Citation Report
List of articles citing

Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program

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#	Paper	IF	Citations
698	Inherited causes of clonal haematopoiesis in 97,691 whole genomes. *Nature*, **2020**, 586, 763-768	50.4	127
697	Seqminer2: an efficient tool to query and retrieve genotypes for statistical genetics analyses from biobank scale sequence dataset. **2020**, 36, 4951-4954		
696	A Sardinian founder mutation in glycoprotein Ib platelet subunit beta (GP1BB) that impacts thrombocytopenia. **2020**, 191, e124-e128		0
695	Comparative genetic analysis of the novel coronavirus (2019-nCoV/SARS-CoV-2) receptor ACE2 in different populations. **2020**, 6, 11		561
694	Increased burden of ultra-rare structural variants localizing to boundaries of topologically associated domains in schizophrenia. **2020**, 11, 1842		22
693	Premature Menopause, Clonal Hematopoiesis, and Coronary Artery Disease in Postmenopausal Women. **2021**, 143, 410-423		31
692	Transcriptional profile of platelets and iPSC-derived megakaryocytes from whole-genome and RNA sequencing. **2021**, 137, 959-968		5
691	An interaction of the 17q12-21 locus with mold exposure in childhood asthma. **2021**, 32, 373-376		
690	A novel locus for exertional dyspnoea in childhood asthma. **2021**, 57,		3
689	Non-coding variants in MYH11, FZD3, and SORCS3 are associated with dementia in women. **2021**, 17, 215-225		3
688	Estimating heritability and its enrichment in tissue-specific gene sets in admixed populations. **2021**, 30, 1521-1534		6
687	Predicting genotype-specific gene regulatory networks.		0
686	Exome-wide age-of-onset analysis reveals exonic variants in ERN1 and SPPL2C associated with Alzheimer's disease. **2021**, 11, 146		1
685	High coverage whole genome sequencing of the expanded 1000 Genomes Project cohort including 602 trios.		31
684	Low-pass sequencing increases the power of GWAS and decreases measurement error of polygenic risk scores compared to genotyping arrays. **2021**, 31, 529-537		14
683	A unified genealogy of modern and ancient genomes.		4
682	Genome-wide association study identifies two new loci associated with anti-NMDAR encephalitis.		

681	Collaborative Cohort of Cohorts for COVID-19 Research (C4R) Study: Study Design. **2021**,	1
680	The TOPMed genomic resource for human health. **2021**, 22, 200	3
679	Cardiovascular Informatics: building a bridge to data harmony. **2021**,	3
678	Genetic architecture of four smoking behaviors using partitioned SNP heritability. **2021**, 116, 2498-2508	2
677	Incorporating European GWAS findings improve polygenic risk prediction accuracy of breast cancer among East Asians. **2021**, 45, 471-484	2
676	ATAV: a comprehensive platform for population-scale genomic analyses. **2021**, 22, 149	5
675	Long-read assembly of a Great Dane genome highlights the contribution of GC-rich sequence and mobile elements to canine genomes. **2021**, 118,	6
674	Population-scale detection of non-reference sequence variants using colored de Bruijn Graphs.	
673	Robust, flexible, and scalable tests for Hardy-Weinberg equilibrium across diverse ancestries. **2021**, 218,	0
672	Genetic Risk Stratification: A Paradigm Shift in Prevention of Coronary Artery Disease. **2021**, 6, 287-304	6
671	Why are rare variants hard to impute? Coalescent models reveal theoretical limits in existing algorithms. **2021**, 217,	1
670	RNA-SeQC 2: Efficient RNA-seq quality control and quantification for large cohorts. **2021**,	4
669	NHLBI-CMREF Workshop Report on Pulmonary Vascular Disease´Classification: JACC State-of-the-Art Review. **2021**, 77, 2040-2052	6
668	A novel age-informed approach for genetic association analysis in Alzheimer's disease. **2021**, 13, 72	2
667	Advancing drug discovery using the power of the human genome. **2021**, 254, 418-429	1
666	SmProt: A Reliable Repository with Comprehensive Annotation of Small Proteins Identified from Ribosome Profiling.	0
665	The impact of non-additive genetic associations on age-related complex diseases. **2021**, 12, 2436	10
664	Inverting the model of genomics data sharing with the NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-space (AnVIL).	5

663	Chromosome Xq23 is associated with lower atherogenic lipid concentrations and favorable cardiometabolic indices. **2021**, 12, 2182		5
662	Genetic and non-genetic factors affecting the expression of COVID-19-relevant genes in the large airway epithelium. **2021**, 13, 66		6
661	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. **2021**, 108, 656-668		10
660	One scientist couple's five suggestions to solve the 'two body problem'. *Nature*, **2021**,	50.4	0
659	Novel recessive locus for body mass index in childhood asthma. **2021**, 76, 1227-1230		0
658	A System for Phenotype Harmonization in the National Heart, Lung, and Blood Institute Trans-Omics for Precision Medicine (TOPMed) Program. **2021**, 190, 1977-1992		3
657	Interpreting type 1 diabetes risk with genetics and single-cell epigenomics. *Nature*, **2021**, 594, 398-402	50.4	20
656	Leveraging a founder population to identify novel rare-population genetic determinants of lipidome.		0
655	Genome-wide association study of cardiac troponin I in the general population. **2021**, 30, 2027-2039		5
654	Two-phase sample selection strategies for design and analysis in post-genome wide association fine-mapping studies.		
653	Genetic structure correlates with ethnolinguistic diversity in eastern and southern Africa.		0
652	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder.		
651	A spatially aware likelihood test to detect sweeps from haplotype distributions.		0
650	Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. **2021**, 108, 919-928		13
649	The genetic background of the associations between sense of coherence and mental health, self-esteem and personality. **2021**, 1		0
648	Genetic and epigenetic features of promoters with ubiquitous chromatin accessibility support ubiquitous transcription of cell-essential genes. **2021**, 49, 5705-5725		0
647	Sparse Allele Vectors and the Savvy Software Suite. **2021**,		1
646	Large trans-ethnic meta-analysis identifies AKR1C4 as a novel gene associated with age at menarche. **2021**, 36, 1999-2010		1

645	Novel loci and Mapuche genetic ancestry are associated with pubertal growth traits in Chilean boys. **2021**, 140, 1651-1661	0
644	The Dockstore: enhancing a community platform for sharing reproducible and accessible computational protocols. **2021**, 49, W624-W632	2
643	Germline risk of clonal haematopoiesis. **2021**, 22, 603-617	11
642	Genomic risk prediction for breast cancer in older women.	
641	Identification of putative causal loci in whole-genome sequencing data via knockoff statistics. **2021**, 12, 3152	3
640	Stability of Polygenic Scores Across Discovery Genome-Wide Association Studies.	2
639	Genetic evaluation of cardiomyopathies in Qatar identifies enrichment of pathogenic sarcomere gene variants and possible founder disease mutations in the Arabs. **2021**, 9, e1709	1
638	Genome sequencing unveils a regulatory landscape of platelet reactivity. **2021**, 12, 3626	6
637	Undisclosed, unmet and neglected challenges in multi-omics studies.	5
636	Recognition and inhibition of SARS-CoV-2 by humoral innate immunity pattern recognition molecules.	1
635	Insights on Genetic and Environmental Factors in Parkinson Disease from a regional Swedish Case-Control Cohort.	
634	Fine-mapping, trans-ancestral and genomic analyses identify causal variants, cells, genes and drug targets for type 1 diabetes. **2021**, 53, 962-971	28
633	XCVATR: Characterization of Variant Impact on the Embeddings of Single -Cell and Bulk RNA-Sequencing Samples.	0
632	Identifying the Lipidomic Effects of a Rare Loss-of-Function Deletion in. **2021**, 14, e003232	1
631	Clonal hematopoiesis in sickle cell disease.	1
630	The effect of protein mutations on drug binding suggests ensuing personalised drug selection. **2021**, 11, 13452	3
629	Mutational Landscape of the Proglucagon-Derived Peptides. **2021**, 12, 698511	3
628	The missing link between genetic association and regulatory function.	1

627	Mutation saturation for fitness effects at human CpG sites.	3
626	North Carolina Macular Dystrophy: Phenotypic Variability and Computational Analysis of Disease-Associated Noncoding Variants. **2021**, 62, 16	2
625	Association between ABO and Duffy blood types and circulating chemokines and cytokines. **2021**, 22, 161-171	3
624	The ARIC (Atherosclerosis Risk In Communities) Study: JACC Focus Seminar 3/8. **2021**, 77, 2939-2959	23
623	Revisiting the pathogenic mechanism of the GJB1 5' UTR c.-103C > T mutation causing CMTX1. **2021**, 22, 149-160	
622	New susceptibility loci for severe COVID-19 by detailed GWAS analysis in European populations.	2
621	Genetic epidemiology approach to estimating birth incidence and current disease prevalence for rhizomelic chondrodysplasia punctata. **2021**, 16, 300	0
620	A complete reference genome improves analysis of human genetic variation.	9
619	Germ-line mutations in predispose to familial papillary thyroid cancer. **2021**, 118,	1
618	Genomic Risk Prediction for Breast Cancer in Older Women. **2021**, 13,	1
617	Impact of natural selection on global patterns of genetic variation, and association with clinical phenotypes, at genes involved in SARS-CoV-2 infection. **2021**,	0
616	Characterizing the Genetic Architecture of Parkinson's Disease in Latinos. **2021**, 90, 353-365	8
615	Multi-resolution characterization of molecular taxonomies in bulk and single-cell transcriptomics data. **2021**, 49, e98	1
614	The Thousand Polish Genomes Project a national database of Polish variant allele frequencies.	0
613	Sodium Sensitivity, Sodium Resistance, and Incidence of Hypertension: A Longitudinal Follow-Up Study of Dietary Sodium Intervention. **2021**, 78, 155-164	3
612	Monogenic and Polygenic Models of Coronary Artery Disease. **2021**, 23, 107	0
611	Family SES Is Associated with the Gut Microbiome in Infants and Children. **2021**, 9,	5
610	Impact of natural selection on global patterns of genetic variation, and association with clinical phenotypes, at genes involved in SARS-CoV-2 infection. **2021**,	4

