Resource Summary Report

Generated by dkNET on Sep 5, 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: 20260904T000130+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 5119 mentions in open access literature.

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

Fuentes-Bolanos NA, et al. (2026) Integrated Germline and Somatic Molecular Profiling to Detect Cancer Predisposition Has a High Clinical Impact in Poor-Prognosis Pediatric Cancer. Clinical cancer research : an official journal of the American Association for Cancer Research, 32(10), 2047.

Chatterjee S, et al. (2026) Pathogenesis of Noonan Syndrome is Modulated by NOC2L, a Novel Interactor of LZTR1 Leading to Impaired P53 Signalling. The Journal of clinical endocrinology and metabolism, 111(5), 1418.

Uddin I, et al. (2026) Novel biallelic DNHD1 variants associated with male infertility with severe MMAF phenotype. Asian journal of andrology, 28(2), 179.

Zheng W, et al. (2026) Comprehensive clinical and genetic architecture of familial amyotrophic lateral sclerosis in China: A 15-year cohort study with 302 families. Neural regeneration research, 21(6), 2573.

Yu L, et al. (2026) Identification of a novel CLCN2 homozygous variant in a man with leukoencephalopathy and infertility: a case report and literature review. Frontiers in genetics, 17, 1761205.

Akkus N, et al. (2026) Whole Exome Sequencing in Patients With Developmental Delay/Intellectual Disability (DD/ID), Epilepsy and the First Turkish Patient Diagnosed With

BCL11A-Related Intellectual Disability. Molecular genetics & genomic medicine, 14(1), e70180.

Ishigami D, et al. (2026) Delineating the Genetic Basis of RNF213-related vasculopathies: The association of PKHD1 variants with bilateral cerebral vasculopathy. European journal of human genetics : EJHG, 34(6), 788.

Wang Y, et al. (2026) Identification and validation of CARS1 p.E712V and NF1 p.Q2002X in sporadic Moyamoya disease across 30 trio pedigrees. NPJ genomic medicine, 11(1), 9.

Han C, et al. (2026) Whole-exome sequencing for the genetic diagnosis of early-onset high myopia and associated hereditary eye disorders. BMC medical genomics, 19(1).

Bas I, et al. (2026) Unclassified Inborn Errors of Immunity Patients without any Pathogenic Variant in Targeted Next-Generation Sequencing: Long-Term Follow-up and Whole Exome Sequencing Results. Journal of clinical immunology, 46(1).

Zennami K, et al. (2026) Genomic Profiling in Localized Prostate Cancer: Associations With Biochemical Recurrence and Response to Salvage Radiotherapy. Cancer science, 117(5), 1469.

Hong F, et al. (2026) Pulmonary sequestration with a rare renal artery subtype: a comprehensive clinicopathological analysis of 20 pediatric cases. BMC pulmonary medicine, 26(1).

Rajeh A, et al. (2026) Somatic mutations reveal hyperactive Notch signaling in prurigo nodularis. JCI insight, 11(8).

Liao R, et al. (2026) Integrative multi-omics profiling reveals distinct evolutionary and immunogenic features of brain oligometastasis in lung adenocarcinoma. Genome medicine, 18(1).

Wu Y, et al. (2026) Identification of De Novo ZBTB18 Variant in a Patient With Global Developmental Delay, Seizures, and Juvenile Facies. International journal of pediatrics, 2026, 7226289.

Xu H, et al. (2026) Comprehensive genomic profiling of neuroendocrine neoplasms of the colorectum. Frontiers in genetics, 17, 1792341.

Zhang W, et al. (2026) Epilepsy associated with SYNGAP1 gene variants: clinical features of six cases and a literature review. Frontiers in pediatrics, 14, 1789561.

Scholl S, et al. (2026) Unveiling Candidate Markers for Drug Resistance or Synthetic Lethality in Cervical Cancer: Integrative Analysis of Genetic and Pharmacoprofiling. Cancer reports (Hoboken, N.J.), 9(6), e70599.

Treccarichi S, et al. (2026) A de novo Loss-of-function Variant in RAPGEF6 Supports its Role in Neuropsychiatric Disorders. Journal of molecular neuroscience : MN, 76(2).

Gao J, et al. (2026) Genetic heterogeneity correlated with phenotypic variability in 6 Chinese families with Alport syndrome. Frontiers in genetics, 17, 1840344.