Resource Summary Report

Generated by NIF on Apr 9, 2025

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: database, data or information resource

Defining Citation: PMID:27899611

Keywords: exome sequencing, exome, sequencing, variant, grch37/hg19, gene, region,

transcript, multi-allelic variant, FASEB list

Funding:

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: ExAc

Resource ID: SCR_004068

Alternate IDs: nlx_158505

Record Creation Time: 20220129T080222+0000

Record Last Update: 20250409T060328+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 4620 mentions in open access literature.

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

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.

Liu Z, et al. (2025) Circulating tumor DNA analysis for prediction of prognosis and molecular insights in patients with resectable gastric cancer: results from a prospective study. MedComm, 6(2), e70065.

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.

Wang Z, et al. (2025) Optimizing the NGS-based discrimination of multiple lung cancers from the perspective of evolution. NPJ precision oncology, 9(1), 14.

Fan X, et al. (2025) Genotype-phenotype correlations for 17 Chinese families with inherited retinal dystrophies due to homozygous variants. Scientific reports, 15(1), 3043.

Dong J, et al. (2025) A Case Study Identified a New Mutation in the TTN Gene for Inherited Hypertrophic Cardiomyopathy. International journal of general medicine, 18, 447.

Ayenew B, et al. (2025) Pesticide Residues, Glyphosate Adsorption and Degradation Characteristics in Ethiopian Agricultural Soils. Environmental health insights, 19, 11786302241311679.

Zheng H, et al. (2025) Interpreting Variants of Uncertain Significance in PCD: Abnormal Splicing Caused by a Missense Variant of DNAAF3. Molecular genetics & genomic medicine, 13(1), e70036.

Lee D, et al. (2025) Increased local DNA methylation disorder in AMLs with DNMT3A-destabilizing variants and its clinical implication. Nature communications, 16(1), 560.

Cheng L, et al. (2025) Circulating Tumor DNA Detection for Recurrence Monitoring of Stage I Non-Small Cell Lung Cancer Treated With Microwave Ablation. Thoracic cancer, 16(2), e15534.

Liu Q, et al. (2025) RHOBTB2 Variant p.Arg511Gln Causes Developmental and Epileptic Encephalopathy Type 64 in an Infant: A Case Report and Hotspot Variant Analysis. Molecular genetics & genomic medicine, 13(1), e70059.

Cheng Y, et al. (2025) Stromal architecture and fibroblast subpopulations with opposing effects on outcomes in hepatocellular carcinoma. Cell discovery, 11(1), 1.

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.

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

Lutokhina Y, et al. (2025) Incidence and Impact of Myocarditis in Genetic Cardiomyopathies: Inflammation as a Potential Therapeutic Target. Genes, 16(1).

Schwarz JM, et al. (2025) Somatic DNA Variants in Epilepsy Surgery Brain Samples from Patients with Lesional Epilepsy. International journal of molecular sciences, 26(2).

Cui LM, et al. (2025) Analysis of a Series of 26 Cases With Prenatal Skeletal Dysplasia via Multiplatform Genetic Detection. Molecular genetics & genomic medicine, 13(1), e70062.

Krull JE, et al. (2024) Follicular lymphoma B cells exhibit heterogeneous transcriptional states with associated somatic alterations and tumor microenvironments. Cell reports.

Medicine, 5(3), 101443.

Anselmino N, et al. (2024) Integrative Molecular Analyses of the MD Anderson Prostate Cancer Patient-derived Xenograft (MDA PCa PDX) Series. Clinical cancer research : an official journal of the American Association for Cancer Research, 30(10), 2272.