

Resource Summary Report

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Genome Aggregation Database

RRID:SCR_014964

Type: Tool

Proper Citation

Genome Aggregation Database (RRID:SCR_014964)

Resource Information

URL: <http://gnomad.broadinstitute.org/>

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Description: Database that aggregates exome and genome sequencing data from large-scale sequencing projects. The gnomAD data set contains individuals sequenced using multiple exome capture methods and sequencing chemistries. Raw data from the projects have been reprocessed through the same pipeline, and jointly variant-called to increase consistency across projects.

Abbreviations: gnomAD

Synonyms: gnomAD 2.0, gnomAD Browser, gnomAD version 2.0, Exome Aggregation Consortium

Resource Type: data or information resource, database

Keywords: database, genome, , bio.tools, FASEB list

Funding: Broad Institute

Availability: Open source, Available to the biomedical community, The community can contribute to this resource

Resource Name: Genome Aggregation Database

Resource ID: SCR_014964

Alternate IDs: biotools:gnomad

Alternate URLs: https://github.com/macarthur-lab/gnomad_browser/issues,
<https://bio.tools/gnomad>

License: ODC Open Database License

License URLs: <http://gnomad.broadinstitute.org/terms>

Record Creation Time: 20220129T080323+0000

Record Last Update: 20260801T191012+0000

Ratings and Alerts

No rating or validation information has been found for Genome Aggregation Database.

No alerts have been found for Genome Aggregation Database.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 4229 mentions in open access literature.

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

Nishina S, et al. (2026) Novel biallelic CDK9 variants are associated with retinal dystrophy without CHARGE-like malformation syndrome. Journal of human genetics, 71(2), 97.

Browne IM, et al. (2026) The Prognostic and Predictive Impact of ctDNA Levels in Patients with Advanced Breast Cancer Enrolled on the plasmaMATCH Trial. Clinical cancer research : an official journal of the American Association for Cancer Research, 32(1), 148.

Fischer MC, et al. (2026) Splicing and frameshift variants in QSER1 may be involved in developmental phenotypes. HGG advances, 7(1), 100539.

Zhao XC, et al. (2026) Unexpectedly high levels of normally spliced transcripts from the pathogenic SLC10A7 alleles in a recessive form of skeletal dysplasia. HGG advances, 7(1), 100545.

Xu H, et al. (2026) CUL1 variants cause severe neurodevelopmental disorders: Insights from human genetics and a zebrafish model of microcephaly. HGG advances, 7(1), 100542.

Drost M, et al. (2026) Routine RNA-based analysis of potential splicing variants facilitates genomic diagnostics and reveals limitations of in silico prediction tools. HGG advances, 7(1), 100521.

Ncir WB, et al. (2026) First LDLRAP1 and Recurrent LDLR Mutations in Tunisian Families With Familial Hypercholesterolemia. Journal of cellular and molecular medicine, 30(1), e70997.

Leban T, et al. (2026) Multiomics Data Synthesis of FAM83H in Amelogenesis Imperfecta. International dental journal, 76(1), 109293.

- Watanabe T, et al. (2026) Whole exome sequencing in Japanese spinocerebellar ataxia identifies novel variants. *Journal of human genetics*, 71(1), 35.
- Elliott MJ, et al. (2025) Ultrasensitive Detection and Monitoring of Circulating Tumor DNA Using Structural Variants in Early-Stage Breast Cancer. *Clinical cancer research : an official journal of the American Association for Cancer Research*, 31(8), 1520.
- Chauhan PS, et al. (2025) Genomic and Epigenomic Analysis of Plasma Cell-Free DNA Identifies Stemness Features Associated with Worse Survival in Lethal Prostate Cancer. *Clinical cancer research : an official journal of the American Association for Cancer Research*, 31(1), 151.
- Minami S, et al. (2025) Auditory neuropathy spectrum disorder and related auditory features in patients with hearing loss associated with the MT-TS1 m.7471dup variant. *Mitochondrion*, 84, 102056.
- Deng M, et al. (2025) Mutational landscape of glomus tumor and clinical application of genomic profiling based on next-generation sequencing technology. *Pathology, research and practice*, 275, 156230.
- Gaikwad AS, et al. (2025) Functional and clinical insights into nuclear receptor variants for advancing precision diagnostics in male infertility. *EBioMedicine*, 119, 105899.
- Krishnan T, et al. (2025) Clonal Hematopoiesis of Indeterminate Potential and its Association with Treatment Outcomes and Adverse Events in Patients with Solid Tumors. *Cancer research communications*, 5(1), 66.
- Suda K, et al. (2025) Low OLFM1 and BMP6 Expression Predicts Recurrence in Early-Stage Nonsquamous NSCLC with Pure Solid Tumor Appearance. *Cancer research communications*, 5(12), 2186.
- Arenillas C, et al. (2025) Convergent Genetic Adaptation in Human Tumors Developed Under Systemic Hypoxia and in Populations Living at High Altitudes. *Cancer discovery*, 15(5), 1037.
- Gharbi W, et al. (2025) Functional genetic variants in immune-checkpoint molecules and susceptibility to Nasopharyngeal carcinoma in a Tunisian case-control study. *Molecular biology reports*, 53(1), 230.
- Stanciu A, et al. (2025) Personalized ctDNA detection and genomic profiling in the NeoRHEA Study. *NPJ breast cancer*, 11(1), 137.
- Bouzaki A, et al. (2025) Integration of Dose Surface Maps and Genetic Data Identifies the Lower Posterior Rectum as a Key Region for Toxicity after Prostate Cancer Radiotherapy. *Clinical cancer research : an official journal of the American Association for Cancer Research*, 31(23), 5006.