*, **2007**, 23, 1565-7	7.2	97
206	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. *Nature Neuroscience*, **2017**, 20, 1661-1668	25.5	95
205	Retinal transcriptome and eQTL analyses identify genes associated with age-related macular degeneration. *Nature Genetics*, **2019**, 51, 606-610	36.3	93
204	Efficient study designs for test of genetic association using sibship data and unrelated cases and controls. *American Journal of Human Genetics*, **2006**, 78, 778-792	11	92
203	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals. *Nature Communications*, **2018**, 9, 3391	17.4	90
202	Genome-wide linkage analysis of a composite index of neuroticism and mood-related scales in extreme selected sibships. *Human Molecular Genetics*, **2004**, 13, 2173-82	5.6	90
201	Genome-wide meta-analysis of psoriatic arthritis identifies susceptibility locus at REL. *Journal of Investigative Dermatology*, **2012**, 132, 1133-40	4.3	89
200	Functional gene group analysis reveals a role of synaptic heterotrimeric G proteins in cognitive ability. *American Journal of Human Genetics*, **2010**, 86, 113-25	11	89
199	The power to detect linkage disequilibrium with quantitative traits in selected samples. *American Journal of Human Genetics*, **2001**, 68, 1463-74	11	87
198	Functional equivalence of genome sequencing analysis pipelines enables harmonized variant calling across human genetics projects. *Nature Communications*, **2018**, 9, 4038	17.4	87
197	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. *Hypertension*, **2017**,	8.5	85
196	Mutations in a BTB-Kelch protein, KLHL7, cause autosomal-dominant retinitis pigmentosa. *American Journal of Human Genetics*, **2009**, 84, 792-800	11	84
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