Goncalo Abecasis

List of Publications by Year in Descending Order

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	Clonal hematopoiesis in sickle cell disease.. *Journal of Clinical Investigation*, **2022**,	15.9	7
385	Genetic determinants of telomere length from 109,122 ancestrally diverse whole-genome sequences in TOPMed.. *Cell Genomics*, **2022**, 2, 100084-100084		1
384	Transethnic analysis of psoriasis susceptibility in South Asians and Europeans enhances fine-mapping in the MHC and genomewide.. *Human Genetics and Genomics Advances*, **2022**, 3, 100069-100069	3.8	0
383	Analysis of rare genetic variation underlying cardiometabolic diseases and traits among 200,000 individuals in the UK Biobank.. *Nature Genetics*, **2022**,	36.3	4
382	Genome-wide analysis provides genetic evidence that ACE2 influences COVID-19 risk and yields risk scores associated with severe disease.. *Nature Genetics*, **2022**,	36.3	9
381	Assessing the contribution of rare variants to complex trait heritability from whole-genome sequence data.. *Nature Genetics*, **2022**,	36.3	6
380	Whole genome sequencing reveals host factors underlying critical Covid-19.. *Nature*, **2022**,	50.4	8
379	Nasal airway transcriptome-wide association study of asthma reveals genetically driven mucus pathobiology.. *Nature Communications*, **2022**, 13, 1632	17.4	2
378	Mendelian randomization supports bidirectional causality between telomere length and clonal hematopoiesis of indeterminate potential.. *Science Advances*, **2022**, 8, eabl6579	14.3	3
377	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation.. *Nature Genetics*, **2022**,	36.3	7
376	Impact of natural selection on global patterns of genetic variation and association with clinical phenotypes at genes involved in SARS-CoV-2 infection.. *Proceedings of the National Academy of Sciences of the United States of America*, **2022**, 119, e2123000119	11.5	0
375	The power of genetic diversity in genome-wide association studies of lipids. *Nature*, **2021**,	50.4	24
374	Genome-wide analysis of 53,400 people with irritable bowel syndrome highlights shared genetic pathways with mood and anxiety disorders. *Nature Genetics*, **2021**, 53, 1543-1552	36.3	11
373	Association of mitochondrial DNA copy number with cardiometabolic diseases.. *Cell Genomics*, **2021**, 1,		1
372	A high-resolution HLA reference panel capturing global population diversity enables multi-ancestry fine-mapping in HIV host response. *Nature Genetics*, **2021**, 53, 1504-1516	36.3	7
371	Exome sequencing and analysis of 454,787 UK Biobank participants. *Nature*, **2021**, 599, 628-634	50.4	34
370	LocusZoom.js: Interactive and embeddable visualization of genetic association study results. *Bioinformatics*, **2021**,	7.2	8

369	Chromosome Xq23 is associated with lower atherogenic lipid concentrations and favorable cardiometabolic indices. *Nature Communications*, **2021**, 12, 2182	17.4	5
368	Computationally efficient whole-genome regression for quantitative and binary traits. *Nature Genetics*, **2021**, 53, 1097-1103	36.3	51
367	Clonal hematopoiesis associated with epigenetic aging and clinical outcomes. *Aging Cell*, **2021**, 20, e13366	9.9	9
366	The trans-ancestral genomic architecture of glycemic traits. *Nature Genetics*, **2021**, 53, 840-860	36.3	44
365	Genome sequencing unveils a regulatory landscape of platelet reactivity. *Nature Communications*, **2021**, 12, 3626	17.4	6
364	Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals. *American Journal of Human Genetics*, **2021**, 108, 1350-1355	11	25
363	Mapping the 17q12-21.1 Locus for Variants Associated with Early-Onset Asthma in African Americans. *American Journal of Respiratory and Critical Care Medicine*, **2021**, 203, 424-436	10.2	5
362	Identifying Novel Susceptibility Genes for Colorectal Cancer Risk From a Transcriptome-Wide Association Study of 125,478 Subjects. *Gastroenterology*, **2021**, 160, 1164-1178.e6	13.3	15
361	mutation alters immune system activation, inflammation, and risk of autoimmunity. *Multiple Sclerosis Journal*, **2021**, 27, 1332-1340	5	4
360	Sex-dimorphic genetic effects and novel loci for fasting glucose and insulin variability. *Nature Communications*, **2021**, 12, 24	17.4	30
359	Genetic architectures of proximal and distal colorectal cancer are partly distinct. *Gut*, **2021**, 70, 1325-1334	19.2	7
358	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. *Nature*, **2021**, 590, 290-299	50.4	268
357	Whole-genome association analyses of sleep-disordered breathing phenotypes in the NHLBI TOPMed program. *Genome Medicine*, **2021**, 13, 136	14.4	3
356	Whole-genome sequencing in diverse subjects identifies genetic correlates of leukocyte traits: The NHLBI TOPMed program. *American Journal of Human Genetics*, **2021**, 108, 1836-1851	11	1
355	Presence and transmission of mitochondrial heteroplasmic mutations in human populations of European and African ancestry. *Mitochondrion*, **2021**, 60, 33-42	4.9	0
354	Whole genome sequence analysis of platelet traits in the NHLBI trans-omics for precision medicine initiative. *Human Molecular Genetics*, **2021**,	5.6	2
353	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. *Molecular Psychiatry*, **2021**, 26, 5239-5250	15.1	3
352	GWAS of stool frequency provides insights into gastrointestinal motility and irritable bowel syndrome.. *Cell Genomics*, **2021**, 1, None		2

