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: consortium, data or information resource, data set, database, organization portal, 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: r3d100010180, nlx_143819, OMICS_00261

Alternate URLs: https://doi.org/10.17616/R3CP4M

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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 5881 mentions in open access literature.

Listed below are recent publications. The full list is available at kravitz2 .

Kawazu M, et al. (2026) Genomic and Transcriptomic Analysis of High-Grade Endometrial Carcinoma Reveals Biological Heterogeneity and Molecular Classification Challenges. Cancer research communications, 6(4), 961.

Tang C, et al. (2026) Investigating the determinants of immunotherapy response in the primary tumour of clear cell renal cell carcinoma (RCC). BMJ oncology, 5(2), e001031.

Camerini L, et al. (2026) Harsh parenting and rs11621961 at the SERPINA6/1 locus: gene-environment interaction effects on hair cortisol in a Brazilian population-based longitudinal study. Stress (Amsterdam, Netherlands), 29(1), 2611613.

Kore P, et al. (2026) Interplay between Genetic Ancestry, Self-reported Race and Ethnicity, and Clinical Factors in Pediatric Acute Lymphoblastic Leukemia: A REDIAL Consortium Report. Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology, 35(5), 710.

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Lyu S, et al. (2026) Brain age gap as biomarker linking cardiovascular diseases genetic susceptibility and causality. iScience, 29(3), 114987.

Ma Y, et al. (2026) Gene-culture coevolution shapes olfactory receptor gene diversity in Orang Asli populations. Cell reports, 117181.

Lee JK, et al. (2026) The Pan-Tumor Landscape of Gene Amplifications and Copy Number Amplification Ratio for Established and Emerging Clinical Targets. Clinical cancer research : an official journal of the American Association for Cancer Research, 32(13), 2653.

Hu X, et al. (2026) Spatial2GWAS: a database for linking spatial transcriptomic regions with GWAS traits. Nucleic acids research, 54(D1), D1309.

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Shabnaz S, et al. (2026) Causal Effects of Hydrophilic Bile Acids on Carfilzomib-Related Cardiovascular Events in Multiple Myeloma: A Mendelian Randomization Study. Clinical pharmacology and therapeutics, 119(6), 1494.

Luo Y, et al. (2026) AI-based multiomics profiling reveals complementary omics contributions to personalized prediction of cardiovascular disease. Nature communications, 17(1).

Nicholas JC, et al. (2026) Cross-ancestry comparison of aptamer and antibody protein measures. Nature communications, 17(1), 1054.

Zheng W, et al. (2026) Comprehensive clinical and genetic architecture of familial amyotrophic lateral sclerosis in China: A 15-year cohort study with 302 families. Neural regeneration research, 21(6), 2573.

Nishiyama NC, et al. (2026) eQTL in diseased colon tissue identifies potential target genes associated with IBD. Nature communications, 17(1).

Priebe O, et al. (2026) Haplotype-aware segmentation with HapASeg increases accuracy of detecting homolog-specific somatic copy number alterations. Genome biology, 27(1).

Favier A, et al. (2026) COBT: a gene-based rare variant burden test for case-only study designs using aggregated genotypes from public reference cohorts. Genome medicine, 18(1).

Cheung C, et al. (2026) APOE and plasma AD biomarkers: The role of genetic ancestry in Hispanics/Latinos. Alzheimer's & dementia : the journal of the Alzheimer's Association, 22(3), e71213.