609	Somatic mutations in benign breast disease tissues and association with breast cancer risk. **2021**, 14, 185	0
608	A fast Data-Driven method for genotype imputation, phasing, and local ancestry inference: MendelImpute.jl. **2021**,	0
607	BinomiRare: A robust test for association of a rare genetic variant with a binary outcome for mixed models and any case-control proportion. **2021**, 2,	1
606	Genomewide Association Studies in Pharmacogenomics. **2021**, 110, 637-648	5
605	Profiling variable-number tandem repeat variation across populations using repeat-pangenome graphs. **2021**, 12, 4250	4
604	A strategy for building and using a human reference pangenome. **2019**, 8, 1751	2
603	Polygenic Risk Prediction using Gradient Boosted Trees Captures Non-Linear Genetic Effects and Allele Interactions in Complex Phenotypes.	1
602	Whole-exome imputation within UK Biobank powers rare coding variant association and fine-mapping analyses. **2021**, 53, 1260-1269	7
601	GCAT|Panel, a comprehensive structural variant haplotype map of the Iberian population from high-coverage whole-genome sequencing.	1
600	Summix: A method for detecting and adjusting for population structure in genetic summary data. **2021**, 108, 1270-1282	0
599	Identification of novel and rare variants associated with handgrip strength using whole genome sequence data from the NHLBI Trans-Omics in Precision Medicine (TOPMed) Program. **2021**, 16, e0253611	1
598	Effective variant filtering and expected candidate variant yield in studies of rare human disease. **2021**, 6, 60	8
597	Protein prediction for trait mapping in diverse populations.	
596	LoFTK: a framework for fully automated calculation of predicted Loss-of-Function variants.	
595	Progress and Research Priorities in Imaging Genomics for Heart and Lung Disease: Summary of an NHLBI Workshop. **2021**, 14, e012943	1
594	Using de novo assembly to identify structural variation of eight complex immune system gene regions. **2021**, 17, e1009254	5
593	Using attentive gated neural networks to quantify the impact of non-coding variants on transcription factor binding affinity.	
592	Quantitative Human Paleogenetics: What can Ancient DNA Tell us About Complex Trait Evolution?. **2021**, 12, 703541	5

591	Epigenome-wide association study of mitochondrial genome copy number. **2021**,	0
590	Enhanced mixture interpretation with macrohaplotypes based on long-read DNA sequencing. **2021**, 135, 2189-2198	1
589	Gene expression in African Americans and Latinos reveals ancestry-specific patterns of genetic architecture.	0
588	Genome-wide association studies. **2021**, 1,	50
587	Population sequencing data reveal a compendium of mutational processes in the human germ line. **2021**, 373, 1030-1035	7
586	Analyses of biomarker traits in diverse UK biobank participants identify associations missed by European-centric analysis strategies. **2021**,	6
585	Decoding disease: from genomes to networks to phenotypes. **2021**, 22, 774-790	7
584	Exploring human-genome gut-microbiome interaction in Parkinson's disease. **2021**, 7, 74	4
583	Partitioning gene-level contributions to complex-trait heritability by allele frequency identifies disease-relevant genes.	1
582	Whole-genome association analyses of sleep-disordered breathing phenotypes in the NHLBI TOPMed program. **2021**, 13, 136	3
581	Linking genome variants to disease: scalable approaches to test the functional impact of human mutations. **2021**, 30, R187-R197	4
580	Privacy-preserving genotype imputation in a trusted execution environment. **2021**, 12, 983-993.e7	3
579	An Association Test of the Spatial Distribution of Rare Missense Variants within Protein Structures Improves Statistical Power of Sequencing Studies.	
578	3'aQTL-atlas: an atlas of 3'UTR alternative polyadenylation quantitative trait loci across human normal tissues. **2021**,	0
577	Building a Chinese pan-genome of 486 individuals. **2021**, 4, 1016	2
576	Genome-Wide Association Meta-Analysis Supports Genes Involved in Valve and Cardiac Development to Associate With Mitral Valve Prolapse. **2021**, 14, e003148	0
575	DIAPH1 Variants in Non-East Asian Patients With Sporadic Moyamoya Disease. **2021**, 78, 993-1003	7
574	Early-Onset Atrial Fibrillation and Heritable Heart Disease-To Test or Not to Test?. **2021**,	1

573	STAAR Workflow: A cloud-based workflow for scalable and reproducible rare variant analysis.	
572	Genome-wide Association Study Identifies 2 New Loci Associated With Anti-NMDAR Encephalitis. **2021**, 8,	3
571	SVAT: Secure Outsourcing of Variant Annotation and Genotype Aggregation.	
570	A natural variant in ANP32B impairs influenza virus replication in human cells. **2021**, 102,	3
569	Presence and transmission of mitochondrial heteroplasmic mutations in human populations of European and African ancestry. **2021**, 60, 33-42	0
568	Protein-coding repeat polymorphisms strongly shape diverse human phenotypes. **2021**, 373, 1499-1505	15
567	Clinical autism subscales have common genetic liability that is heritable, pleiotropic, and generalizable to the general population.	0
566	The Opportunities and Challenges of Integrating Population Histories Into Genetic Studies for Diverse Populations: A Motivating Example From Native Hawaiians. **2021**, 12, 643883	0
565	Evaluation of Vicinity-based Hidden Markov Models for Genotype Imputation.	
564	Fast two-stage phasing of large-scale sequence data. **2021**, 108, 1880-1890	8
563	Current Developments in Detection of Identity-by-Descent Methods and Applications. **2021**, 12, 722602	0
562	Leveraging genomic diversity for discovery in an EHR-linked biobank: the UCLA ATLAS Community Health Initiative.	2
561	Tracing the Distribution of European Lactase Persistence Genotypes Along the Americas. **2021**, 12, 671079	0
560	HIF-1Pulmonary Phenotype Wide Association Study Unveils a Link to Inflammatory Airway Conditions. **2021**, 12, 756645	3
559	Stroke Genetics: Turning Discoveries into Clinical Applications. **2021**, 52, 2974-2982	0
558	A population-specific missense variant rs1597000001 in CETP promotes a favorable lipid profile and reduces CETP activity.	
557	Genetic and environmental correlations between complex phenotypes differ by race/ethnicity and sex.	0
556	Pathogenic variants in nucleoporin TPR (translocated promoter region, nuclear basket protein) cause severe intellectual disability in humans. **2021**,	0

555	Strategies to improve pharmacogenomic-guided treatment options for patients with hemoglobinopathies. **2021**, 14, 883-885	
554	SmProt: A Reliable Repository with Comprehensive Annotation of Small Proteins Identified from Ribosome Profiling. **2021**,	1
553	Qatar Genome: Insights on Genomics from the Middle East.	1
552	Open Imputation Server provides secure Imputation services with provable genomic privacy.	0
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550	Evolutionary conservation in noncoding genomic regions. **2021**, 37, 903-918	2
549	Mutations in spike protein and allele variations in ACE2 impact targeted therapy strategies against SARS-CoV-2. **2021**, 42, 170-181	0
548	A unifying framework for rare variant association testing in family-based designs, including higher criticism approaches, SKATs, and burden tests. **2020**,	1
547	Accurate, scalable cohort variant calls using DeepVariant and GLnexus. **2021**,	18
546	Genetic correlates of phenotypic heterogeneity in autism.	2
545	Contribution of enhancer-driven and master-regulator genes to autoimmune disease revealed using functionally informed SNP-to-gene linking strategies.	3
544	Bidirectional Mendelian randomization supports bidirectional causality between telomere length and clonal hematopoiesis of intermediate potential.	1
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542	ARHap: Association Rule Haplotype Phasing. **2021**, PP,	0
541	ConVarT: a search engine for matching human genetic variants with variants from non-human species. **2021**,	2
540	A multi-ethnic whole genome sequencing study to identify novel loci for bone mineral density. **2021**,	0
539	Whole genome sequence analysis of blood lipid levels in >66,000 individuals.	0
538	Integrating de novo and inherited variants in over 42,607 autism cases identifies mutations in new moderate risk genes.	2