351	A Sardinian founder mutation in glycoprotein Ib platelet subunit beta (GP1BB) that impacts thrombocytopenia. *British Journal of Haematology*, **2020**, 191, e124-e128	4.5	0
350	Lung Function in African American Children with Asthma Is Associated with Novel Regulatory Variants of the KIT Ligand and Gene-By-Air-Pollution Interaction. *Genetics*, **2020**, 215, 869-886	4	3
349	Scalable generalized linear mixed model for region-based association tests in large biobanks and cohorts. *Nature Genetics*, **2020**, 52, 634-639	36.3	41
348	Association Analysis and Meta-Analysis of Multi-Allelic Variants for Large-Scale Sequence Data. *Genes*, **2020**, 11,	4.2	1
347	Whole-Genome Sequencing Identifies Novel Functional Loci Associated with Lung Function in Puerto Rican Youth. *American Journal of Respiratory and Critical Care Medicine*, **2020**, 202, 962-972	10.2	1
346	Exploring and visualizing large-scale genetic associations by using PheWeb. *Nature Genetics*, **2020**, 52, 550-552	36.3	41
345	Age-of-onset information helps identify 76 genetic variants associated with allergic disease. *PLoS Genetics*, **2020**, 16, e1008725	6	10
344	De novo mutations across 1,465 diverse genomes reveal mutational insights and reductions in the Amish founder population. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 2560-2569	11.5	29
343	Ancestry-agnostic estimation of DNA sample contamination from sequence reads. *Genome Research*, **2020**, 30, 185-194	9.7	15
342	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. *Human Molecular Genetics*, **2020**, 29, 2022-2034	5.6	9
341	LabWAS: Novel findings and study design recommendations from a meta-analysis of clinical labs in two independent biobanks. *PLoS Genetics*, **2020**, 16, e1009077	6	6
340	Asthma and its relationship to mitochondrial copy number: Results from the Asthma Translational Genomics Collaborative (ATGC) of the Trans-Omics for Precision Medicine (TOPMed) program. *PLoS ONE*, **2020**, 15, e0242364	3.7	2
339	Integrating comprehensive functional annotations to boost power and accuracy in gene-based association analysis. *PLoS Genetics*, **2020**, 16, e1009060	6	3
338	Type 2 and interferon inflammation strongly regulate SARS-CoV-2 related gene expression in the airway epithelium **2020**,		30
337	Identification of CFTR variants in Latino patients with cystic fibrosis from the Dominican Republic and Puerto Rico. *Pediatric Pulmonology*, **2020**, 55, 533-540	3.5	3
336	Whole genome sequence analysis of pulmonary function and COPD in 19,996 multi-ethnic participants. *Nature Communications*, **2020**, 11, 5182	17.4	6
335	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. *Nature*, **2020**, 586, 749-756	50.4	122
334	Type 2 and interferon inflammation regulate SARS-CoV-2 entry factor expression in the airway epithelium. *Nature Communications*, **2020**, 11, 5139	17.4	68

