Kari Stefansson

List of Publications by Year in Descending Order

686 papers	128,531 citations	183 h-index	347 g-index
755 ext. papers	154,052 ext. citations	18.5 avg, IF	6.91 L-index

#	Paper	IF	Citations
686	Response by Bjï¿½nsson et al to Letter Regarding Article, "Large-Scale Screening for Monogenic and Clinically Defined Familial Hypercholesterolemia in Iceland".. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2022**, 42, e46-e47	9.4	
685	Functional dissection of inherited non-coding variation influencing multiple myeloma risk.. *Nature Communications*, **2022**, 13, 151	17.4	0
684	Rare SLC13A1 variants associate with intervertebral disc disorder highlighting role of sulfate in disc pathology.. *Nature Communications*, **2022**, 13, 634	17.4	3
683	Genome-wide analysis of 102,084 migraine cases identifies 123 risk loci and subtype-specific risk alleles.. *Nature Genetics*, **2022**, 54, 152-160	36.3	13
682	Population-level deficit of homozygosity unveils CPSF3 as an intellectual disability syndrome gene.. *Nature Communications*, **2022**, 13, 705	17.4	0
681	Established risk loci for systemic lupus erythematosus at NCF2, STAT4, TNPO3, IRF5 and ITGAM associate with distinct clinical manifestations: a Danish genome-wide association study.. *Joint Bone Spine*, **2022**, 105357	2.9	
680	Genetic overlap analysis of endometriosis and asthma identifies shared loci implicating sex hormones and thyroid signalling pathways.. *Human Reproduction*, **2022**, 37, 366-383	5.7	2
679	Reconstruction of a large-scale outbreak of SARS-CoV-2 infection in Iceland informs vaccination strategies.. *Clinical Microbiology and Infection*, **2022**,	9.5	2
678	A genome-wide meta-analysis identifies 50 genetic loci associated with carpal tunnel syndrome.. *Nature Communications*, **2022**, 13, 1598	17.4	1
677	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome.. *American Journal of Human Genetics*, **2022**, 109, 601-617	11	0
676	Mapping genomic loci implicates genes and synaptic biology in schizophrenia.. *Nature*, **2022**,	50.4	35
675	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation.. *Nature Genetics*, **2022**,	36.3	7
674	Data Resource Profile: The Copenhagen Hospital Biobank (CHB). *International Journal of Epidemiology*, **2021**, 50, 719-720e	7.8	3
673	Genome-wide association study of panic disorder reveals genetic overlap with neuroticism and depression. *Molecular Psychiatry*, **2021**, 26, 4179-4190	15.1	8
672	Large-scale integration of the plasma proteome with genetics and disease. *Nature Genetics*, **2021**, 53, 1712-1721	36.3	17
671	Comment on "Evaluating the cardiovascular safety of sclerostin inhibition using evidence from meta-analysis of clinical trials and human genetics". *Science Translational Medicine*, **2021**, 13, eabe8497	17.5	1
670	A genome-wide meta-analysis uncovers six sequence variants conferring risk of vertigo. *Communications Biology*, **2021**, 4, 1148	6.7	3

