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A global reference for human genetic variation

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334	Why do Diseases Start One Sided? Clues From HLA-B27 Acute Anterior Uveitis. 76-82	О
334	Why do Diseases Start One Sided? Clues From HLA-B27 Acute Anterior Uveitis. 76-82 Whole Exome Sequencing for the Diagnosis of Rare Genetic Neurodevelopmental Disorders Associated with Cerebellar Atrophy.	0
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226	New Genetic Insights in Rheumatoid Arthritis using Taxonomy3 $\ \ \ \ \ \ \ \ \ \ \ \ \ \ \ \ \ \ \$	0
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181	Genome Analysis Using Whole-Exome Sequencing of Non-Syndromic Cleft Lip and/or Palate from Malagasy Trios Identifies Variants Associated with Cilium-Related Pathways and Asian Genetic Ancestry. 2023 , 14, 665	O
180	Demographic Modeling of Admixed Latin American Populations from Whole Genomes.	O
179	Genome wide association study based on clustering by obesity-related variables shed light on a genetic architecture of obesity in Japanese and UK population.	O
178	Circulating white blood cell traits and colorectal cancer risk: A Mendelian randomization study.	О
177	Chromosome-scale Genome Assembly of the Yellow Nutsedge (Cyperus esculentus). 2023, 15,	O
176	Improved computations for relationship inference using low-coverage sequencing data. 2023, 24,	О
175	Functional and regulatory diversification of circadian rhythmperiodgenes during the evolution of vertebrates.	O
174	A crowdsourcing database for the copy-number variation of the Spanish population. 2023, 17,	О
173	Split-Transformer Impute (STI): Genotype Imputation Using a Transformer-Based Model.	O
172	Recent advances in CRISPR-based genome editing technology and its applications in cardiovascular research. 2023 , 10,	0
171	Identifying polymorphic cis-regulatory variants as risk markers for lung carcinogenesis and chemotherapy responses in tobacco smokers from eastern India. 2023 , 13,	O
170	Longitudinal Reading Measures and Genome Imputation in the National Child Development Study: Prospects for Future Reading Research. 1-11	0
169	Associations of atrial natriuretic peptide with measures of insulin and adipose depots. 2023, 11,	O
168	Rescuing early Parkinson-induced hyposmia prevents dopaminergic system failure.	O
167	Leveraging Base Pair Mammalian Constraint to Understand Genetic Variation and Human Disease.	O
166	The genetic overlap between AlzheimerâʿʿB disease, amyotrophic lateral sclerosis, Lewy body dementia, and ParkinsonâʿʿB disease. 2023 ,	O
165	Shared genetic architecture between attention-deficit/hyperactivity disorder and lifespan.	O

164	A deep population reference panel of tandem repeat variation.	0
163	ancIBD - Screening for identity by descent segments in human ancient DNA.	o
162	The genetic basis of endometriosis and comorbidity with other pain and inflammatory conditions. 2023 , 55, 423-436	0
161	The genetic architecture of pain intensity in a sample of 598,339 U.S. veterans.	o
160	dbAQP-SNP: a database of missense single-nucleotide polymorphisms in human aquaporins. 2023 , 2023,	0
159	Genetically predicted circulating levels of cytokines and the risk of osteoarthritis: A mendelian randomization study. 14,	O
158	The genetic architecture of pain intensity in a sample of 598,339 U.S. veterans.	O
157	Learning a restricted Boltzmann machine using biased Monte Carlo sampling. 2023, 14,	О
156	Inferring biological kinship in ancient datasets: comparing the response of ancient DNA-specific software packages to low coverage data. 2023 , 24,	O
155	Genetic architecture of spatial electrical biomarkers for cardiac arrhythmia and relationship with cardiovascular disease. 2023 , 14,	o
154	Genome-wide analysis of genetic pleiotropy and causal genes across three age-related ocular disorders. 2023 , 142, 507-522	O
153	A retrospective analysis of preemptive pharmacogenomic testing in 22,918 individuals from China. 2023 , 37,	O
152	Evaluating performance and applications of sample-wise cell deconvolution methods on human brain transcriptomic data.	O
151	Association of atopic dermatitis with conjunctivitis and other ocular surface diseases: A bidirectional two-sample Mendelian randomization study.	O
150	Exploration of genotype-by-environment interactions affecting gene expression responses in porcine immune cells. 14,	0
149	Hypercholesterolemia in the Malaysian Cohort Participants: Genetic and Non-Genetic Risk Factors. 2023 , 14, 721	o
148	NOMe-HiC: joint profiling of genetic variant, DNA methylation, chromatin accessibility, and 3D genome in the same DNA molecule. 2023 , 24,	0
147	Unlocking the genome of perch âlFrom genes to ecology and back again.	O