333	Inherited causes of clonal haematopoiesis in 97,691 whole genomes. *Nature*, **2020**, 586, 763-768	50.4	127
332	MEPE loss-of-function variant associates with decreased bone mineral density and increased fracture risk. *Nature Communications*, **2020**, 11, 4093	17.4	4
331	A Novel Recurrent Genetic Variant Is Associated With a Dysplasia-Associated Arterial Disease Exhibiting Dissections and Fibromuscular Dysplasia. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2020**, 40, 2686-2699	9.4	11
330	Complex genetic signatures in immune cells underlie autoimmunity and inform therapy. *Nature Genetics*, **2020**, 52, 1036-1045	36.3	16
329	Dynamic incorporation of multiple in silico functional annotations empowers rare variant association analysis of large whole-genome sequencing studies at scale. *Nature Genetics*, **2020**, 52, 969-983	36.3	33
328	Loss-of-function genomic variants highlight potential therapeutic targets for cardiovascular disease. *Nature Communications*, **2020**, 11, 6417	17.4	17
327	Meta-analysis of up to 622,409 individuals identifies 40 novel smoking behaviour associated genetic loci. *Molecular Psychiatry*, **2020**, 25, 2392-2409	15.1	45
326	Genes for Good: Engaging the Public in Genetics Research via Social Media. *American Journal of Human Genetics*, **2019**, 105, 65-77	11	8
325	Exploring various polygenic risk scores for skin cancer in the phenomes of the Michigan genomics initiative and the UK Biobank with a visual catalog: PRSWeb. *PLoS Genetics*, **2019**, 15, e1008202	6	18
324	Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls. *Nature*, **2019**, 570, 71-76	50.4	129
323	Sex-specific and pleiotropic effects underlying kidney function identified from GWAS meta-analysis. *Nature Communications*, **2019**, 10, 1847	17.4	22
322	Relative impact of indels versus SNPs on complex disease. *Genetic Epidemiology*, **2019**, 43, 112-117	2.6	3
321	Retinal transcriptome and eQTL analyses identify genes associated with age-related macular degeneration. *Nature Genetics*, **2019**, 51, 606-610	36.3	93
320	emeraLD: rapid linkage disequilibrium estimation with massive datasets. *Bioinformatics*, **2019**, 35, 164-166	4.62	4
319	Meta-MultiSKAT: Multiple phenotype meta-analysis for region-based association test. *Genetic Epidemiology*, **2019**, 43, 800-814	2.6	7
318	Estimation of DNA contamination and its sources in genotyped samples. *Genetic Epidemiology*, **2019**, 43, 980-995	2.6	3
317	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. *Nature Genetics*, **2019**, 51, 452-469	36.3	44
316	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

315	Exome Chip Meta-analysis Fine Maps Causal Variants and Elucidates the Genetic Architecture of Rare Coding Variants in Smoking and Alcohol Use. *Biological Psychiatry*, **2019**, 85, 946-955	7.9	35
314	Discovery of common and rare genetic risk variants for colorectal cancer. *Nature Genetics*, **2019**, 51, 76-87	36.3	177
313	Narrow-sense heritability estimation of complex traits using identity-by-descent information. *Heredity*, **2018**, 121, 616-630	3.6	14
312	Genome-wide analysis of disease progression in age-related macular degeneration. *Human Molecular Genetics*, **2018**, 27, 929-940	5.6	37
311	Genome-wide Study of Atrial Fibrillation Identifies Seven Risk Loci and Highlights Biological Pathways and Regulatory Elements Involved in Cardiac Development. *American Journal of Human Genetics*, **2018**, 102, 103-115	11	53
310	Improved score statistics for meta-analysis in single-variant and gene-level association studies. *Genetic Epidemiology*, **2018**, 42, 333-343	2.6	3
309	A Mixed-Effects Model for Powerful Association Tests in Integrative Functional Genomics. *American Journal of Human Genetics*, **2018**, 102, 904-919	11	20
308	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
307	Biobank-driven genomic discovery yields new insight into atrial fibrillation biology. *Nature Genetics*, **2018**, 50, 1234-1239	36.3	254
306	Proper conditional analysis in the presence of missing data: Application to large scale meta-analysis of tobacco use phenotypes. *PLoS Genetics*, **2018**, 14, e1007452	6	11
305	Efficiently controlling for case-control imbalance and sample relatedness in large-scale genetic association studies. *Nature Genetics*, **2018**, 50, 1335-1341	36.3	375
304	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals. *Nature Communications*, **2018**, 9, 3391	17.4	90
303	Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes. *Nature Communications*, **2018**, 9, 2252	17.4	71
302	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
301	Genome-wide association study of delay discounting in 23,217 adult research participants of European ancestry. *Nature Neuroscience*, **2018**, 21, 16-18	25.5	56
300	Association of Rare Predicted Loss-of-Function Variants in Cellular Pathways with Sub-Phenotypes in Age-Related Macular Degeneration. *Ophthalmology*, **2018**, 125, 398-406	7.3	7
299	Imputation-Aware Tag SNP Selection To Improve Power for Large-Scale, Multi-ethnic Association Studies. *G3: Genes, Genomes, Genetics*, **2018**, 8, 3255-3267	3.2	17
298	Association Between Titin Loss-of-Function Variants and Early-Onset Atrial Fibrillation. *JAMA - Journal of the American Medical Association*, **2018**, 320, 2354-2364	27.4	75