669	1q21.1 distal copy number variants are associated with cerebral and cognitive alterations in humans. *Translational Psychiatry*, **2021**, 11, 182	8.6	6
668	Allele frequency of variants reported to cause adenine phosphoribosyltransferase deficiency. *European Journal of Human Genetics*, **2021**, 29, 1061-1070	5.3	1
667	Identification of genetic loci associated with nocturnal enuresis: a genome-wide association study. *The Lancet Child and Adolescent Health*, **2021**, 5, 201-209	14.5	7
666	Variable number tandem repeats mediate the expression of proximal genes. *Nature Communications*, **2021**, 12, 2075	17.4	10
665	Germline variants at SOHLH2 influence multiple myeloma risk. *Blood Cancer Journal*, **2021**, 11, 76	7	1
664	Long-read sequencing of 3,622 Icelanders provides insight into the role of structural variants in human diseases and other traits. *Nature Genetics*, **2021**, 53, 779-786	36.3	44
663	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. *Nature Genetics*, **2021**, 53, 817-829	36.3	83
662	The genetic structure of Norway. *European Journal of Human Genetics*, **2021**, 29, 1710-1718	5.3	1
661	Eleven genomic loci affect plasma levels of chronic inflammation marker soluble urokinase-type plasminogen activator receptor. *Communications Biology*, **2021**, 4, 655	6.7	3
660	The genetic architecture of age-related hearing impairment revealed by genome-wide association analysis. *Communications Biology*, **2021**, 4, 706	6.7	4
659	Polygenic Risk Score-Enhanced Risk Stratification of Coronary Artery Disease in Patients With Stable Chest Pain. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003298	5.2	2
658	Predicting the probability of death using proteomics. *Communications Biology*, **2021**, 4, 758	6.7	2
657	Molecular benchmarks of a SARS-CoV-2 epidemic. *Nature Communications*, **2021**, 12, 3633	17.4	0
656	Distinction between the effects of parental and fetal genomes on fetal growth. *Nature Genetics*, **2021**, 53, 1135-1142	36.3	5
655	Identification of genetic overlap and novel risk loci for attention-deficit/hyperactivity disorder and bipolar disorder. *Molecular Psychiatry*, **2021**, 26, 4055-4065	15.1	15
654	Genome-wide association study identifies 48 common genetic variants associated with handedness. *Nature Human Behaviour*, **2021**, 5, 59-70	12.8	33
653	Increased absorption of phytosterols is the simplest and most plausible explanation for coronary artery disease risk not accounted for by non-HDL cholesterol in high cholesterol absorbers. *European Heart Journal*, **2021**, 42, 283-284	9.5	3
652	Lifelong Reduction in LDL (Low-Density Lipoprotein) Cholesterol due to a Gain-of-Function Mutation in. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003029	5.2	5

651	A polygenic resilience score moderates the genetic risk for schizophrenia. *Molecular Psychiatry*, **2021**, 26, 800-815	15.1	15
650	Sex-dimorphic genetic effects and novel loci for fasting glucose and insulin variability. *Nature Communications*, **2021**, 12, 24	17.4	30
649	A meta-analysis uncovers the first sequence variant conferring risk of Bell's palsy. *Scientific Reports*, **2021**, 11, 4188	4.9	1
648	Loss-of-Function Variants in the Tumor-Suppressor Gene Confer Increased Cancer Risk. *Cancer Research*, **2021**, 81, 1954-1964	10.1	2
647	PopDel identifies medium-size deletions simultaneously in tens of thousands of genomes. *Nature Communications*, **2021**, 12, 730	17.4	4
646	A genome-wide meta-analysis yields 46 new loci associating with biomarkers of iron homeostasis. *Communications Biology*, **2021**, 4, 156	6.7	11
645	Effects of copy number variations on brain structure and risk for psychiatric illness: Large-scale studies from the ENIGMA working groups on CNVs. *Human Brain Mapping*, **2021**,	5.9	6
644	Genetic insight into sick sinus syndrome. *European Heart Journal*, **2021**, 42, 1959-1971	9.5	7
643	Genetic analyses of gynecological disease identify genetic relationships between uterine fibroids and endometrial cancer, and a novel endometrial cancer genetic risk region at the WNT4 1p36.12 locus. *Human Genetics*, **2021**, 140, 1353-1365	6.3	5
642	Cognition in Schizophrenia. *Biological Psychiatry*, **2021**, 90, 4-5	7.9	
641	Identification of 371 genetic variants for age at first sex and birth linked to externalising behaviour. *Nature Human Behaviour*, **2021**,	12.8	5
640	Genetic insights into biological mechanisms governing human ovarian ageing. *Nature*, **2021**, 596, 393-397	50.4	28
639	Sequence variants in malignant hyperthermia genes in Iceland: classification and actionable findings in a population database. *European Journal of Human Genetics*, **2021**, 29, 1819-1824	5.3	0
638	Identifying the Common Genetic Basis of Antidepressant Response. *Biological Psychiatry Global Open Science*, **2021**,		4
637	Genetic variants associated with platelet count are predictive of human disease and physiological markers. *Communications Biology*, **2021**, 4, 1132	6.7	0
636	The CRTAC1 Protein in Plasma Is Associated With Osteoarthritis and Predicts Progression to Joint Replacement: A Large-Scale Proteomics Scan in Iceland. *Arthritis and Rheumatology*, **2021**, 73, 2025-2034	9.5	6
635	Analysis of Diffusion Tensor Imaging Data From the UK Biobank Confirms Dosage Effect of 15q11.2 Copy Number Variation on White Matter and Shows Association With Cognition. *Biological Psychiatry*, **2021**, 90, 307-316	7.9	1
634	A genome-wide association study with 1,126,563 individuals identifies new risk loci for Alzheimer's disease. *Nature Genetics*, **2021**, 53, 1276-1282	36.3	40