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537	Genomic insights into the population history and biological adaptation of Southwestern Chinese Hmong-Mien people.		
536	A massive effort links protein-coding gene variants to health. *Nature*, **2021**, 599, 561-563	50.4	1
535	APOE missense variant R145C is associated with increased Alzheimer disease risk in African ancestry individuals with the APOE /4 genotype.		0
534	Beyond GWAS: from simple associations to functional insights. **2021**, 1		2
533	Association of mitochondrial DNA copy number with cardiometabolic diseases.. **2021**, 1,		1
532	Lymph node-resident dendritic cells drive T2 cell development involving MARCH1. **2021**, 6, eabh0707		2
531	A high-resolution HLA reference panel capturing global population diversity enables multi-ancestry fine-mapping in HIV host response. **2021**, 53, 1504-1516		7
530	Challenges at the APOE locus: A robust quality control approach for accurate APOE genotyping.		0
529	Inferring the mode and strength of ongoing selection.		
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527	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. **2021**, 13, 153		4
526	Integrating thousands of PTEN variant activity and abundance measurements reveals variant subgroups and new dominant negatives in cancers. **2021**, 13, 165		0
525	Adjusting for Common Variant Polygenic Scores Improves Yield in Rare Variant Association Analyses.		0
524	End-Truncated LAMB1 Causes a Hippocampal Memory Defect and a Leukoencephalopathy. **2021**, 90, 962-975		0
523	Exome sequencing and analysis of 454,787 UK Biobank participants. *Nature*, **2021**, 599, 628-634	50.4	34
522	An intermediate effect size variant in UMOD confers risk for chronic kidney disease.		0
521	Genetic polymorphisms in ADRB2 and ADRB1 are associated with differential survival in heart failure patients taking blockers. **2021**,		
520	Purifying selection acts on germline methylation to modify the CpG mutation rate at promoters.		2

519	The limits of personalization in precision medicine: Polygenic risk scores and racial categorization in a precision breast cancer screening trial. **2021**, 16, e0258571	1
518	Mid-pass whole genome sequencing enables biomedical genetic studies of diverse populations. **2021**, 22, 666	
517	A multi-ethnic polygenic risk score is associated with hypertension prevalence and progression throughout adulthood.	0
516	A framework for detecting noncoding rare variant associations of large-scale whole-genome sequencing studies.	0
515	A population-specific reference panel for improved genotype imputation in African Americans. **2021**, 4, 1269	0
514	Biobank-scale inference of ancestral recombination graphs enables genealogy-based mixed model association of complex traits.	0
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509	Mutation saturation for fitness effects at human CpG sites. **2021**, 10,	4
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498	Polygenic risk prediction and SNCA haplotype analysis in a Latino Parkinson disease cohort.	
497	Perfect and imperfect views of ultraconserved sequences. **2021**,	1
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495	MEDTEC Students against Coronavirus: Investigating the Role of Hemostatic Genes in the Predisposition to COVID-19 Severity. **2021**, 11,	1
494	Clinical utility of polygenic risk scores for coronary artery disease. **2021**,	7
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492	CCR5 and Biological Complexity: The Need for Data Integration and Educational Materials to Address Genetic/Biological Reductionism at the Interface of Ethical, Legal, and Social Implications.. **2021**, 12, 790041	1
491	Genetic and functional evidence links a missense variant in to lower LDL and fibrinogen. **2021**, 374, 1221-1227	1
490	Genome-wide analysis of common and rare variants via multiple knockoffs at biobank scale, with an application to Alzheimer disease genetics. **2021**, 108, 2336-2353	0
489	Trellis for efficient data and task management in the VA Million Veteran Program. **2021**, 11, 23229	0
488	Phenome risk classification enables phenotypic imputation and gene discovery in developmental stuttering. **2021**, 108, 2271-2283	3
487	Variant interpretation using population databases: lessons from gnomAD. **2021**,	10
486	APOE Missense Variant R145C is Associated with Increased Alzheimer's Disease Risk in African Ancestry Individuals with the APOE /4 Genotype.	
485	High Coverage Whole Genome Sequencing of the Expanded 1000 Genomes Project Cohort Including 602 Trios.	4
484	NDUFV1 mutations in complex I deficiency: Case reports and review of symptoms. **2021**, 44, e20210149	0

483	Chromosomal imbalances detected via RNA-sequencing in 28 cancers.. **2022**,	1
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479	Expanding the pool of public controls for GWAS via a method for combining genotypes from arrays and sequencing.	
478	First genome-wide association study of esophageal atresia identifies three genetic risk loci at , //, and .. **2022**, 3, 100093	
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458	Pharmacogenetics of Between-Individual Variability in Plasma Clearance of Bedaquiline and Clofazimine in South Africa.. **2022**,	1
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456	Variation and impact of polygenic hematological traits in monogenic sickle cell disease.	0
455	Deborah A. Nickerson (1954 2021). **2022**, 32, v-vi	
454	Genetic modifiers of Huntington disease differentially influence motor and cognitive domains.	0
453	Missense variants causing Wiedemann-Steiner syndrome preferentially occur in the KMT2A-CXXC domain and are accurately classified using AlphaFold2.	
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445	Core and Flanking bHLH-PAS:DNA interactions mediate specificity and drive obesity.		0
444	Challenges at the APOE locus: a robust quality control approach for accurate APOE genotyping.. **2022**, 14, 22		
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440	Collaborative Cohort of Cohorts for COVID-19 Research (C4R) Study: Study Design.. **2022**,		3
439	GCAT|Panel, a comprehensive structural variant haplotype map of the Iberian population from high-coverage whole-genome sequencing.. **2022**,		0
438	missense variant rs373863828 has both direct and indirect effects on type 2 diabetes and fasting glucose in Polynesian peoples living in Samoa and Aotearoa New Zealand.. **2022**, 10,		0
437	RaPID-Query for Fast Identity by Descent Search and Genealogical Analysis.		
436	Genome-Wide Characterization of a Highly Penetrant Form of Hyperlipoproteinemia Associated With Genetically Elevated Cardiovascular Risk.. **2022**, CIRCGEN121003489		1
435	Methylation risk scores are associated with a collection of phenotypes within electronic health record systems.		0
434	Haplotype-enhanced inference of somatic copy number profiles from single-cell transcriptomes.		0
433	Disease association of cyclase-associated protein (CAP): Lessons from gene-targeted mice and human genetic studies.. **2022**, 101, 151207		0
432	The power of genetic diversity in genome-wide association studies of lipids. *Nature*, **2021**,	50.4	24
431	The HUNT Study: a population-based cohort for genetic research.		3
430	Lamina Propria Phagocyte Profiling Reveals Targetable Signaling Pathways in Refractory Inflammatory Bowel Disease. **2022**, 1, 380-392		3

429	Linkage and Association Studies of Substance Use Disorders. **2022**, 177-204	
428	Protein prediction for trait mapping in diverse populations.. **2022**, 17, e0264341	0
427	Genome-wide significant risk loci for mood disorders in the Old Order Amish founder population.	
426	A unified genealogy of modern and ancient genomes.. **2022**, 375, eabi8264	1
425	An association test of the spatial distribution of rare missense variants within protein structures identify Alzheimer's disease-related patterns.. **2022**,	1
424	The combination of autism and exceptional cognitive ability increases risk for suicidal ideation.	0
423	Genome-wide association study of brain arteriolosclerosis.. **2022**, 271678X211066299	0
422	Inverted genomic regions between reference genome builds in humans impact imputation accuracy and decrease the power of association testing.	
421	Association of SPI1 Haplotypes with Altered SPI1 Gene Expression and Alzheimer's Disease Risk.. **2022**,	1
420	Rare Genetic Variants Associated With Myocardial Fibrosis: Multi-Ethnic Study of Atherosclerosis.. **2022**, 9, 804788	0
419	The UCLA ATLAS Community Health Initiative: promoting precision health research in a diverse biobank.	0
418	Integrating external controls in case-control studies improves power for rare-variant tests.. **2022**,	0
417	Rare coding variants in RCN3 are associated with blood pressure.. **2022**, 23, 148	
416	Allele imputation for the killer cell immunoglobulin-like receptor KIR3DL1/S1.. **2022**, 18, e1009059	1
415	Exome-wide association study to identify rare variants influencing COVID-19 outcomes: Results from the Host Genetics Initiative.	0
414	Assessing the contribution of rare variants to complex trait heritability from whole-genome sequence data.. **2022**,	6
413	Whole-genome sequencing as an investigational device for return of hereditary disease risk and pharmacogenomic results as part of the All of Us Research Program.. **2022**, 14, 34	0
412	Discovering missing heritability in whole-genome sequencing data.. **2022**,	1