297	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
296	Genetics of blood lipids among ~300,000 multi-ethnic participants of the Million Veteran Program. *Nature Genetics*, **2018**, 50, 1514-1523	36.3	260
295	Functional equivalence of genome sequencing analysis pipelines enables harmonized variant calling across human genetics projects. *Nature Communications*, **2018**, 9, 4038	17.4	87
294	Genetic signature to provide robust risk assessment of psoriatic arthritis development in psoriasis patients. *Nature Communications*, **2018**, 9, 4178	17.4	61
293	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
292	Genomic history of the Sardinian population. *Nature Genetics*, **2018**, 50, 1426-1434	36.3	42
291	Genotype Imputation from Large Reference Panels. *Annual Review of Genomics and Human Genetics* , **2018**, 19, 73-96	9.7	68
290	Association of Polygenic Risk Scores for Multiple Cancers in a Phenome-wide Study: Results from The Michigan Genomics Initiative. *American Journal of Human Genetics*, **2018**, 102, 1048-1061	11	83
289	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
288	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
287	Population- and individual-specific regulatory variation in Sardinia. *Nature Genetics*, **2017**, 49, 700-707	36.3	24
286	Mitogenome Diversity in Sardinians: A Genetic Window onto an Islandâ Past. *Molecular Biology and Evolution*, **2017**, 34, 1230-1239	8.3	43
285	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
284	Overexpression of the Cytokine BAFF and Autoimmunity Risk. *New England Journal of Medicine*, **2017**, 376, 1615-1626	59.2	198
283	A Fast and Accurate Algorithm to Test for Binary Phenotypes and Its Application to PheWAS. *American Journal of Human Genetics*, **2017**, 101, 37-49	11	66
282	Bivariate Analysis of Age-Related Macular Degeneration Progression Using Genetic Risk Scores. *Genetics*, **2017**, 206, 119-133	4	31
281	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. *Nature Communications*, **2017**, 8, 15382	17.4	136
280	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes*, **2017**, 66, 2888-2902	8.2	414

