

# Resource Summary Report

Generated by [SPARC SAWG](#) on Sep 17, 2026

## ExomeDepth

RRID:SCR\_002663

Type: Tool

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### Proper Citation

ExomeDepth (RRID:SCR\_002663)

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### 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:** 20260912T195544+0000

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### Ratings and Alerts

No rating or validation information has been found for ExomeDepth.

No alerts have been found for ExomeDepth.

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## Data and Source Information

**Source:** [SciCrunch Registry](#)

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## Usage and Citation Metrics

We found 295 mentions in open access literature.

**Listed below are recent publications.** The full list is available at [SPARC SAWG](#) .

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).