633	Genetic propensities for verbal and spatial ability have opposite effects on body mass index and risk of schizophrenia. *Intelligence*, **2021**, 88, 101565	3	0
632	The Genetic Architecture of Depression in Individuals of East Asian Ancestry: A Genome-Wide Association Study. *JAMA Psychiatry*, **2021**, 78, 1258-1269	14.5	7
631	Deciphering osteoarthritis genetics across 826,690 individuals from 9 populations. *Cell*, **2021**, 184, 4784-4818.e17	38.6	24
630	Large-Scale Screening for Monogenic and Clinically Defined Familial Hypercholesterolemia in Iceland. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2021**, 41, 2616-2628	9.4	4
629	Differences between germline genomes of monozygotic twins. *Nature Genetics*, **2021**, 53, 27-34	36.3	24
628	Cohort profile: Copenhagen Hospital Biobank - Cardiovascular Disease Cohort (CHB-CVDC): Construction of a large-scale genetic cohort to facilitate a better understanding of heart diseases. *BMJ Open*, **2021**, 11, e049709	3	0
627	Genetic predisposition to hypertension is associated with preeclampsia in European and Central Asian women. *Nature Communications*, **2020**, 11, 5976	17.4	30
626	Habitual sleep disturbances and migraine: a Mendelian randomization study. *Annals of Clinical and Translational Neurology*, **2020**, 7, 2370-2380	5.3	6
625	Multi-ancestry GWAS of the electrocardiographic PR interval identifies 202 loci underlying cardiac conduction. *Nature Communications*, **2020**, 11, 2542	17.4	16
624	FLT3 stop mutation increases FLT3 ligand level and risk of autoimmune thyroid disease. *Nature*, **2020**, 584, 619-623	50.4	23
623	Genome-wide association identifies seven loci for pelvic organ prolapse in Iceland and the UK Biobank. *Communications Biology*, **2020**, 3, 129	6.7	7
622	Assessing thyroid cancer risk using polygenic risk scores. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 5997-6002	11.5	17
621	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. *Nature Communications*, **2020**, 11, 3368	17.4	22
620	Genetic Risk of Coronary Artery Disease, Features of Atherosclerosis, and Coronary Plaque Burden. *Journal of the American Heart Association*, **2020**, 9, e014795	6	9
619	Eighty-eight variants highlight the role of T cell regulation and airway remodeling in asthma pathogenesis. *Nature Communications*, **2020**, 11, 393	17.4	29
618	Predicted loss and gain of function mutations in ACO1 are associated with erythropoiesis. *Communications Biology*, **2020**, 3, 189	6.7	8
617	Genome-wide meta-analysis identifies eight new susceptibility loci for cutaneous squamous cell carcinoma. *Nature Communications*, **2020**, 11, 820	17.4	13
616	Association of Copy Number Variation of the 15q11.2 BP1-BP2 Region With Cortical and Subcortical Morphology and Cognition. *JAMA Psychiatry*, **2020**, 77, 420-430	14.5	24