146	Statistical Methods for Disease Risk Prediction with Genotype Data. 2023, 331-347	0
145	Polygenic scores for tobacco use provide insights into systemic health risks in a diverse EHR-linked biobank in Los Angeles.	O
144	Standardizing and applying a mating-based whole-genome simulation approach reveals caution in using chromosome-level PCA and kinship estimates.	0
143	An Ensemble Penalized Regression Method for Multi-ancestry Polygenic Risk Prediction.	O
142	Enrichment of self-domestication and neural crest function loci in the heritability of neurodevelopmental disorders.	0
141	Complete sequences of six major histocompatibility complex haplotypes, including all the major MHC class II structures.	O
140	Evaluation of polygenic score for hypertrophic cardiomyopathy in the general population and across clinical settings.	0
139	PDIVAS: Pathogenicity predictor for Deep-Intronic Variants causing Aberrant Splicing.	О
138	Genome-wide association study identifies four pan-ancestry loci for suicidal ideation in the Million Veteran Program. 2023 , 19, e1010623	0
137	Models based on best-available information support a low inbreeding load and potential for recovery in the vaquita. 2023 , 130, 183-187	O
136	Maternal and fetal origins of offspring blood pressure: statistical analysis using genetic correlation and genetic risk score-based Mendelian randomization.	O
135	Distribution of a novel CYP2C haplotype in Native American populations. 14,	O
134	Genetic and Environmental interactions contribute to immune variation in rewilded mice.	0
133	Interactions of genetic risks for autism and the broad autism phenotypes. 14,	O
132	IL16 and factor V gene variations are associated with asparaginase-related thrombosis in childhood acute lymphoblastic leukemia patients. 2023 , 24, 199-206	0
131	Genomic analyses of hair from Ludwig van Beethoven. 2023 ,	O
130	Investigating Vitamin D Receptor Genetic Markers in a Cluster Headache Meta-Analysis. 2023 , 24, 5950	0
129	A theoretical base for non-invasive prenatal paternity testing. 2023 , 346, 111649	O

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124	Current allele distribution of the human longevity gene APOE in Europe can mainly be explained by ancient admixture.	O
123	An HLA map of the world: A comparison of HLA frequencies in 200 worldwide populations reveals diverse patterns for class I and class II. 14,	О
122	Dried fruit intake causally protects against low back pain: A Mendelian randomization study. 10,	О
121	Homologous recombination deficiency signatures in gastrointestinal and thoracic cancers correlate with platinum therapy duration. 2023 , 7,	O
120	Genome-wide association study of Klebsiella pneumoniae urinary tract infection in Taiwanese patients identifies potential genetic risk factors.	О
119	A common variant rs2054564 in ADAMTS17 is associated with susceptibility to lumbar spondylosis. 2023 , 13,	О
118	Analysis of evolutionary dynamics and clonal architecture in prostate cancer.	0
117	Causal effects on complex traits are similar for common variants across segments of different continental ancestries within admixed individuals. 2023 , 55, 549-558	O
116	Inferring CpG methylation signatures accumulated along human history from genetic variation catalogs.	O
115	The role of a multicentre data repository in ocular inflammation: The Ocular Autoimmune Systemic Inflammatory Infectious Study (OASIS).	o
114	A genome-wide association study of frailty identifies significant genetic correlation with neuropsychiatric, cardiovascular, and inflammation pathways.	0
113	Tuberculosis severity associates with variants and eQTLs related to vascular biology and infection-induced inflammation. 2023 , 19, e1010387	o
112	Multi-source data approach for personalized outcome prediction in lung cancer screening: update from the NELSON trial. 2023 , 38, 445-454	0
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108	Genome-wide association study of population-standardised cognitive performance phenotypes in a rural South African community. 2023 , 6,	0
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106	Real-world disparities and ethical considerations with access to CFTR modulator drugs: Mind the gap!. 14,	0
105	Reference-free phylogeny from sequencing data. 2023 , 16,	О
104	Gene-by-Sex Interactions: Genome-Wide Association Study Reveals Five SNPs Associated with Obesity and Overweight in a Male Population. 2023 , 14, 799	0
103	Pharmacogenetics and Adverse Events in the Use of Fluoropyrimidine in a Cohort of Cancer Patients on Standard of Care Treatment in Zimbabwe. 2023 , 13, 588	O
102	The genetics of incontinence: A scoping review.	O
101	A regulatory variant at 19p13.3 is associated with primary biliary cholangitis risk and ARID3A expression. 2023 , 14,	О
100	Do Poor Diet and Lifestyle Behaviors Modify the Genetic Susceptibility to Impulsivity in the General Population?. 2023 , 15, 1625	0
99	Epigenome-wide association study of serum folate in maternal peripheral blood leukocytes. 2023 , 15, 39-52	О
98	Deletion mapping of regulatory elements for GATA3 in T´cells reveals a distal enhancer involved in allergic diseases. 2023 , 110, 703-714	0
97	Evolutionary Genetics and Admixture in African Populations. 2023, 15,	О
96	Phenome-wide Mendelian randomization study of plasma triglyceride levels and 2600 disease traits. 12,	O
95	An atlas of genetic scores to predict multi-omic traits. 2023 , 616, 123-131	O
94	The impact of damaging epilepsy and cardiac genetic variant burden in sudden death in the young.	0
93	Tensor decomposition based feature extraction and classification to detect natural selection from genomic data.	O