411	Neurobiological, familial and genetic risk factors for dimensional psychopathology in the Adolescent Brain Cognitive Development study.. **2022**,	0
410	The hematopoietic saga of clonality in sickle cell disease.. **2022**, 132,	1
409	A multi-phenotype analysis reveals 19 novel susceptibility loci for basal cell carcinoma and 15 for squamous cell carcinoma.	0
408	A ZMIZ1 Pathogenic Variant for Neurodevelopmental Disorder With Dysmorphic Facies and Distal Skeletal Anomalies.. **2022**, 13, 840577	0
407	Secondary analyses for genome-wide association studies using expression quantitative trait loci.. **2022**,	0
406	Aspirin and the risk of colorectal cancer according to genetic susceptibility among older individuals.. **2022**,	2
405	ABO O blood group as a risk factor for platelet reactivity in heparin-induced thrombocytopenia.. **2022**,	0
404	Meta-analysis fine-mapping is often miscalibrated at single-variant resolution.	1
403	Nasal airway transcriptome-wide association study of asthma reveals genetically driven mucus pathobiology.. **2022**, 13, 1632	2
402	Whole-genome sequencing of 1,171 elderly admixed individuals from S□ Paulo, Brazil.. **2022**, 13, 1004	4
401	Polygenic risk for prostate cancer: Decreasing relative risk with age but little impact on absolute risk.. **2022**,	0
400	Cohort profile: genomic data for 26 622 individuals from the Canadian Longitudinal Study on Aging (CLSA).. **2022**, 12, e059021	1
399	Fungal microbiome in inflammatory bowel disease: a critical assessment.. **2022**, 132,	2
398	Advances in understanding the molecular basis of clonal hematopoiesis.. **2022**,	0
397	A multi-dimensional integrative scoring framework for predicting functional variants in the human genome.. **2022**, 109, 446-456	1
396	Genetic modifiers of Huntington disease differentially influence motor and cognitive domains.. **2022**,	2
395	The motif composition of variable-number tandem repeats impacts gene expression.	0
394	Polygenic transcriptome risk scores for COPD and lung function improve cross-ethnic portability of prediction in the NHLBI TOPMed program.. **2022**,	0

393	Predicted leukocyte telomere length and risk of germ cell tumours.. **2022**,	
392	MicroRNA-eQTLs in the developing human neocortex link miR-4707-3p expression to brain size.	
391	From pharmacogenetics to pharmaco-omics: Milestones and future directions.. **2022**, 3, 100100	1
390	Diastolic Blood Pressure Alleles Improve Congenital Heart Defect Repair Outcomes.. **2022**, CIRCRESAHA121319842	0
389	An Amish founder population reveals rare-population genetic determinants of the human lipidome.. **2022**, 5, 334	0
388	Accurate genome-wide germline profiling from decade-old archival tissue DNA reveals the contribution of common variants to precancer disease outcome.	0
387	Maternal effect genes as risk factors for congenital heart defects.. **2022**, 3, 100098	0
386	Admixture Mapping of Alzheimer's disease in Caribbean Hispanics identifies a new locus on 22q13.1.. **2022**,	2
385	A complete reference genome improves analysis of human genetic variation.. **2022**, 376, eabl3533	12
384	Impaired neurogenesis alters brain biomechanics in a neuroprogenitor-based genetic subtype of congenital hydrocephalus.. **2022**,	0
383	RAREsim: A simulation method for very rare genetic variants.. **2022**,	
382	Partitioning gene-level contributions to complex-trait heritability by allele frequency identifies disease-relevant genes.. **2022**,	
381	Mendelian randomization supports bidirectional causality between telomere length and clonal hematopoiesis of indeterminate potential.. **2022**, 8, eabl6579	3
380	Novel missense SETD1A variants in Japanese patients with schizophrenia: Resequencing and association analysis.. **2022**, 310, 114481	
379	New insights into the genetic etiology of Alzheimer's disease and related dementias.. **2022**,	27
378	North and East African mitochondrial genetic variation needs further characterization towards precision medicine.	
377	Genomic Characterization of Host Factors Related to SARS-CoV-2 Infection in People with Dementia and Control Populations: The GR@ACE/DEGESCO Study.. **2021**, 11,	2
376	Whole genome sequencing identifies common and rare structural variants contributing to hematologic traits in the NHLBI TOPMed program.	1

375	Driving mosaicism: somatic variants in reference population databases and effect on variant interpretation in rare genetic disease.. **2021**, 15, 71	0
374	Deviation from baseline mutation burden provides powerful and robust rare-variants association test for complex diseases.. **2021**,	1
373	Clonal hematopoiesis is driven by aberrant activation of TCL1A.	1
372	The Michigan Genomics Initiative: a biobank linking genotypes and electronic clinical records in Michigan Medicine patients.	3
371	Clonal hematopoiesis is associated with protection from Alzheimer's disease.	0
370	Genetic Contributors of Incident Stroke in 10,700 African Americans With Hypertension: A Meta-Analysis From the Genetics of Hypertension Associated Treatments and Reasons for Geographic and Racial Differences in Stroke Studies.. **2021**, 12, 781451	
369	Multi-Omic Approaches to Identify Genetic Factors in Metabolic Syndrome.. **2021**, 12, 3045-3084	0
368	A spectrum of recessiveness among Mendelian disease variants in UK Biobank.	0
367	A homozygous CAP2 pathogenic variant in a neonate presenting with rapidly progressive cardiomyopathy and nemaline rods. **2021**,	0
366	De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. **2021**, 6, 104	0
365	A polygenic risk score predicts mosaic loss of chromosome Y in circulating blood cells.. **2021**, 11, 205	0
364	MOMIC: A Multi-Omics Pipeline for Data Analysis, Integration and Interpretation. **2022**, 12, 3987	
363	Genotype error biases trio-based estimates of haplotype phase accuracy.	
362	A spatially aware likelihood test to detect sweeps from haplotype distributions.. **2022**, 18, e1010134	1
361	Genetics of chronic obstructive pulmonary disease: understanding the pathobiology and heterogeneity of a complex disorder.. **2022**,	7
360	Polygenic risk, population structure and ongoing difficulties with race in human genetics.. **2022**, 377, 20200427	1
359	Exome sequencing of families from Ghana reveals known and candidate hearing impairment genes.. **2022**, 5, 369	4
358	How HLA diversity is apportioned: influence of selection and relevance to transplantation.. **2022**, 377, 20200420	2

357	Whole Genome DNA and RNA Sequencing of Whole Blood Elucidates the Genetic Architecture of Gene Expression Underlying a Wide Range of Diseases.. **2022**,	0
356	A loss-of-function IFNAR1 allele in Polynesia underlies severe viral diseases in homozygotes.. **2022**, 219,	2
355	Beyond GWAS of Colorectal Cancer: Evidence of Interaction with Alcohol Consumption and Putative Causal Variant for the 10q24.2 Region.. **2022**, OF1-OF13	0
354	An atlas of genetic scores to predict multi-omic traits.	0
353	Genome-wide studies reveal factors associated with circulating uromodulin and its relations with complex diseases.. **2022**,	1
352	Improved sequence mapping using a complete reference genome and lift-over.	0
351	GWAS of Hematuria.. **2022**,	0
350	The Thousand Polish Genomes-A Database of Polish Variant Allele Frequencies.. **2022**, 23,	4
349	Genome interpretation using in silico predictors of variant impact.. **2022**, 1	0
348	SCN1A Variants as the Underlying Cause of Genetic Epilepsy with Febrile Seizures Plus in Two Multi-Generational Colombian Families. **2022**, 13, 754	
347	Whole-exome sequencing identifies rare genetic variants associated with human plasma metabolites.. **2022**,	0
346	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation.. **2022**,	7
345	Whole exome sequencing identifies novel germline variants of SLC15A4 gene as potentially cancer predisposing in familial colorectal cancer.. **2022**, 1	
344	Genomic risk score for advanced osteoarthritis in older adults.. **2022**,	0
343	Promoting the genomic revolution in Africa through the Nigerian 100K Genome Project.. **2022**, 54, 531-536	1
342	Accounting for population structure in genetic studies of cystic fibrosis. **2022**, 100117	
341	Meta-imputation: An efficient method to combine genotype data after imputation with multiple reference panels.. **2022**,	1
340	TOP-LD: A tool to explore linkage disequilibrium with TOPMed whole-genome sequence data.. **2022**,	0

339	Genotyping and population structure of the China Kadoorie Biobank.	0
338	The translational challenges of precision oncology.. **2022**,	3
337	Independent Replication and Drug-specificity of an Antidepressant Response Polygenic Risk Score.	
336	The Value of Rare Genetic Variation in the Prediction of Common Obesity in European Ancestry Populations.. **2022**, 13, 863893	
335	Gene-based whole genome sequencing meta-analysis of 250 circulating proteins in three isolated European populations.. **2022**, 101509	0
334	Sex differences in the genetic architecture of cognitive resilience to Alzheimer's disease.. **2022**,	0
333	Opportunities and challenges for the use of common controls in sequencing studies.. **2022**,	0
332	Importance of Including Non-European Populations in Large Human Genetic Studies to Enhance Precision Medicine.. **2022**,	2
331	Impact of natural selection on global patterns of genetic variation and association with clinical phenotypes at genes involved in SARS-CoV-2 infection.. **2022**, 119, e2123000119	0
330	A bioinformatics pipeline for estimating mitochondrial DNA copy number and heteroplasmy levels from whole genome sequencing data.. **2022**, 4, lqac034	0
329	Computational and experimental methods for classifying variants of unknown clinical significance.. **2022**, 8,	0
328	SLCO5A1 and synaptic assembly genes contribute to impulsivity in juvenile myoclonic epilepsy.	
327	Association of APOE-Independent Alzheimer Disease Polygenic Risk Score With Brain Amyloid Deposition in Asymptomatic Older Adults. 10.1212/WNL.0000000000200544	
326	Lack of association of CYP2B6 pharmacogenetics with cyclophosphamide toxicity in patients with cancer.	
325	Genomic analyses of 10,376 individuals in the Westlake BioBank for Chinese (WBBC) pilot project. **2022**, 13,	0
324	Lipoprotein(a) beyond the kringle IV repeat polymorphism: The complexity of genetic variation in the LPA gene. **2022**, 349, 17-35	16
323	Detecting Tandem Repeat Expansions Using Short-Read Sequencing for Clinical Use. **2022**, 15-42	0
322	A spectrum of recessiveness among Mendelian disease variants in UK Biobank. **2022**,	0