615	Genome-wide association and Mendelian randomisation analysis provide insights into the pathogenesis of heart failure. *Nature Communications*, **2020**, 11, 163	17.4	140
614	Association of Genetically Predicted Lipid Levels With the Extent of Coronary Atherosclerosis in Icelandic Adults. *JAMA Cardiology*, **2020**, 5, 13-20	16.2	15
613	Identification of Genetic Loci Shared Between Attention-Deficit/Hyperactivity Disorder, Intelligence, and Educational Attainment. *Biological Psychiatry*, **2020**, 87, 1052-1062	7.9	5
612	A large-scale genome-wide association study meta-analysis of cannabis use disorder. *Lancet Psychiatry,the*, **2020**, 7, 1032-1045	23.3	43
611	Sequence Variants in TAAR5 and Other Loci Affect Human Odor Perception and Naming. *Current Biology*, **2020**, 30, 4643-4653.e3	6.3	10
610	Discovery of rare variants associated with blood pressure regulation through meta-analysis of 1.3 million individuals. *Nature Genetics*, **2020**, 52, 1314-1332	36.3	26
609	Large genome-wide association study identifies three novel risk variants for restless legs syndrome. *Communications Biology*, **2020**, 3, 703	6.7	11
608	GWAS of thyroid stimulating hormone highlights pleiotropic effects and inverse association with thyroid cancer. *Nature Communications*, **2020**, 11, 3981	17.4	21
607	Genetic variability in the absorption of dietary sterols affects the risk of coronary artery disease. *European Heart Journal*, **2020**, 41, 2618-2628	9.5	26
606	MEPE loss-of-function variant associates with decreased bone mineral density and increased fracture risk. *Nature Communications*, **2020**, 11, 4093	17.4	4
605	Humoral Immune Response to SARS-CoV-2 in Iceland. *New England Journal of Medicine*, **2020**, 383, 1724-1734	59.1	593
604	Common and Rare Sequence Variants Influencing Tumor Biomarkers in Blood. *Cancer Epidemiology Biomarkers and Prevention*, **2020**, 29, 225-235	4	3
603	Dose response of the 16p11.2 distal copy number variant on intracranial volume and basal ganglia. *Molecular Psychiatry*, **2020**, 25, 584-602	15.1	24
602	Classical Human Leukocyte Antigen Alleles and C4 Haplotypes Are Not Significantly Associated With Depression. *Biological Psychiatry*, **2020**, 87, 419-430	7.9	9
601	The nature of Neanderthal introgression revealed by 27,566 Icelandic genomes. *Nature*, **2020**, 582, 78-83	50.4	33
600	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. *International Journal of Epidemiology*, **2020**, 49, 1022-1031	7.8	15
599	Spread of SARS-CoV-2 in the Icelandic Population. *New England Journal of Medicine*, **2020**, 382, 2302-2315	59.2	842
598	Transcriptome-wide association study of multiple myeloma identifies candidate susceptibility genes. *Human Genomics*, **2019**, 13, 37	6.8	5

597	Genome-wide association meta-analyses and fine-mapping elucidate pathways influencing albuminuria. *Nature Communications*, **2019**, 10, 4130	17.4	43
596	Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics*, **2019**, 51, 1459-1474	36.3	122
595	Shared heritability and functional enrichment across six solid cancers. *Nature Communications*, **2019** , 10, 431	17.4	45
594	Characterizing mutagenic effects of recombination through a sequence-level genetic map. *Science*, **2019**, 363,	33.3	97
593	Comparing migraine with and without aura to healthy controls using RNA sequencing. *Cephalalgia*, **2019**, 39, 1435-1444	6.1	9
592	Genome-wide association study implicates CHRNA2 in cannabis use disorder. *Nature Neuroscience*, **2019**, 22, 1066-1074	25.5	60
591	A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics*, **2019**, 51, 957-972	36.3	217
590	GBA and APOE ε4 associate with sporadic dementia with Lewy bodies in European genome wide association study. *Scientific Reports*, **2019**, 9, 7013	4.9	24
589	GWAS of bone size yields twelve loci that also affect height, BMD, osteoarthritis or fractures. *Nature Communications*, **2019**, 10, 2054	17.4	36
588	Genome-wide association study identifies 30 loci associated with bipolar disorder. *Nature Genetics*, **2019**, 51, 793-803	36.3	662
587	Tracking Five Millennia of Horse Management with Extensive Ancient Genome Time Series. *Cell*, **2019**, 177, 1419-1435.e31	56.2	110
586	Sequence variation at ANAPC1 accounts for 24% of the variability in corneal endothelial cell density. *Nature Communications*, **2019**, 10, 1284	17.4	13
585	Clinical characteristics of patients with colorectal cancer with double somatic mismatch repair mutations compared with Lynch syndrome. *Journal of Medical Genetics*, **2019**, 56, 462-470	5.8	31
584	Meta-Analysis of Genomewide Association Studies Reveals Genetic Variants for Hip Bone Geometry. *Journal of Bone and Mineral Research*, **2019**, 34, 1284-1296	6.3	16
583	Interrogating the Genetic Determinants of Tourette's Syndrome and Other Tic Disorders Through Genome-Wide Association Studies. *American Journal of Psychiatry*, **2019**, 176, 217-227	11.9	95
582	Roadmap for a precision-medicine initiative in the Nordic region. *Nature Genetics*, **2019**, 51, 924-930	36.3	12
581	A Missense Variant in PTPN22 is a Risk Factor for Drug-induced Liver Injury. *Gastroenterology*, **2019**, 156, 1707-1716.e2	13.3	59
580	Identification of common genetic risk variants for autism spectrum disorder. *Nature Genetics*, **2019**, 51, 431-444	36.3	746

