

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: data set, data or information resource, organization portal, database, consortium, 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](http://www.1000genomes.org/data#DataAccess)

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Resource ID: SCR_006828

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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 5486 mentions in open access literature.

Listed below are recent publications. The full list is available at [nidm-terms](#) .

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.

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M??kitie O, et al. (2025) A de novo PRPF8 Pathogenic Variant in Transient Severe Hypophosphatemia with Delayed Puberty and Growth Failure. *Hormone research in paediatrics*, 98(6), 677.

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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.

Blandin C, et al. (2025) Genetics and Bone Mineral Density Predict the Fractures in Adults With Osteogenesis Imperfecta: A Prospective Study. *The Journal of clinical endocrinology and metabolism*, 110(8), 2307.

Le Borgne J, et al. (2025) X-chromosome-wide association study for Alzheimer's disease. *Molecular psychiatry*, 30(6), 2335.

Wonkam A, et al. (2025) FLT1 and other candidate fetal haemoglobin modifying loci in sickle cell disease in African ancestries. *Nature communications*, 16(1), 2092.

Fiorica PN, et al. (2025) Germline Pathogenic DROSHA Variants Are Linked to Pineoblastoma and Wilms Tumor Predisposition. *Clinical cancer research : an official journal of the American Association for Cancer Research*, 31(8), 1491.