279	A Low-Frequency Inactivating Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. *Diabetes*, **2017**, 66, 2019-2032	0.9	29
278	Exome-wide association study of plasma lipids in >300,000 individuals. *Nature Genetics*, **2017**, 49, 1758-1766	36.3	310
277	Exome chip meta-analysis identifies novel loci and East Asian-specific coding variants that contribute to lipid levels and coronary artery disease. *Nature Genetics*, **2017**, 49, 1722-1730	36.3	83
276	Shared genetic origin of asthma, hay fever and eczema elucidates allergic disease biology. *Nature Genetics*, **2017**, 49, 1752-1757	36.3	256
275	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
274	LASER server: ancestry tracing with genotypes or sequence reads. *Bioinformatics*, **2017**, 33, 2056-2058	7.2	15
273	A Scalable Bayesian Method for Integrating Functional Information in Genome-wide Association Studies. *American Journal of Human Genetics*, **2017**, 101, 404-416	11	41
272	Improving power of association tests using multiple sets of imputed genotypes from distributed reference panels. *Genetic Epidemiology*, **2017**, 41, 744-755	2.6	13
271	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
270	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. *Nature Neuroscience*, **2017**, 20, 1661-1668	25.5	95
269	Characterization of ADME gene variation in 21 populations by exome sequencing. *Pharmacogenetics and Genomics*, **2017**, 27, 89-100	1.9	24
268	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Scientific Data*, **2017**, 4, 170179	8.2	22
267	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. *Human Molecular Genetics*, **2017**, 26, 4301-4313	5.6	25
266	Genome-wide physical activity interactions in adiposity - A meta-analysis of 200,452 adults. *PLoS Genetics*, **2017**, 13, e1006528	6	103
265	Optimal sequencing strategies for identifying disease-associated singletons. *PLoS Genetics*, **2017**, 13, e1006811	6	15
264	Next-generation genotype imputation service and methods. *Nature Genetics*, **2016**, 48, 1284-1287	36.3	1369
263	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
262	Prosaposin is a regulator of progranulin levels and oligomerization. *Nature Communications*, **2016**, 7, 11992	17.4	49

261	A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. *Nature Communications*, **2016**, 7, 13357	17.4	46
260	Genetic variants in CETP increase risk of intracerebral hemorrhage. *Annals of Neurology*, **2016**, 80, 730-740	9.4	24
259	52 Genetic Loci Influencing Myocardial Mass. *Journal of the American College of Cardiology*, **2016**, 68, 1435-1448	15.1	76
258	Omics-squared: human genomic, transcriptomic and phenotypic data for genetic analysis workshop 19. *BMC Proceedings*, **2016**, 10, 71-77	2.3	14
257	Independent test assessment using the extreme value distribution theory. *BMC Proceedings*, **2016**, 10, 245-249	2.3	1
256	Platelet-Related Variants Identified by Exomechip Meta-analysis in 157,293 Individuals. *American Journal of Human Genetics*, **2016**, 99, 40-55	11	61
255	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
254	Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. *Science*, **2016**, 351, 1166-71	33.3	325
253	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
252	RVTESTS: an efficient and comprehensive tool for rare variant association analysis using sequence data. *Bioinformatics*, **2016**, 32, 1423-6	7.2	199
251	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
250	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
249	Fine-Mapping of Common Genetic Variants Associated with Colorectal Tumor Risk Identified Potential Functional Variants. *PLoS ONE*, **2016**, 11, e0157521	3.7	5
248	The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47	50.4	704
247	Phenotypic Characterization of Genetically Lowered Human Lipoprotein(a) Levels. *Journal of the American College of Cardiology*, **2016**, 68, 2761-2772	15.1	127
246	Targeted exonic sequencing of GWAS loci in the high extremes of the plasma lipids distribution. *Atherosclerosis*, **2016**, 250, 63-8	3.1	9
245	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. *Nature Genetics*, **2016**, 48, 593-9	36.3	204
244	Reference-based phasing using the Haplotype Reference Consortium panel. *Nature Genetics*, **2016**, 48, 1443-1448	36.3	699

243	Graphical algorithm for integration of genetic and biological data: proof of principle using psoriasis as a model. *Bioinformatics*, **2015**, 31, 1243-9	7.2	7
242	Unified representation of genetic variants. *Bioinformatics*, **2015**, 31, 2202-4	7.2	226
241	Author reply: To PMID 24974817. *Ophthalmology*, **2015**, 122, e46-7	7.3	1
240	Correcting for Sample Contamination in Genotype Calling of DNA Sequence Data. *American Journal of Human Genetics*, **2015**, 97, 284-90	11	29
239	A single SNP surrogate for genotyping HLA-C*06:02 in diverse populations. *Journal of Investigative Dermatology*, **2015**, 135, 1177-1180	4.3	4
238	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
237	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
236	Genome-wide association study of susceptibility loci for breast cancer in Sardinian population. *BMC Cancer*, **2015**, 15, 383	4.8	9
235	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
234	A global reference for human genetic variation. *Nature*, **2015**, 526, 68-74	50.4	8599
233	Genome-wide association analyses based on whole-genome sequencing in Sardinia provide insights into regulation of hemoglobin levels. *Nature Genetics*, **2015**, 47, 1264-71	36.3	49
232	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
231	Population genetic differentiation of height and body mass index across Europe. *Nature Genetics*, **2015**, 47, 1357-62	36.3	186
230	Height-reducing variants and selection for short stature in Sardinia. *Nature Genetics*, **2015**, 47, 1352-1356	36.3	71
229	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
228	Reply: To PMID 25456150. *Ophthalmology*, **2015**, 122, e61-2	7.3	
227	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. *Nature*, **2015**, 518, 102-6	50.4	463
226	Rare variant genotype imputation with thousands of study-specific whole-genome sequences: implications for cost-effective study designs. *European Journal of Human Genetics*, **2015**, 23, 975-83	5.3	57