321	The global prevalence and ethnic heterogenicity of iron-refractory iron deficiency anemia.	
320	Developing and validating a multivariable prediction model which predicts progression of intermediate to late age-related macular degeneration the PINNACLE trial protocol.	1
319	Association of Rare APOE Missense Variants V236E and R251G With Risk of Alzheimer Disease.	2
318	Genotype error biases trio-based estimates of haplotype phase accuracy. **2022**, 109, 1016-1025	0
317	Genetic correlates of phenotypic heterogeneity in autism.	1
316	Network Approaches to Integrate Analyses of Genetics and Metabolomics Data with Applications to Fetal Programming Studies. **2022**, 12, 512	
315	Novel genes and sex differences in COVID-19 severity.	2
314	Corin Missense Variants, Blood Pressure, and Hypertension in 11´322 Black Individuals: Insights From REGARDS and the Jackson Heart Study. **2022**, 11,	
313	XSI - A genotype compression tool for compressive genomics in large biobanks.	1
312	Open problems in human trait genetics. **2022**, 23,	2
311	Building a Vertically-Integrated Genomic Learning Health System: The Colorado Center for Personalized Medicine Biobank.	1
310	Admixture mapping screening of CKD traits and risk factors in U.S. Hispanic/Latino individuals from Central America country-of-origin.	
309	Robust and accurate estimation of paralog-specific copy number for duplicated genes using whole-genome sequencing. **2022**, 13,	
308	Clinical autism subscales have common genetic liabilities that are heritable, pleiotropic, and generalizable to the general population. **2022**, 12,	0
307	A multi-ethnic polygenic risk score is associated with hypertension prevalence and progression throughout adulthood. **2022**, 13,	3
306	Genome-wide meta-analysis of iron status biomarkers and the effect of iron on all-cause mortality in HUNT. **2022**, 5,	0
305	Polygenic risk prediction and SNCA haplotype analysis in a Latino Parkinson's disease cohort. **2022**,	0
304	Patterns and distribution of de novo mutations in multiplex Middle Eastern families.	

303	Insulin Response to Oral Glucose and Cardiometabolic Disease: A Mendelian Randomization Study to Assess Potential Causality.	
302	A de novo start-loss in EFTUD2 associated with mandibulofacial dysostosis with microcephaly: case report. **2022**, 8, a006206	1
301	Missense variants causing Wiedemann-Steiner syndrome preferentially occur in the KMT2A-CXXC domain and are accurately classified using AlphaFold2. **2022**, 18, e1010278	0
300	Elucidation of the genetic causes of bicuspid aortic valve disease.	0
299	Mixed model-based eQTL analysis reveals lncRNAs associated with regulation of genes involved in sex determination and spermatogenesis: The key to understanding human gender imbalance. **2022**, 99, 107713	
298	Current status and future perspectives of the evaluation of missense variants by using three-dimensional structures of proteins. **2022**,	
297	The Location of Missense Variants in the Human GIP Gene Is Indicative for Natural Selection. 13,	
296	On the prospect of achieving accurate joint estimation of selection with population history. **2022**, 14,	1
295	Whole Exome Sequencing Enhanced Imputation Identifies 85 Metabolite Associations in the Alpine CHRIS Cohort. **2022**, 12, 604	0
294	Genetic Determinants in Airways Obstructive Diseases: The Case of Asthma Chronic Obstructive Pulmonary Disease Overlap. **2022**,	
293	Genotyping, sequencing and analysis of 140,000 adults from the Mexico City Prospective Study.	0
292	Investigating the characteristics of genes and variants associated with self-reported hearing difficulty in older adults in the UK Biobank. **2022**, 20,	0
291	Systems Analysis of de novo Mutations in Congenital Heart Diseases Identified a Molecular Network in Hypoplastic Left Heart Syndrome.	
290	Whole-exome sequencing in 415,422 individuals identifies rare variants associated with mitochondrial DNA copy number.	
289	Thousands of human mutation clusters are explained by short-range template switching. gr.276478.121	
288	Genome-wide association study of ACE inhibitor-induced cough implicates neuropeptides and shows genetic overlap with chronic dry cough.	
287	IBD-based estimation of X chromosome effective population size with application to sex-specific demographic history.	
286	Contributions of the Women Health Initiative to Cardiovascular Research. **2022**, 80, 256-275	0

285	Estimating the genome-wide mutation rate from thousands of unrelated individuals.		
284	Continuous Bayesian Variant Interpretation Accounts for Incomplete Penetrance among Mendelian Cardiac Channelopathies.		
283	Polygenic Scores of Alzheimer Disease Risk Genes Add Only Modestly to APOE in Explaining Variation in Amyloid PET Burden. **2022**, 1-11		
282	Use of Polygenic Risk Scores for Coronary Heart Disease in Ancestrally Diverse Populations.		
281	Proteomics and Population Biology in the Cardiovascular Health Study (CHS): design of a study with mentored access and active data sharing.		1
280	A locus at 19q13.31 significantly reduces the ApoE 4 risk for Alzheimer Disease in African Ancestry. **2022**, 18, e1009977		0
279	Detailed stratified GWAS analysis for severe COVID-19 in four European populations.		0
278	How Functional Genomics Can Keep Pace With VUS Identification. 9,		0
277	Whole exome sequencing study identifies candidate loss of function variants and locus heterogeneity in familial cholesteatoma.		
276	Genetic Contributors of Efficacy and Adverse Metabolic Effects of Chlorthalidone in African Americans from the Genetics of Hypertension Associated Treatments (GenHAT) Study. **2022**, 13, 1260		0
275	Including diverse and admixed populations in genetic epidemiology research.		0
274	A minimal role for synonymous variation in human disease.		0
273	Des recommandations fran ises pour le d ficit en alpha1-antitrypsine´; enfin !. **2022**,		
272	Insights From a Large-Scale Whole-Genome Sequencing Study of Systolic Blood Pressure, Diastolic Blood Pressure, and Hypertension. **2022**, 79, 1656-1667		1
271	The sequences of 150,119 genomes in the UK Biobank. *Nature*, **2022**, 607, 732-740	50.4	6
270	Genome-wide association study in minority children with asthma implicates DNAH5 in bronchodilator responsiveness. **2022**, 12,		0
269	CMDB: the comprehensive population genome variation database of China.		1
268	Recommendations on the use and reporting of race, ethnicity, and ancestry in genetic research: Experiences from the NHLBI TOPMed program. **2022**, 100155		0

267	Germline Mutations in CIDEB and Protection against Liver Disease. **2022**, 387, 332-344	1
266	Imputation of ancient genomes.	0
265	Germline genomic and phenomic landscape of clonal hematopoiesis in 323,112 individuals.	1
264	dRFEtools: Dynamic recursive feature elimination for omics.	0
263	Whole-genome sequencing of the UK Biobank.	0
262	Multiset correlation and factor analysis enables exploration of multi-omic data.	0
261	Fixing reference errors efficiently improves sequencing results.	0
260	Whole genome sequence association analysis of fasting glucose and fasting insulin levels in diverse cohorts from the NHLBI TOPMed program. **2022**, 5,	1
259	Loss-of-function variants in SAT1 cause X-linked childhood-onset systemic lupus erythematosus. annrheumdis-2022-2227	
258	Fast, accurate local ancestry inference with FLARE.	0
257	Common Variants Near ZIC1 and ZIC4 in Autopsy-Confirmed Multiple System Atrophy.	1
256	Quantitative trait locus (xQTL) approaches identify risk genes and drug targets from non-coding genome findings.	0
255	Identification of a de novo mutation of FOXG1gene and comprehensive analysis for molecular factors in Chinese FOXG1-related Rett syndrome.	
254	Recurrent mutation in the ancestry of a rare variant.	
253	Non-linear machine learning models incorporating SNPs and PRS improve polygenic prediction in diverse human populations. **2022**, 5,	0
252	Secure secondary utilization system of genomic data using quantum secure cloud.	
251	Nontargeted and Targeted Metabolomic Profiling Reveals Novel Metabolite Biomarkers of Incident Diabetes in African Americans.	0
250	DDX58 is Associated with Susceptibility to Severe Influenza Virus Infection in Children and Adolescents.	