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91	A scalable variational approach to characterize pleiotropic components across thousands of human diseases and complex traits using GWAS summary statistics.	O
90	Somatic and germline aberrations in homologous recombination repair genes in Chinese prostate cancer patients. 13,	О
89	Comprehensive evaluations of individual discrimination, kinship analysis, genetic relationship exploration and biogeographic origin prediction in Chinese Dongxiang group by a 60-plex DIP panel. 2023 , 160,	O
88	Whole-exome screening for primary congenital glaucoma in Lebanon. 1-12	О
87	The EN-TEx resource of multi-tissue personal epigenomes & wariant-impact models. 2023, 186, 1493-15	l1. e 40
86	A single-nucleus transcriptome-wide association study implicates novel genes in depression pathogenesis.	0
85	Statistical examination of shared loci in neuropsychiatric diseases using genome-wide association study summary statistics.	O
84	Frequencies of variants in genes associated with dyslipidemias identified in Costa Rican genomes. 14,	O
83	A Complex Intrachromosomal Rearrangement Disrupting IRF6 in a Family with Popliteal Pterygium and Van der Woude Syndromes. 2023 , 14, 849	O
82	Estimating the Prevalence of LAMA2 Congenital Muscular Dystrophy using Population Genetic Databases. 2023 , 1-7	0
81	Analysis of genetic dominance in the UK Biobank. 2023 , 379, 1341-1348	O
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76	Colocalization of blood cell traits GWAS associations and variation in PU.1 genomic occupancy prioritizes causal noncoding regulatory variants.	О
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73	Identifying the potential causal role of insomnia symptoms on 11,409 health-related outcomes: a phenome-wide Mendelian randomisation analysis in UK Biobank. 2023 , 21,	o
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71	Genetic impacts on DNA methylation help elucidate regulatory genomic processes.	О
70	Two New Cases of Bachmannâ B upp Syndrome Identified through the International Center for Polyamine Disorders. 2023 , 11, 29	O
69	Genetic associations among internalizing and externalizing traits with polysubstance use among young adults.	О
68	Comparing Pruning and Thresholding with Continuous Shrinkage Polygenic Score Methods in a Large Sample of Ancestrally Diverse Adolescents from the ABCD Study $\!$	О
67	OTTERS: a powerful TWAS framework leveraging summary-level reference data. 2023, 14,	О
66	A global view of the genetic basis of Alzheimer disease.	0
65	StocSum: stochastic summary statistics for whole genome sequencing studies.	О
64	The causal association between smoking initiation, alcohol and coffee consumption, and womenâß reproductive health: A two-sample Mendelian randomization analysis. 14,	0
63	Julia for biologists.	o
62	Human variation impacting MCOLN2 restricts Salmonella Typhi replication by magnesium deprivation. 2023 , 100290	0
61	Multiomics of human aortic endothelial cells reveals cell subtypes with heterogeneous responses to canonical endothelial-to-mesenchymal perturbations.	o
60	Echoes from the last Green Sahara: whole genome analysis of Fulani, a key population to unveil the genetic evolutionary history of Africa.	0
59	GWAS for Systemic Sclerosis Identified six novel susceptibility loci including penetrating FcEReceptor Region.	O
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55	Recent advances in Forensic DNA Phenotyping of appearance, ancestry and age. 2023 , 65, 102870	Ο
54	Identification of Compound Heterozygous EVC2 Gene Variants in Two Mexican Families with EllisâNan Creveld Syndrome. 2023 , 14, 887	0
53	High Polygenic Risk is Associated with Earlier Initiation and Escalation of Treatment in Early Primary Open Angle Glaucoma. 2023 ,	O
52	A gene-level test for directional selection on gene expression.	Ο
51	A resampling-based approach to share reference panels.	O
50	Genetics and epigenetics in the obesity phenotyping scenario.	Ο
49	Brain structure, phenotypic and genetic correlates of reading performance.	Ο
48	Revisiting Genetic Epidemiology with a Refined Targeted Gene Panel for Hereditary Hearing Impairment in the Taiwanese Population. 2023 , 14, 880	0
47	Effect of schizophrenia common variants on infant brain volumes: cross-sectional study in 207 term neonates in developing Human Connectome Project. 2023 , 13,	O
46	Influence of Genetics on the Response to Omalizumab in Patients with Severe Uncontrolled Asthma with an Allergic Phenotype. 2023 , 24, 7029	0
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44	omicSynth: an Open Multi-omic Community Resource for Identifying Druggable Targets across Neurodegenerative Diseases.	0
43	Developing an evolutionary baseline model for humans: jointly inferring purifying selection with population history.	O
42	A Novel Variant in VPS13B Underlying Cohen Syndrome. 2023 , 2023, 1-7	0
41	GRAPE: genomic relatedness detection pipeline. 11, 589	O
40	The Impact of Stability Considerations on Genetic Fine-Mapping.	0
39	Optimal HLA imputation of admixed population with dimension reduction.	Ο