225	Fine mapping of eight psoriasis susceptibility loci. *European Journal of Human Genetics*, **2015**, 23, 844-53	5.3	21
224	Reply: To PMID 25456150. *Ophthalmology*, **2015**, 122, e63	7.3	
223	Reply: To PMID 25456150. *Ophthalmology*, **2015**, 122, e58-9	7.3	
222	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
221	Assessing Mitochondrial DNA Variation and Copy Number in Lymphocytes of ~2,000 Sardinians Using Tailored Sequencing Analysis Tools. *PLoS Genetics*, **2015**, 11, e1005306	6	72
220	Whole-genome sequence-based analysis of thyroid function. *Nature Communications*, **2015**, 6, 5681	17.4	56
219	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
218	Improved ancestry estimation for both genotyping and sequencing data using projection procrustes analysis and genotype imputation. *American Journal of Human Genetics*, **2015**, 96, 926-37	11	82
217	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. *Nature Communications*, **2015**, 6, 7001	17.4	122
216	2014 Curt Stern Award: Adventures in human genetics. *American Journal of Human Genetics*, **2015**, 96, 363-6	11	
215	Genetic testing in persons with age-related macular degeneration and the use of the AREDS supplements: to test or not to test?. *Ophthalmology*, **2015**, 122, 212-5	7.3	50
214	New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196	50.4	920
213	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
212	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
211	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
210	Inactivating mutations in NPC1L1 and protection from coronary heart disease. *New England Journal of Medicine*, **2014**, 371, 2072-82	59.2	307
209	No clinically significant association between CFH and ARMS2 genotypes and response to nutritional supplements: AREDS report number 38. *Ophthalmology*, **2014**, 121, 2173-80	7.3	76
208	Rare-variant association analysis: study designs and statistical tests. *American Journal of Human Genetics*, **2014**, 95, 5-23	11	596

207	Fine mapping major histocompatibility complex associations in psoriasis and its clinical subtypes. *American Journal of Human Genetics*, **2014**, 95, 162-72	11	151
206	Data for Genetic Analysis Workshop 18: human whole genome sequence, blood pressure, and simulated phenotypes in extended pedigrees. *BMC Proceedings*, **2014**, 8, S2	2.3	64
205	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
204	Prevalence of CKD and its relationship to eGFR-related genetic loci and clinical risk factors in the SardiNIA study cohort. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 1533-44	12.7	27
203	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
202	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. *Nature Genetics*, **2014**, 46, 826-36	36.3	199
201	Meta-analysis of gene-level tests for rare variant association. *Nature Genetics*, **2014**, 46, 200-4	36.3	142
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8	Fine-mapping of an expanded set of type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps	18
7	Protein-Coding Variants Implicate Novel Genes Related to Lipid Homeostasis Contributing to Body Fat Distribution	1
6	Genetic discovery and translational decision support from exome sequencing of 20,791 type 2 diabetes cases and 24,440 controls from five ancestries	2
5	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program	68
4	Whole exome sequencing and characterization of coding variation in 49,960 individuals in the UK Biobank	56
3	Tissue-Specific Alteration of Metabolic Pathways Influences Glycemic Regulation	4
2	Clonal hematopoiesis in sickle cell disease	1
1	Whole genome sequencing identifies common and rare structural variants contributing to hematologic traits in the NHLBI TOPMed program	1