579	Sequence variants with large effects on cardiac electrophysiology and disease. *Nature Communications*, **2019**, 10, 4803	17.4	12
578	Attention-deficit hyperactivity disorder shares copy number variant risk with schizophrenia and autism spectrum disorder. *Translational Psychiatry*, **2019**, 9, 258	8.6	39
577	Disentangling the genetics of lean mass. *American Journal of Clinical Nutrition*, **2019**, 109, 276-287	7	24
576	The mother's risk of premature death after child loss across two centuries. *ELife*, **2019**, 8,	8.9	5
575	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. *Nature Genetics*, **2019**, 51, 452-469	36.3	44
574	Genetic predisposition to mosaic Y chromosome loss in blood. *Nature*, **2019**, 575, 652-657	50.4	83
573	Brain age prediction using deep learning uncovers associated sequence variants. *Nature Communications*, **2019**, 10, 5409	17.4	86
572	GraphTyper2 enables population-scale genotyping of structural variation using pangenome graphs. *Nature Communications*, **2019**, 10, 5402	17.4	43
571	Lipoprotein(a) Concentration and Risks of Cardiovascular Disease and Diabetes. *Journal of the American College of Cardiology*, **2019**, 74, 2982-2994	15.1	61
570	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. *Cell*, **2019**, 179, 1469-1482.e11	56.2	402
569	Association of Whole-Genome and NETRIN1 Signaling Pathway-Derived Polygenic Risk Scores for Major Depressive Disorder and White Matter Microstructure in the UK Biobank. *Biological Psychiatry: Cognitive Neuroscience and Neuroimaging*, **2019**, 4, 91-100	3.4	12
568	Reciprocal White Matter Changes Associated With Copy Number Variation at 15q11.2 BP1-BP2: A Diffusion Tensor Imaging Study. *Biological Psychiatry*, **2019**, 85, 563-572	7.9	20
567	Discovery of the first genome-wide significant risk loci for attention deficit/hyperactivity disorder. *Nature Genetics*, **2019**, 51, 63-75	36.3	826
566	Genome-wide meta-analysis identifies new loci and functional pathways influencing Alzheimer's disease risk. *Nature Genetics*, **2019**, 51, 404-413	36.3	771
565	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
564	A loss-of-function variant in ALOX15 protects against nasal polyps and chronic rhinosinusitis. *Nature Genetics*, **2019**, 51, 267-276	36.3	44
563	Sequence variants associating with urinary biomarkers. *Human Molecular Genetics*, **2019**, 28, 1199-1211	5.6	13
562	Genome-wide analysis yields new loci associating with aortic valve stenosis. *Nature Communications* , **2018**, 9, 987	17.4	56