38	Variant calling and benchmarking in an era of complete human genome sequences.	О
37	Celiac Disease Is a Risk Factor for Mature T and NK Cell Lymphoma: A Mendelian Randomization Study. 2023 , 24, 7216	О
36	TEAD1 trapping by the Q353Râllamin A/C causes dilated cardiomyopathy. 2023 , 9,	O
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34	Sex-biased gene regulation varies across human populations as a result of adaptive evolution.	0
33	Ultra-fast genotyping of SNPs and short indels using GPU acceleration.	О
32	Heritability Estimation Approaches Utilizing Genome-Wide Data. 2023, 3,	O
31	Identifying genetic variants for amyloid 🛭 n subcortical vascular cognitive impairment. 15,	О
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28	mitoSplitter: A mitochondrial variants-based method for efficient demultiplexing of pooled single-cell RNA-seq.	o
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26	Genotype-by-environment interactions in chronic back pain. 2023,	0
25	Multi-omic underpinnings of epigenetic aging and human longevity. 2023, 14,	О
24	A rarefaction approach for measuring population differences in rare and common variation.	0
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19	Diverse evolutionary rates and gene duplication patterns among families of functional olfactory receptor genes in humans. 2023 , 18, e0282575	O
18	Prevalence of Monogenic Bone Disorders in a Dutch Cohort of Atypical Femur Fracture Patients.	O
17	Wrestling with Social and Behavioral Genomics: Risks, Potential Benefits, and Ethical Responsibility. 2023 , 53,	O
16	Kernel-based genetic association analysis for microbiome phenotypes identifies host genetic drivers of beta-diversity. 2023 , 11,	O
15	Ovarian Cancer and Parkinsonâ Disease: A Bidirectional Mendelian Randomization Study. 2023 , 12, 2961	O
14	Single-cell genomics meets human genetics.	0
13	Genomics in reproductive medicine: Current and future applications. 2023, 695-719	O
12	PRDM1 DNA-binding zinc finger domain is required for normal limb development and is disrupted in split hand/foot malformation. 2023 , 16,	0
11	Next-generation sequencing for gene panels, clinical exome, and whole-genome analysis. 2023 , 743-766	O
10	Potential utility of risk stratification for multicancer screening with liquid biopsy tests. 2023, 7,	O
9	Ability of a polygenic risk score to refine colorectal cancer risk in Lynch syndrome.	O
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7	Idiopathic pulmonary fibrosis and the role of genetics in the era of precision medicine. 10,	O
6	Genetic, clinical, and pathological study of patients with severe hypertension-associated renal microangiopathy.	O
5	The power of TOPMed imputation for the discovery of Latino-enriched rare variants associated with type 2 diabetes.	O
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Identification of neuropathology-based subgroups in multiple sclerosis using a data-driven approach.

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