Resource Summary Report

Generated by RRID on Apr 28, 2025

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: database, data or information resource

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: 20250428T053849+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 3746 mentions in open access literature.

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

Smith TB, et al. (2025) Bi-allelic variants in DAP3 result in reduced assembly of the mitoribosomal small subunit with altered apoptosis and a Perrault-syndrome-spectrum phenotype. American journal of human genetics, 112(1), 59.

Alayoubi AM, et al. (2025) Zellweger syndrome; identification of mutations in PEX19 and PEX26 gene in Saudi families. Annals of medicine, 57(1), 2447400.

Van Haute L, et al. (2025) Pathogenic PDE12 variants impair mitochondrial RNA processing causing neonatal mitochondrial disease. EMBO molecular medicine, 17(1), 193.

Lutz S, et al. (2025) Unveiling the Digital Evolution of Molecular Tumor Boards. Targeted oncology, 20(1), 27.

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.

Bayam E, et al. (2025) Bi-allelic variants in WDR47 cause a complex neurodevelopmental syndrome. EMBO molecular medicine, 17(1), 129.

Verhoeven WMA, et al. (2025) X-Linked Autism Type 9 Caused by a Hemizygote Pathogenic Variant in the TMLHE Gene: Etiological Diagnosis in an Adult Male with Moderate Intellectual Disability. International medical case reports journal, 18, 111.

Katsonis P, et al. (2025) Meta-EA: a gene-specific combination of available computational tools for predicting missense variant effects. Nature communications, 16(1), 159.

Scherer N, et al. (2025) Coupling metabolomics and exome sequencing reveals graded effects of rare damaging heterozygous variants on gene function and human traits. Nature genetics, 57(1), 193.

Robinson K, et al. (2025) Rare variants in PRKCI cause Van der Woude syndrome and other features of peridermopathy. medRxiv : the preprint server for health sciences.

Zhang S, et al. (2025) Exploratory analysis of a Novel RACK1 mutation and its potential role in epileptic seizures via Microglia activation. Journal of neuroinflammation, 22(1), 27.

Fishman V, et al. (2025) GENA-LM: a family of open-source foundational DNA language models for long sequences. Nucleic acids research, 53(2).

Park JE, et al. (2025) Carrier Frequency and Incidence of MUTYH-Associated Polyposis Based on Database Analysis in East Asians and Koreans. Annals of laboratory medicine, 45(1), 77.

Englisch C, et al. (2025) Association of clonal haematopoiesis with recurrent venous thromboembolism: A case-control study. British journal of haematology, 206(1), 263.

Kesdiren E, et al. (2025) Heterozygous variants in the teashirt zinc finger homeobox 3 (TSHZ3) gene in human congenital anomalies of the kidney and urinary tract. European journal of human genetics : EJHG, 33(1), 44.

Huang X, et al. (2025) Mutation spectra and genotype?phenotype analysis of congenital hypothyroidism in a neonatal population. Biomedical reports, 22(2), 30.

Kokuryo T, et al. (2025) Whole-genome Sequencing Analysis of Bile Tract Cancer Reveals Mutation Characteristics and Potential Biomarkers. Cancer genomics & proteomics, 22(1), 34.

Biar CG, et al. (2025) Curated loci prime editing (cliPE) for accessible multiplexed assays of variant effect (MAVEs). ArXiv.

Magistrati M, et al. (2025) De Novo DNM1L Pathogenic Variant Associated with Lethal Encephalocardiomyopathy-Case Report and Literature Review. International journal of molecular sciences, 26(2).

Huang C, et al. (2025) Comparative genetic analysis of blood and semen samples in sperm

donors from Hunan, China. Annals of medicine, 57(1), 2447421.