Resource Summary Report

Generated by NIF on Aug 23, 2026

ExomeDepth

RRID:SCR_002663
Type: Tool

Proper Citation

ExomeDepth (RRID:SCR_002663)

Resource Information

URL: http://cran.r-project.org/web/packages/ExomeDepth/

Proper Citation: ExomeDepth (RRID:SCR_002663)

Description: Software that calls copy number variants (CNVs) from targeted sequence data, typically exome sequencing experiments designed to identify the genetic basis of Mendelian disorders.

Resource Type: software resource

Defining Citation: PMID:22942019

Keywords: software package, unix/linux, mac os x, windows, r, bio.tools

Availability: Free, Available for download, Freely available

Resource Name: ExomeDepth

Resource ID: SCR_002663

Alternate IDs: OMICS_05443, biotools:exomedepth

Alternate URLs: https://bio.tools/exomedepth

License: GNU General Public License, v3

Record Creation Time: 20220129T080214+0000

Record Last Update: 20260815T182218+0000

Ratings and Alerts

No rating or validation information has been found for ExomeDepth.

No alerts have been found for ExomeDepth.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 295 mentions in open access literature.

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

Borges RL, et al. (2026) Unbalanced chromatin binding of Polycomb complexes drives neurodevelopmental disorders. Molecular cell, 86(4), 604.

Lezirovitz K, et al. (2026) A Structural Variant in the 5' Regulatory Region of DIAPH3 Segregates with Postlingual Hearing Loss and Auditory Neuropathy in a Multigenerational Brazilian Family. International archives of otorhinolaryngology, 30(2), 1.

Wang L, et al. (2026) Genetic analysis of fetal skeletal dysplasia via whole exome sequencing and non-invasive prenatal diagnosis. Annals of medicine, 58(1), 2606517.

Nie H, et al. (2026) Identification and functional characterization of a novel pathogenic COL1A1 splicing variant in a Chinese family with osteogenesis imperfecta. Frontiers in genetics, 17, 1758799.

Quaynor SK, et al. (2026) Consensus-based Detection of Aetiologic Copy Number Variants For Syndromic Orofacial Clefts Utilising Whole Exome Sequencing of Case Parent Trios. Research square.

Marsal-Olivan A, et al. (2026) Integrating preconception carrier screening into public health: lessons learned from a pilot implementation study. Journal of assisted reproduction and genetics, 43(6), 1711.

Lim S, et al. (2026) Development and Performance Validation of a Comprehensive Liquid Biopsy Genotyping Panel for Pan-cancer Analysis. Annals of laboratory medicine, 46(2), 210.

Zhang Q, et al. (2026) Application of chromosomal microarray analysis and trio whole-exome sequencing in first-trimester prenatal diagnosis for high-risk pregnancies. BMC pregnancy and childbirth, 26(1).

Yousaf H, et al. (2026) Expanding the repertoire of loss-of-function variants in HACE1 causing complex spastic paraplegia: literature review and recommendations on clinical management. Human genomics, 20(1).

Zhang Q, et al. (2026) Recurrent migraine with visual aura as the primary phenotype of familial neuronal intranuclear inclusion disease. Frontiers in neurology, 17, 1747202.

Deng X, et al. (2026) The genotypic spectrum of complex febrile seizures: insights from high-risk population genetic screening in a pediatric cohort. Frontiers in neuroscience, 20,

1828448.

Nabbout F, et al. (2026) Familial Tooth Agenesis in Lebanese Patients: Clinical Characterization and Whole-Exome Identification of Rare CACNA2D2 and TRIO Variants. Cureus, 18(5), e108369.

Chang L, et al. (2026) RP9 revisited; RP9 p.(H137L) remains a likely cause of dominant splicing factor-Retinitis Pigmentosa. European journal of human genetics : EJHG, 34(2), 227.

Diogo-Cavassana S, et al. (2026) Genetic Landscape of Hearing Loss in Brazilian Patients Reveals Population-Specific Variants and Clinical Correlations. Clinical genetics, 110(2), 210.

Pérez-Béliz E, et al. (2026) Adrenal mixed corticomedullary tumors: report of a case with molecular characterization and systematic review. Virchows Archiv : an international journal of pathology, 488(2), 367.

Tosi M, et al. (2026) A multi-omics study on monozygotic twins discordant for amyotrophic lateral sclerosis and literature review underline a potential role for innate immunity and epigenetic dysregulation in disease mechanisms. Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 47(3), 230.

Zenteno JC, et al. (2026) The landscape of 605 genetically confirmed distinct rare diseases in a single center in Mexico (2005-2025). Orphanet journal of rare diseases, 21(1).

Wang L, et al. (2026) Analysis of copy number variations and candidate genes in recurrent pregnancy loss. PeerJ, 14, e20889.

Lee H, et al. (2026) Targeted Sequencing of Human Aorta Tissue Reveals Undiagnosed Heritable Thoracic Aortic Disease. Interdisciplinary cardiovascular and thoracic surgery, 41(5).

Price-Kuehne F, et al. (2026) Expanding the Spectrum of Src-Family Kinase-Related Autoinflammatory Diseases: Monogenic Vasculitis Caused By Germline Pathogenic Variants in HCK and FGR. Journal of clinical immunology, 46(1).