249	An intermediate-effect size variant in UMOD confers risk for chronic kidney disease. **2022**, 119,	1
248	A survey of genome-wide association studies, polygenic scores and UK Biobank highlights resources for autoimmune disease genetics. 13,	
247	MUC5B, telomere length and longitudinal quantitative interstitial lung changes: the MESA Lung Study. thoraxjnl-2021-218139	
246	Genome-wide association study of ischemic stroke risk in Sickle Cell confirms ADAMTS2, CDK18, uncovers 12 novel loci.	
245	GAWMerge expands GWAS sample size and diversity by combining array-based genotyping and whole-genome sequencing. **2022**, 5,	
244	Life-Course Associations between Blood Pressure-Related Polygenic Risk Scores and Hypertension in the Bogalusa Heart Study. **2022**, 13, 1473	1
243	Integrating de novo and inherited variants in 42,607 autism cases identifies mutations in new moderate-risk genes.	2
242	Scalable approaches for functional analyses of whole-genome sequencing non-coding variants.	
241	Real-World Evaluation of a Population Germline Genetic Screening Initiative for Family Medicine Patients. **2022**, 12, 1297	0
240	Grand challenges in stroke genomics. 1,	
239	Methylation risk scores are associated with a collection of phenotypes within electronic health record systems. **2022**, 7,	1
238	Multiancestry exome sequencing reveals INHBE mutations associated with favorable fat distribution and protection from diabetes. **2022**, 13,	0
237	Proteomic profiling platforms head to head: Leveraging genetics and clinical traits to compare aptamer- and antibody-based methods. **2022**, 8,	2
236	Rare genetic variants explain missing heritability in smoking.	0
235	Extent to which array genotyping and imputation with large reference panels approximate deep whole-genome sequencing. **2022**,	0
234	Single-cell multi-cohort dissection of the schizophrenia transcriptome.	0
233	Systems analysis of de novo mutations in congenital heart diseases identified a protein network in the hypoplastic left heart syndrome. **2022**,	0
232	Analyses of rare predisposing variants of lung cancer in 6,004 whole genomes in Chinese. **2022**,	0

231	Cystatin C is associated with adverse COVID-19 outcomes in diverse populations. **2022**, 25, 105040	1
230	Challenges Related to the Use of Next-Generation Sequencing for the Optimization of Drug Therapy. **2022**,	0
229	Evolving Approaches to Identifying Genetic Risk Variants for Sleep Disorders. **2022**, 3-20	0
228	SNPs Classification and Terminology: dbSNP Reference SNP (rs) Gene and Consequence Annotation. **2022**, 3-12	0
227	Single Nucleotide Polymorphisms (SNPs). **2022**,	0
226	FAVOR: Functional Annotation of Variants Online Resource and Annotator for Variation across the Human Genome.	0
225	Evaluation of vicinity-based hidden Markov models for genotype imputation. **2022**, 23,	0
224	Pharmacokinetic and pharmacogenetic associations with dolutegravir neuropsychiatric adverse events in an African population.	0
223	Endophenotype effect sizes support variant pathogenicity in monogenic disease susceptibility genes. **2022**, 13,	0
222	Ancestry-driven recalibration of tumor mutational burden and disparate clinical outcomes in response to immune checkpoint inhibitors. **2022**,	0
221	Leveraging genomic diversity for discovery in an electronic health record linked biobank: the UCLA ATLAS Community Health Initiative. **2022**, 14,	0
220	Multimodal biomedical AI. **2022**, 28, 1773-1784	5
219	Polygenic risk scores for dyslipidemia and atherosclerotic cardiovascular disease: progress toward clinical implementation. **2022**, 101702	0
218	Diverse ancestry whole-genome sequencing association study identifies TBX5 and PTK7 as susceptibility genes for posterior urethral valves. 11,	0
217	Interpreting the spectrum of gamma-secretase complex missense variation in the context of hidradenitis suppurativa—An in-silico study. 13,	1
216	Genetic Regulators of Sputum Mucin Concentration and Their Associations with COPD Phenotypes.	0
215	Whole-exome sequencing in 415,422 individuals identifies rare variants associated with mitochondrial DNA copy number. **2022**, 100147	0
214	Association of Whole-Person Eigen-Polygenic Risk Scores with Alzheimer's Disease.	0

213	Towards a global view of multiple sclerosis genetics. **2022**, 18, 613-623	1
212	Comparison of calling pipelines for whole genome sequencing: an empirical study demonstrating the importance of mapping and alignment.	0
211	Compressed Data Structures for Population-Scale Positional Burrows Wheeler Transforms.	0
210	The Phenotype-Genotype Reference Map: Improving biobank data science through replication.	0
209	Use of race, ethnicity, and ancestry data in health research. **2022**, 2, e0001060	0
208	Landscape of germline cancer predisposition mutations testing and management in pediatrics: Implications for research and clinical care. 10,	0
207	EagleImp: fast and accurate genome-wide phasing and imputation in a single tool.	1
206	Genetic structure correlates with ethnolinguistic diversity in eastern and southern Africa. **2022**, 109, 1667-1679	0
205	Rapid, Reference-Free human genotype imputation with denoising autoencoders. 11,	0
204	Social and scientific motivations to move beyond groups in allele frequencies: The TOPMed experience. **2022**, 109, 1582-1590	0
203	Haplotype-aware analysis of somatic copy number variations from single-cell transcriptomes.	0
202	A Haptoglobin (HP) Structural Variant Alters the Effect of APOE Alleles on Alzheimer Disease.	0
201	Leveraging interindividual variability of regulatory activity for refining genetic regulation of gene expression in schizophrenia.	0
200	TIVAN-indel: A computational framework for annotating and predicting noncoding regulatory small insertion and deletion.	0
199	High-coverage whole-genome sequencing of the expanded 1000 Genomes Project cohort including 602 trios. **2022**, 185, 3426-3440.e19	9
198	Deep neural networks with controlled variable selection for the identification of putative causal genetic variants. **2022**, 4, 761-771	0
197	MagicalRsq: Machine-learning-based genotype imputation quality calibration. **2022**,	0
196	Assessing a multivariate model of brain-mediated genetic influences on disordered eating in the ABCD cohort.	0

195	Proteomic analysis of diabetes genetic risk scores identifies complement C2 and neuropilin-2 as predictors of type 2 diabetes: the Atherosclerosis Risk in Communities (ARIC) Study.	0
194	SVAT: Secure outsourcing of variant annotation and genotype aggregation. **2022**, 23,	0
193	Characterizing alcohol expectancies in the ABCD Study: associations with familial and psychosocial factors and relevant polygenic scores.	0
192	GABBR1 monoallelic de novo variants linked to neurodevelopmental delay and epilepsy. **2022**, 109, 1885-1893	2
191	The power of TOPMed imputation for the discovery of Latino enriched rare variants associated with type 2 diabetes.	0
190	Transcriptional and genetic sex differences for schizophrenia across the dorsolateral prefrontal cortex, hippocampus, and caudate nucleus.	0
189	Complement component C4 structural variation and quantitative traits contribute to sex-biased vulnerability in systemic sclerosis. **2022**, 7,	0
188	Human Genomics, History. **2022**,	0
187	Accurate rare variant phasing of whole-genome and whole-exome sequencing data in the UK Biobank.	0
186	Trans-ancestry meta-analysis of genome wide association studies of inhibitory control.	0
185	Genome-wide association study identifies multiple HLA loci for sarcoidosis susceptibility.	0
184	The new uORFdb: integrating literature, sequence, and variation data in a central hub for uORF research.	1
183	Evolutionary coupling analysis guides identification of mistrafficking-sensitive variants in cardiac K+ channels: Validation with hERG. 13,	0
182	Clonal Hematopoiesis of Indeterminate Potential and Kidney Function Decline in the General Population. **2022**,	0
181	The contribution of functional HNF1A variants and polygenic susceptibility to risk of type 2 diabetes in ancestrally diverse populations.	0
180	Whole genome sequence analysis of blood lipid levels in >66,000 individuals. **2022**, 13,	1
179	A saturated map of common genetic variants associated with human height. **2022**, 610, 704-712	2
178	Investigating the role of common and rare variants in multiplex multiple sclerosis families reveals an increased burden of common risk variation. **2022**, 12,	0

177	Advances and Applications of Polygenic Scores for Coronary Artery Disease. **2023**, 74,	0
176	The HUNT study: A population-based cohort for genetic research. **2022**, 2, 100193	0
175	Potential Misrepresentation of Inherited Breast Cancer Risk by Common Germline Alleles.	0
174	The association between mitochondrial DNA copy number, low-density lipoprotein cholesterol, and cardiovascular disease risk.	0
173	PRKCE non-coding variants influence on transcription as well as translation of its gene. **2022**, 19, 1115-1129	0
172	STAARpipeline: an all-in-one rare-variant tool for biobank-scale whole-genome sequencing data.	0
171	Why are GWASs limp about the X chromosome?.	0
170	Using human genetics to improve safety assessment of therapeutics.	0
169	A framework for detecting noncoding rare-variant associations of large-scale whole-genome sequencing studies.	1
168	Human Molecular Genetics Review Issue 2022. **2022**, 31, R1-R3	0
167	Genome-wide Analysis of Rare Haplotypes Associated with Breast Cancer Risk: Discovery, Replication, and Generalizability Evaluation.	0
166	FGF20 and PGM2 variants are associated with childhood asthma in family-based whole-genome sequencing studies.	0
165	Improving polygenic risk prediction in admixed populations by explicitly modeling ancestral-specific effects via GAUDI.	0
164	Rare coding variants inCHRNB2reduce the likelihood of smoking.	0
163	Exome-wide association study to identify rare variants influencing COVID-19 outcomes: Results from the Host Genetics Initiative. **2022**, 18, e1010367	0
162	Shared genetic risk factors and causal association between psoriasis and coronary artery disease. **2022**, 13,	0
161	Microglial efferocytosis: Diving into the Alzheimer disease gene pool. **2022**, 110, 3513-3533	1
160	Secure secondary utilization system of genomic data using quantum secure cloud. **2022**, 12,	0