561	Common schizophrenia alleles are enriched in mutation-intolerant genes and in regions under strong background selection. *Nature Genetics*, **2018**, 50, 381-389	36.3	787
560	2017 William Allan Award. *American Journal of Human Genetics*, **2018**, 102, 351-353	11	0
559	Rare SCARB1 mutations associate with high-density lipoprotein cholesterol but not with coronary artery disease. *European Heart Journal*, **2018**, 39, 2172-2178	9.5	42
558	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. *Nature Genetics*, **2018**, 50, 559-571	36.3	221
557	Regulatory variants at KLF14 influence type 2 diabetes risk via a female-specific effect on adipocyte size and body composition. *Nature Genetics*, **2018**, 50, 572-580	36.3	82
556	The nature of nurture: Effects of parental genotypes. *Science*, **2018**, 359, 424-428	33.3	409
555	Rare and Common Variants Conferring Risk of Tooth Agenesis. *Journal of Dental Research*, **2018**, 97, 515-522	5.2	19
554	Profile of common prostate cancer risk variants in an unscreened Romanian population. *Journal of Cellular and Molecular Medicine*, **2018**, 22, 1574-1582	5.6	4
553	Multiancestry association study identifies new asthma risk loci that colocalize with immune-cell enhancer marks. *Nature Genetics*, **2018**, 50, 42-53	36.3	246
552	Reconstructing an African haploid genome from the 18th century. *Nature Genetics*, **2018**, 50, 199-205	36.3	12
551	Life-Course Genome-wide Association Study Meta-analysis of Total Body BMD and Assessment of Age-Specific Effects. *American Journal of Human Genetics*, **2018**, 102, 88-102	11	119
550	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. *Neuron*, **2018**, 98, 743-753.e4	13.9	42
549	Genome-wide association analyses identify 44 risk variants and refine the genetic architecture of major depression. *Nature Genetics*, **2018**, 50, 668-681	36.3	1301
548	Genome-wide analyses using UK Biobank data provide insights into the genetic architecture of osteoarthritis. *Nature Genetics*, **2018**, 50, 549-558	36.3	122
547	Polygenic risk scores for schizophrenia and bipolar disorder associate with addiction. *Addiction Biology*, **2018**, 23, 485-492	4.6	58
546	Genome-wide association study across European and African American ancestries identifies a SNP in DNMT3B contributing to nicotine dependence. *Molecular Psychiatry*, **2018**, 23, 1911-1919	15.1	52
545	Does Childhood Trauma Moderate Polygenic Risk for Depression? A Meta-analysis of 5765 Subjects From the Psychiatric Genomics Consortium. *Biological Psychiatry*, **2018**, 84, 138-147	7.9	48
544	Biobank-driven genomic discovery yields new insight into atrial fibrillation biology. *Nature Genetics*, **2018**, 50, 1234-1239	36.3	254

543	PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. *Nature Communications*, **2018**, 9, 2904	17.4	39
542	Coding variants in and increase risk of atrial fibrillation. *Communications Biology*, **2018**, 1, 68	6.7	21
541	Genome-wide association and HLA fine-mapping studies identify risk loci and genetic pathways underlying allergic rhinitis. *Nature Genetics*, **2018**, 50, 1072-1080	36.3	52
540	A rare missense mutation in MYH6 associates with non-syndromic coarctation of the aorta. *European Heart Journal*, **2018**, 39, 3243-3249	9.5	29
539	Relatedness disequilibrium regression estimates heritability without environmental bias. *Nature Genetics*, **2018**, 50, 1304-1310	36.3	84
538	A truncating mutation in EPOR leads to hypo-responsiveness to erythropoietin with normal haemoglobin. *Communications Biology*, **2018**, 1, 49	6.7	8
537	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
536	Multiancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. *Nature Genetics*, **2018**, 50, 524-537	36.3	536
535	Genome-wide associations for benign prostatic hyperplasia reveal a genetic correlation with serum levels of PSA. *Nature Communications*, **2018**, 9, 4568	17.4	24
534	Multiple transmissions of de novo mutations in families. *Nature Genetics*, **2018**, 50, 1674-1680	36.3	50
533	Meta-analysis of Alzheimer's disease on 9,751 samples from Norway and IGAP study identifies four risk loci. *Scientific Reports*, **2018**, 8, 18088	4.9	25
532	Large-scale genome-wide meta-analysis of polycystic ovary syndrome suggests shared genetic architecture for different diagnosis criteria. *PLoS Genetics*, **2018**, 14, e1007813	6	166
531	Genome-wide association meta-analysis yields 20 loci associated with gallstone disease. *Nature Communications*, **2018**, 9, 5101	17.4	29
530	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
529	A homozygous loss-of-function mutation leading to CYBC1 deficiency causes chronic granulomatous disease. *Nature Communications*, **2018**, 9, 4447	17.4	54
528	Variants in NKX2-5 and FLNC Cause Dilated Cardiomyopathy and Sudden Cardiac Death. *Circulation Genomic and Precision Medicine*, **2018**, 11, e002151	5.2	19
527	Meta-analysis of Icelandic and UK data sets identifies missense variants in SMO, IL11, COL11A1 and 13 more new loci associated with osteoarthritis. *Nature Genetics*, **2018**, 50, 1681-1687	36.3	67
526	Identification of Lynch syndrome risk variants in the Romanian population. *Journal of Cellular and Molecular Medicine*, **2018**, 22, 6068-6076	5.6	4

