Resource Summary Report

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1000 Genomes: A Deep Catalog of Human Genetic Variation

RRID:SCR_006828
Type: Tool

Proper Citation

1000 Genomes: A Deep Catalog of Human Genetic Variation (RRID:SCR_006828)

Resource Information

URL: http://www.1000genomes.org/

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Description: International collaboration producing an extensive public catalog of human genetic variation, including SNPs and structural variants, and their haplotype contexts, in an effort to provide a foundation for investigating the relationship between genotype and phenotype. The genomes of about 2500 unidentified people from about 25 populations around the world were sequenced using next-generation sequencing technologies. Redundant sequencing on various platforms and by different groups of scientists of the same samples can be compared. The results of the study are freely and publicly accessible to researchers worldwide. The consortium identified the following populations whose DNA will be sequenced: Yoruba in Ibadan, Nigeria; Japanese in Tokyo; Chinese in Beijing; Utah residents with ancestry from northern and western Europe; Luhya in Webuye, Kenya; Maasai in Kinyawa, Kenya; Toscani in Italy; Gujarati Indians in Houston; Chinese in metropolitan Denver; people of Mexican ancestry in Los Angeles; and people of African ancestry in the southwestern United States. The goal Project is to find most genetic variants that have frequencies of at least 1% in the populations studied. Sequencing is still too expensive to deeply sequence the many samples being studied for this project. However, any particular region of the genome generally contains a limited number of haplotypes. Data can be combined across many samples to allow efficient detection of most of the variants in a region. The Project currently plans to sequence each sample to about 4X coverage; at this depth sequencing cannot provide the complete genotype of each sample, but should allow the detection of most variants with frequencies as low as 1%. Combining the data from 2500 samples should allow highly accurate estimation (imputation) of the variants and genotypes for each sample that were not seen directly by the light sequencing. All samples from the

1000 genomes are available as lymphoblastoid cell lines (LCLs) and LCL derived DNA from the Coriell Cell Repository as part of the NHGRI Catalog. The sequence and alignment data generated by the 1000genomes project is made available as quickly as possible via their mirrored ftp sites. ftp://ftp.1000genomes.ebi.ac.uk ftp://ftp-trace.ncbi.nlm.nih.gov/1000genomes

Abbreviations: 1000 Genomes

Synonyms: International 1000 Genomes Project, 1000 Genomes Project

Resource Type: portal, data or information resource, consortium, database, data set, organization portal

Keywords: genetic variation, gene, next-generation sequencing, sequence, alignment, genome, single-nucleotide polymorphism, structural variant, haplotype, genome-wide association study, pharmacology, genetics, biomarker, consortium, data sharing, genotype, phenotype, FASEB list

Funding: Wellcome Trust Sanger Institute; Hinxton; United Kingdom ;
Beijing Genomics Institute; Shenzhen; China ;
NHGRI ;
454 Life Sciences Roche ;
Life Technologies ;
Illumina

Availability: Free, Public, Restrictions apply,
Http://www.1000genomes.org/data#DataAccess

Resource Name: 1000 Genomes: A Deep Catalog of Human Genetic Variation

Resource ID: SCR_006828

Alternate IDs: nlx_143819, OMICS_00261

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Ratings and Alerts

No rating or validation information has been found for 1000 Genomes: A Deep Catalog of Human Genetic Variation.

No alerts have been found for 1000 Genomes: A Deep Catalog of Human Genetic Variation.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 5011 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

Liu Z, et al. (2025) Circulating tumor DNA analysis for prediction of prognosis and molecular insights in patients with resectable gastric cancer: results from a prospective study. MedComm, 6(2), e70065.

Lake AM, et al. (2025) Sexual Trauma, Polygenic Scores, and Mental Health Diagnoses and Outcomes. JAMA psychiatry, 82(1), 75.

Zurel H, et al. (2025) Characterization of Y chromosome diversity in newfoundland and labrador: evidence for a structured founding population. European journal of human genetics : EJHG, 33(1), 98.

Wang C, et al. (2025) Integrating electronic health records and GWAS summary statistics to predict the progression of autoimmune diseases from preclinical stages. Nature communications, 16(1), 180.

Pampari A, et al. (2025) ChromBPNet: bias factorized, base-resolution deep learning models of chromatin accessibility reveal cis-regulatory sequence syntax, transcription factor footprints and regulatory variants. bioRxiv : the preprint server for biology.

Eulalio T, et al. (2025) regionalpcs improve discovery of DNA methylation associations with complex traits. Nature communications, 16(1), 368.

Gálvez-Montosa F, et al. (2025) Polymorphisms within autophagy-related genes as susceptibility biomarkers for pancreatic cancer: A meta-analysis of three large European cohorts and functional characterization. International journal of cancer, 156(2), 339.

Iida N, et al. (2025) Systematically developing a registry of splice-site creating variants utilizing massive publicly available transcriptome sequence data. Nature communications, 16(1), 426.

Koptekin D, et al. (2025) Pre-processing of paleogenomes: mitigating reference bias and postmortem damage in ancient genome data. Genome biology, 26(1), 6.

Cheng L, et al. (2025) Circulating Tumor DNA Detection for Recurrence Monitoring of Stage I Non-Small Cell Lung Cancer Treated With Microwave Ablation. Thoracic cancer, 16(2), e15534.

Martins Rodrigues F, et al. (2025) Germline predisposition in multiple myeloma. iScience, 28(1), 111620.

Zhou M, et al. (2025) A copy number variation detection method based on OCSVM algorithm using multi strategies integration. Scientific reports, 15(1), 3526.

Mowlaei ME, et al. (2025) STICI: Split-Transformer with integrated convolutions for genotype imputation. Nature communications, 16(1), 1218.

Kanjira SC, et al. (2025) Polygenic prediction of major depressive disorder and related traits in African ancestries UK Biobank participants. Molecular psychiatry, 30(1), 151.

Liu Y, et al. (2025) Novel genetic variants in the NLRP3 inflammasome-related PANX1 and APP genes predict survival of patients with hepatitis B virus-related hepatocellular carcinoma. Clinical & translational oncology : official publication of the Federation of Spanish Oncology Societies and of the National Cancer Institute of Mexico, 27(2), 630.

Weng LC, et al. (2025) The impact of common and rare genetic variants on bradyarrhythmia development. Nature genetics, 57(1), 53.

Yang L, et al. (2025) A novel de novo GABRA2 gene missense variant causing developmental epileptic encephalopathy in a Chinese patient. Annals of clinical and translational neurology, 12(1), 137.

Victor Atoki A, et al. (2025) Exploring the versatility of Drosophila melanogaster as a model organism in biomedical research: a comprehensive review. Fly, 19(1), 2420453.

Davis CN, et al. (2025) Utility of Candidate Genes From an Algorithm Designed to Predict Genetic Risk for Opioid Use Disorder. JAMA network open, 8(1), e2453913.

Zeng Y, et al. (2025) Mapping the chromothripsis landscape in urothelial carcinoma unravels great intratumoral and intertumoral heterogeneity. iScience, 28(1), 111510.