Resource Summary Report

Generated by NIF on Aug 1, 2026

ExAc

RRID:SCR_004068
Type: Tool

Proper Citation

ExAc (RRID:SCR_004068)

Resource Information

URL: http://exac.broadinstitute.org/

Proper Citation: ExAc (RRID:SCR_004068)

Description: THIS RESOURCE IS NO LONGER IN SERVICE. Documented on January 9, 2023. An aggregated data platform for genome sequencing data created by a coalition of investigators seeking to aggregate and harmonize exome sequencing data from a variety of large-scale sequencing projects, and to make summary data available for the wider scientific community. The data set provided on this website spans 61,486 unrelated individuals sequenced as part of various disease-specific and population genetic studies. They have removed individuals affected by severe pediatric disease, so this data set should serve as a useful reference set of allele frequencies for severe disease studies. All of the raw data from these projects have been reprocessed through the same pipeline, and jointly variant-called to increase consistency across projects. They ask that you not publish global (genome-wide) analyses of these data until after the ExAC flagship paper has been published, estimated to be in early 2015. If you''re uncertain which category your analyses fall into, please email them. The aggregation and release of summary data from the exomes collected by the Exome Aggregation Consortium has been approved by the Partners IRB (protocol 2013P001477, Genomic approaches to gene discovery in rare neuromuscular diseases).

Abbreviations: ExAC

Synonyms: Exome Aggregation Consortium, ExAC Browser

Resource Type: data or information resource, database

Defining Citation: PMID:27899611

Keywords: exome sequencing, exome, sequencing, variant, grch37/hg19, gene, region, transcript, multi-allelic variant, FASEB list

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: ExAc

Resource ID: SCR_004068

Alternate IDs: nlx_158505, r3d100010468

Alternate URLs: https://doi.org/10.17616/R3QP53

Record Creation Time: 20220129T080222+0000

Record Last Update: 20260729T044052+0000

Ratings and Alerts

No rating or validation information has been found for ExAc.

No alerts have been found for ExAc.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 4800 mentions in open access literature.

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

Giri R, et al. (2026) Pathogenic OTUD3 Mutations Predispose to Ulcerative Colitis Due to Barrier Dysfunction. Cellular and molecular gastroenterology and hepatology, 20(2), 101659.

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.

Guo W, et al. (2026) Pathogenicity of novel GLA gene missense mutations in Fabry disease and the therapeutic impact of migalastat. Journal of advanced research, 79, 549.

Miglierina E, et al. (2026) DIS3 licenses B cells for plasma cell differentiation in humans. Cellular & molecular immunology, 23(1), 31.

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.

Elias R, et al. (2025) Clear-Cell Renal Cell Carcinoma Molecular Subtypes Differ by African and European Genetic Similarity. Cancer research communications, 5(5), 743.

Zhang JT, et al. (2025) Follow-up Analysis Enhances Understanding of Molecular Residual Disease in Localized Non-Small Cell Lung Cancer. Clinical cancer research : an

official journal of the American Association for Cancer Research, 31(7), 1305.

van Ravensteijn SG, et al. (2025) Cemiplimab in Locally Advanced or Metastatic Secondary Angiosarcomas (CEMangio): A Phase II Clinical Trial and Biomarker Analyses. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(17), 3678.

Mäkitie 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.

Gong S, et al. (2025) Exon Sequencing of HNF1? in Chinese Patients with Early-Onset Diabetes. Diabetes & metabolism journal, 49(2), 321.

Zhao W, et al. (2025) GoFCards: an integrated database and analytic platform for gain of function variants in humans. Nucleic acids research, 53(D1), D976.

Bouzid A, et al. (2025) Whole exome sequencing identifies ABHD14A and MRNIP as novel candidate genes for developmental language disorder. Scientific reports, 15(1), 367.

Matsushita K, et al. (2025) Importance of EQA/PT for the detection of genetic variants in comprehensive cancer genome testing. Scientific reports, 15(1), 1036.

Zhu C, et al. (2025) The Oncologist, 2025, Vol, XX, Issue XX Predictive role of ARID1A and B2M mutations and the antigen presentation pathway in the efficacy of definitive chemoradiotherapy for cervical cancer. The oncologist, 30(6).

Ammar M, et al. (2025) Identification of a novel truncated pathogenic variant in PUS1 gene in two siblings of consanguineous Tunisian family: intrafamilial phenotypic variability related to mtDNA copy number. Annals of hematology, 104(2), 943.

Liu RL, et al. (2025) Whole exome sequence reveals genetic profiles of primary cardiomyopathy and genotype-phenotype association in Chinese population. BMC genomics, 26(1), 150.

Siwek JC, et al. (2025) Sliding Window Interaction Grammar (SWING): a generalized interaction language model for peptide and protein interactions. Nature methods, 22(8), 1707.

Zhang L, et al. (2025) Disitamab vedotin combined with toripalimab and radiotherapy for multimodal organ-sparing treatment of muscle invasive bladder cancer: a proof-of-concept study. Neoplasia (New York, N.Y.), 68, 101216.

Chen J, et al. (2025) Neonatal seizures associated with a rare familial 16p11.2 microduplication: A case report. The Journal of international medical research, 53(7), 3000605251358613.

Eid M, et al. (2025) Real-World Data From a Molecular Tumor Board-Assisted Cancer Care From a Single Center in The Czech Republic: Is Precision Oncology an Accessible Option, or a Privilege for a Minority of Patients? Cancer medicine, 14(15), e71119.