159	Analysis of the caudate nucleus transcriptome in individuals with schizophrenia highlights effects of antipsychotics and new risk genes. **2022**, 25, 1559-1568	2
158	Inverted genomic regions between reference genome builds in humans impact imputation accuracy and decrease the power of association testing. **2022**, 100159	0
157	Global perspectives on CYP2D6 associations with primaquine metabolism and Plasmodium vivax radical cure. 13,	0
156	Mosaic chromosomal alterations in blood across ancestries via whole-genome sequencing.	0
155	Genome-wide admixture and association analysis identifies African ancestry specific risk loci of eosinophilic esophagitis in African American. **2022**,	1
154	Shared genetic risk across different presentations of gene test□egative idiopathic nephrotic syndrome.	0
153	FAVOR: functional annotation of variants online resource and annotator for variation across the human genome.	0
152	Meta-analysis fine-mapping is often miscalibrated at single-variant resolution. **2022**, 100210	0
151	Uncovering Cross-Cohort Molecular Features with Multi-Omics Integration Analysis.	0
150	Estimating the genome-wide mutation rate from thousands of unrelated individuals. **2022**,	0
149	Identification of PCSK9-like human gene knockouts using metabolomics, proteomics, and whole-genome sequencing in a consanguineous population. **2022**, 100218	0
148	Genome-wide Analysis of Rare Haplotypes Associated with Breast Cancer Risk.	0
147	From the reference human genome to human pangenome: Premise, promise and challenge. 13,	0
146	Interrogating the Human Diplome: Computational Methods, Emerging Applications, and Challenges. **2023**, 1-30	0
145	A comprehensive knowledgebase of known and predicted human genetic variants associated with COVID-19 susceptibility and severity.	0
144	Association of a Multi-Ancestry Genome-Wide Blood Pressure Polygenic Risk Score with Adverse Cardiovascular Events.	0
143	Genetic Modifiers of Sickle Cell Disease. **2022**, 36, 1097-1124	0
142	Different effects of low muscle mass on the risk of non-alcoholic fatty liver disease and hepatic fibrosis in a prospective cohort.	0

141	The combination of autism and exceptional cognitive ability is associated with suicidal ideation. **2023**, 197, 107698	0
140	A Phenome-Wide Association Study (Phewas) Of´Late Onset Alzheimer Disease Genetic Risk in Children of European Ancestry at Middle Childhood: Results From the ABCD Study.	0
139	IMMerge: merging imputation data at scale.	0
138	Clonal Hematopoiesis and Its Impact on Human Health. **2023**, 74,	0
137	Whole genome DNA and RNA sequencing of whole blood elucidates the genetic architecture of gene expression underlying a wide range of diseases. **2022**, 12,	0
136	Unappreciated Subcontinental Admixture in Europeans and European Americans: Implications for Genetic Epidemiology Studies.	0
135	Can genomic research make a useful contribution to social policy?. **2022**, 9,	0
134	Analyzing the Korean reference genome with meta-imputation increased the imputation accuracy and spectrum of rare variants in the Korean population. 13,	0
133	Imputation of low-coverage sequencing data from 150,119 UK Biobank genomes.	2
132	GhostKnockoff inference empowers identification of putative causal variants in genome-wide association studies. **2022**, 13,	0
131	Whole-exome sequencing study identifies four novel gene loci associated with diabetic kidney disease.	0
130	Eight pharmacokinetic genetic variants are not associated with the risk of bleeding from direct oral anticoagulants in non-valvular atrial fibrillation patients. 13,	1
129	POIROT: A powerful test for parent-of-origin effects in unrelated samples leveraging multiple phenotypes.	0
128	Common and rare variant associations with clonal haematopoiesis phenotypes. **2022**, 612, 301-309	1
127	A High-resolution Haplotype-resolved Reference Panel Constructed from the China Kadoorie Biobank Study.	0
126	Rare variants in SLC34A3 explain missing heritability of urinary stone disease.	0
125	Analytic pipelines to assess the relationship between immune response and germline genetics in human tumors. **2022**, 3, 101809	0
124	The missing link between genetic association and regulatory function. 11,	0

123	Alzheimer's Disease Heterogeneity Explained by Polygenic Risk Scores Derived from Brain Transcriptomic Profiles.	0
122	Identification of a de novo mutation of the FOXG1 gene and comprehensive analysis for molecular factors in Chinese FOXG1-related encephalopathies. 15,	0
121	Identification of reduced ERAP2 expression and a novel HLA allele as components of a risk score for susceptibility to liver injury due to amoxicillin-clavulanate. **2022**,	2
120	Disparities in Inherited Retinal Degenerations. 1-6	0
119	Comparison of calling pipelines for whole genome sequencing: an empirical study demonstrating the importance of mapping and alignment. **2022**, 12,	0
118	Genome-wide association study of multiple neuropathology endophenotypes identifies novel risk loci and provides insights into known Alzheimer's risk loci.	0
117	A Mendelian randomization study to assess the genetic liability of type 1 diabetes mellitus for IgA nephropathy. 13,	0
116	A minimal role for synonymous variation in human disease. **2022**, 109, 2105-2109	0
115	Monopogen: single nucleotide variant calling from single cell sequencing.	0
114	Correlations between complex human phenotypes vary by genetic background, gender, and environment. **2022**, 3, 100844	1
113	Powerful, scalable and resource-efficient meta-analysis of rare variant associations in large whole genome sequencing studies.	0
112	Whole genome sequencing identifies structural variants contributing to hematologic traits in the NHLBI TOPMed program. **2022**, 13,	0
111	Whole-genome sequencing across 449 samples spanning 47 ethnolinguistic groups provides insights into genetic diversity in Nigeria.	0
110	Altered and allele-specific open chromatin landscape reveals epigenetic and genetic regulators of innate immunity in COVID-19. **2022**, 100232	1
109	XCVATR: detection and characterization of variant impact on the Embeddings of single -cell and bulk RNA-sequencing samples. **2022**, 23,	0
108	A survey of proteomic variation across two ethnic groups in Nigeria and its relationship to obesity risk.	0
107	Polygenic Parkinson's disease genetic risk score as risk modifier of parkinsonism in Gaucher disease.	0
106	The NBDC-DDBJ imputation server facilitates the use of controlled access reference panel datasets in Japan. **2022**, 9,	0

105	A multi-phenotype analysis reveals 19 susceptibility loci for basal cell carcinoma and 15 for squamous cell carcinoma. **2022**, 13,	0
104	Editorial: Forensic investigative genetic genealogy and fine-scale structure of human populations. 13,	0
103	Fast, accurate local ancestry inference with FLARE. **2023**,	0
102	15 years of GWAS discovery: Realizing the promise. **2023**,	0
101	Poison exon annotations improve the yield of clinically relevant variants in genomic diagnostic testing.	0
100	Genomic atlas of the plasma metabolome prioritizes metabolites implicated in human diseases. **2023**, 55, 44-53	0
99	Genome-wide association study of early ischaemic stroke risk in Brazilian individuals with sickle cell disease implicates ADAMTS2 and CDK18 and uncovers novel loci.	0
98	MicroRNA-eQTLs in the developing human neocortex link miR-4707-3p expression to brain size. 12,	0
97	The UCLA ATLAS Community Health Initiative: Promoting precision health research in a diverse biobank. **2023**, 3, 100243	0
96	The global prevalence and ethnic heterogeneity of iron-refractory iron deficiency anaemia. **2023**, 18,	0
95	Integration of single-cell multiomic measurements across disease states with genetics identifies mechanisms of beta cell dysfunction in type 2 diabetes.	0
94	Accurate genome-wide genotyping from archival tissue to explore the contribution of common genetic variants to pre-cancer outcomes. **2022**, 20,	0
93	Proteomics Analysis of Genetic Liability of Abdominal Aortic Aneurysm Identifies Plasma Neogenin and Kit Ligand: The ARIC Study.	0
92	xAtlas: scalable small variant calling across heterogeneous next-generation sequencing experiments. **2022**, 12,	1
91	A comparison between similarity matrices for principal component analysis to assess population stratification in sequenced genetic data sets. **2023**, 24,	0
90	The Singapore National Precision Medicine Strategy.	0
89	First GWAS on Alzheimer's Disease in Argentina and Chile populations.	0
88	A harmonized public resource of deeply sequenced diverse human genomes.	0