525	Insights into imprinting from parent-of-origin phased methylomes and transcriptomes. *Nature Genetics*, **2018**, 50, 1542-1552	36.3	56
524	Identification of multiple risk loci and regulatory mechanisms influencing susceptibility to multiple myeloma. *Nature Communications*, **2018**, 9, 3707	17.4	57
523	Variants associating with uterine leiomyoma highlight genetic background shared by various cancers and hormone-related traits. *Nature Communications*, **2018**, 9, 3636	17.4	31
522	Association of BRCA2 K3326* With Small Cell Lung Cancer and Squamous Cell Cancer of the Skin. *Journal of the National Cancer Institute*, **2018**, 110, 967-974	9.7	16
521	Ancient genomes from Iceland reveal the making of a human population. *Science*, **2018**, 360, 1028-1032	33.3	37
520	Estimation of Genetic Correlation via Linkage Disequilibrium Score Regression and Genomic Restricted Maximum Likelihood. *American Journal of Human Genetics*, **2018**, 102, 1185-1194	11	55
519	Genome-wide Association for Major Depression Through Age at Onset Stratification: Major Depressive Disorder Working Group of the Psychiatric Genomics Consortium. *Biological Psychiatry*, **2017**, 81, 325-335	7.9	129
518	Selection against variants in the genome associated with educational attainment. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E727-E732	11.5	84
517	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
516	A genome-wide association study yields five novel thyroid cancer risk loci. *Nature Communications*, **2017**, 8, 14517	17.4	80
515	Sequence variant at 8q24.21 associates with sciatica caused by lumbar disc herniation. *Nature Communications*, **2017**, 8, 14265	17.4	22
514	Diversity in non-repetitive human sequences not found in the reference genome. *Nature Genetics*, **2017**, 49, 588-593	36.3	42
513	Genome-wide association analyses for lung function and chronic obstructive pulmonary disease identify new loci and potential druggable targets. *Nature Genetics*, **2017**, 49, 416-425	36.3	170
512	Description of the EuroTARGET cohort: A European collaborative project on TArgeted therapy in renal cell cancer-GEnetic- and tumor-related biomarkers for response and toxicity. *Urologic Oncology: Seminars and Original Investigations*, **2017**, 35, 529.e9-529.e16	2.8	6
511	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. *Nature Genetics*, **2017**, 49, 834-841	36.3	257
510	Clonal hematopoiesis, with and without candidate driver mutations, is common in the elderly. *Blood* , **2017**, 130, 742-752	2.2	365
509	Loss of Cardioprotective Effects at the Locus as a Result of Gene-Smoking Interactions. *Circulation*, **2017**, 135, 2336-2353	16.7	36
508	15q11.2 CNV affects cognitive, structural and functional correlates of dyslexia and dyscalculia. *Translational Psychiatry*, **2017**, 7, e1109	8.6	52

507	Genome-wide analyses for personality traits identify six genomic loci and show correlations with psychiatric disorders. *Nature Genetics*, **2017**, 49, 152-156	36.3	251
506	Meta-Analysis of Genome-Wide Association Studies for Abdominal Aortic Aneurysm Identifies Four New Disease-Specific Risk Loci. *Circulation Research*, **2017**, 120, 341-353	15.7	97
505	Truncating mutations in RBM12 are associated with psychosis. *Nature Genetics*, **2017**, 49, 1251-1254	36.3	45
504	Variants in the fetal genome near FLT1 are associated with risk of preeclampsia. *Nature Genetics*, **2017**, 49, 1255-1260	36.3	118
503	Identification of sequence variants influencing immunoglobulin levels. *Nature Genetics*, **2017**, 49, 1182-1191	36.3	57
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