87	Multi-ancestry transcriptome-wide association analyses yield insights into tobacco use biology and drug repurposing.	0
86	Exploring the genetic diversity of the Japanese Population: Insights from a Large-Scale Whole Genome Sequencing Analysis.	0
85	Gene burden analysis identifies genes associated with increased risk and severity of adult-onset hearing loss in a diverse hospital-based cohort. **2023**, 19, e1010584	0
84	Is the Association of the Rare rs35667974 IFIH1 Gene Polymorphism With Autoimmune Diseases a Case of RNA Epigenetics?.	0
83	Characterizing Alcohol Expectancies in the ABCD Study: Associations with Sociodemographic Factors, the Immediate Social Environment, and Genetic Propensities.	0
82	Common genetic variants contribute to heritability of age at onset of schizophrenia.	0
81	COVID-19 Mortality in the Colorado Center for Personalized Medicine Biobank. **2023**, 20, 2368	0
80	TIVAN-indel: a computational framework for annotating and predicting non-coding regulatory small insertions and deletions. **2023**, 39,	0
79	Structural variation across 138,134 samples in the TOPMed consortium.	0
78	LoFTK: a framework for fully automated calculation of predicted Loss-of-Function variants and genes. **2023**, 16,	0
77	Design and implementation of a hybrid cloud system for large-scale human genomic research. **2023** , 10,	0
76	Polygenic scores for tobacco use provide insights into systemic health risks in a diverse EHR-linked biobank in Los Angeles.	0
75	Multi-ancestry genome-wide study in >2.5 million individuals reveals heterogeneity in mechanistic pathways of type 2 diabetes and complications.	0
74	Polygenic Parkinson's Disease Genetic Risk Score as Risk Modifier of Parkinsonism in Gaucher Disease.	0
73	Rare variant analyses in large-scale cohorts identified SLC13A1 associated with chronic pain. **2023**, Publish Ahead of Print,	0
72	Translating non-coding genetic associations into a better understanding of immune-mediated disease. **2023**, 16,	0
71	Genetic and environmental contributions to ancestry differences in gene expression in the human brain.	0
70	Fast and accurate Bayesian polygenic risk modeling with variational inference. **2023**,	0

69	Aberrant activation of TCL1A promotes stem cell expansion in clonal haematopoiesis.	0
68	Pharmacotherapy exposure as a marker of disease complexity in bipolar disorder: Associations with clinical & genetic risk factors. **2023**, 323, 115174	0
67	Nutrigenomics in the context of evolution. **2023**, 62, 102656	0
66	Germline rare deleterious variant load alters cancer risk, age of onset and tumor characteristics. **2023**, 7,	0
65	The Michigan Genomics Initiative: A biobank linking genotypes and electronic clinical records in Michigan Medicine patients. **2023**, 3, 100257	0
64	Association of Rare Protein-Truncating DNA Variants in APOB or PCSK9 With Low-density Lipoprotein Cholesterol Level and Risk of Coronary Heart Disease. **2023**, 8, 258	1
63	Human IRF1 governs macrophagic IFN-□mmunity to mycobacteria. **2023**, 186, 621-645.e33	1
62	North and East African mitochondrial genetic variation needs further characterization towards precision medicine. **2023**,	0
61	Integrative genetics-metabolomics analysis of infant bronchiolitis-childhood asthma link: A multicenter prospective study. 13,	0
60	X-Chromosome Association Study in Latin American Cohorts Identifies New Loci in Parkinson Disease.	0
59	eXclusionarY: Ten years later, where are the sex chromosomes in GWAS?.	0
58	Probable Treatment Targets for Diabetic Retinopathy Based on an Integrated Proteomic and Genomic Analysis. **2023**, 12, 8	0
57	WGS Data Collections: How Do Genomic Databases Transform Medicine?. **2023**, 24, 3031	0
56	Cross-continental environmental and genome-wide association study on children and adolescent anxiety and depression.	0
55	Multivariate adaptive shrinkage improves cross-population transcriptome prediction for transcriptome-wide association studies in underrepresented populations.	0
54	Inferring a directed acyclic graph of phenotypes from GWAS summary statistics*.	0
53	Increasing serum iron levels and their role in the risk of infectious diseases: a Mendelian randomization approach.	0
52	Apolipoprotein C3 induces inflammasome activation only in its delipidated form. **2023**, 24, 408-411	0

51	Extensive set of African ancestry-informative markers (AIMs) to study ancestry and population health. 14,	0
50	An Oxylipin-Related Nutrient Pattern and Risk of Type 1 Diabetes in the Diabetes Autoimmunity Study in the Young (DAISY). **2023**, 15, 945	1
49	Modifications of the endosomal compartment in fibroblasts from sporadic Alzheimer disease patients are associated with cognitive impairment. **2023**, 13,	0
48	PBWT: Enabling the Storage and Use of UK Biobank Data on a Commodity Laptop.	0
47	Genome-wide polygenic risk score for major osteoporotic fractures in postmenopausal women using associated single nucleotide polymorphisms. **2023**, 21,	0
46	Whole-genome sequencing analysis in families with recurrent pregnancy loss: A pilot study. **2023**, 18, e0281934	0
45	Computational prediction of human deep intronic variation.	0
44	Association of African Ancestry Specific APOE Missense Variant R145C With Risk of Alzheimer Disease. **2023**, 329, 551	0
43	FixItFelix: improving genomic analysis by fixing reference errors. **2023**, 24,	0
42	Matching Variants for functional characterization of genetic variants.	0
41	Comparison of the structure-function properties of wild-type human apoA-V and a C-terminal truncation associated with elevated plasma triglycerides.	0
40	Hacking hematopoiesis emerging tools for examining variant effects. **2023**, 16,	0
39	Genome-Wide Association Studies and fine-mapping of genomic loci for n-3 and n-6 Polyunsaturated Fatty Acids in Hispanic American and African American Cohorts.	0
38	Whole-genome sequencing of ethnolinguistic diverse northwestern Chinese Hexi Corridor people from the 10K_CPGDP project suggested the differentiated East-West genetic admixture along the Silk Road and their biological adaptations.	0
37	Why does the X chromosome lag behind autosomes in GWAS findings?. **2023**, 19, e1010472	0
36	Unified views on variant impact across many diseases. **2023**,	0
35	Multiomic analyses implicate a neurodevelopmental program in the pathogenesis of cerebral arachnoid cysts. **2023**, 29, 667-678	0
34	Genome-wide significant risk loci for mood disorders in the Old Order Amish founder population.	0

33	Split-Transformer Impute (STI): Genotype Imputation Using a Transformer-Based Model.	0
32	Evolutionary constraint and innovation across hundreds of placental mammals.	0
31	Leveraging Base Pair Mammalian Constraint to Understand Genetic Variation and Human Disease.	0
30	Diversity ofCFTRvariants across ancestries characterized using 454,727 UK biobank whole exome sequences.	0
29	Inferring feature importance with uncertainties with application to large genotype data. **2023**, 19, e1010963	0
28	Polygenic prediction of bipolar disorder in a Latin American sample.	0
27	Whole exome sequencing study identifies candidate loss of function variants and locus heterogeneity in familial cholesteatoma. **2023**, 18, e0272174	0
26	Polygenic scores for tobacco use provide insights into systemic health risks in a diverse EHR-linked biobank in Los Angeles.	0
25	Pharmacogenomic and Statistical Analysis. **2023**, 305-330	0
24	Dual impact of PTEN mutation on CSF dynamics and cortical networks via the dysregulation of neural precursors and their interneuron descendants.	0
23	MultiomicsTracks96: A high throughput PIXUL-Matrix-based toolbox to profile frozen and FFPE tissues multiomes.	0
22	Adjusting for common variant polygenic scores improves yield in rare variant association analyses. **2023**, 55, 544-548	0
21	Whole Genome Analysis of Venous Thromboembolism: the Trans-Omics for Precision Medicine Program. **2023**, 16,	0
20	Correlates of Risk for Disinhibited Behaviors in the Million Veteran Program Cohort.	0
19	Multi-source data approach for personalized outcome prediction in lung cancer screening: update from the NELSON trial. **2023**, 38, 445-454	0
18	Genome-wide analysis of individual coding variants and HLA-II-associated self-immunopeptidomes in ulcerative colitis.	0
17	Genome-wide analysis of oxylipins and oxylipin profiles in a pediatric population. 10,	0
16	Topological Optimisation Structure Design for Personalisation of Hydrogel Controlled Drug Delivery System. **2023**, 16, 2687	0

15	An atlas of genetic scores to predict multi-omic traits. **2023**, 616, 123-131	0
14	Association of polygenic scores with chronic kidney disease phenotypes in a longitudinal study of older adults. **2023**,	0
13	Comparing Pruning and Thresholding with Continuous Shrinkage Polygenic Score Methods in a Large Sample of Ancestrally Diverse Adolescents from the ABCD Study .	0
12	StocSum: stochastic summary statistics for whole genome sequencing studies.	0
11	FOXI3 pathogenic variants cause one form of craniofacial microsomia. **2023**, 14,	0
10	A Novel Variant in VPS13B Underlying Cohen Syndrome. **2023**, 2023, 1-7	0
9	Optimal HLA imputation of admixed population with dimension reduction.	0
8	GBC: a parallel toolkit based on highly addressable byte-encoding blocks for extremely large-scale genotypes of species. **2023**, 24,	0
7	A Phenome-Wide Association Study (PheWAS) of Late Onset Alzheimer Disease Genetic Risk in Children of European Ancestry at Middle Childhood: Results from the ABCD Study.	0
6	Pharmacogenetics of tenofovir renal toxicity in HIV-positive Southern Africans. Publish Ahead of Print,	0
5	The genetic determinants of recurrent somatic mutations in 43,693 blood genomes. **2023**, 9,	0
4	Evolutionary constraint and innovation across hundreds of placental mammals. **2023**, 380,	0
3	The power of TOPMed imputation for the discovery of Latino-enriched rare variants associated with type 2 diabetes.	0
2	Genome-wide Association Identifies Novel Etiological Insights Associated with Parkinson Disease in African and African Admixed Populations.	0
1	Biological and pharmacological functions of the FGF19- and FGF21-coreceptor beta klotho. 14,	0