

Resource Summary Report

Generated by [RRID](#) on Aug 6, 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: 20260801T190152+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 262 mentions in open access literature.

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

Jiang L, et al. (2025) Identification of a novel microdeletion at 9q21.13 in a family with epilepsy, intellectual disability, and speech disorders and literature review. *Frontiers in genetics*, 16, 1616005.

Llansó L, et al. (2025) Repeat Expansions in PLIN4 Cause Autosomal Dominant Vacuolar Myopathy With Sarcolemmal Features. *Annals of clinical and translational neurology*, 12(10), 2136.

Vooren EV, et al. (2025) RPE65 variant p.(E519K) causes a novel dominant adult-onset maculopathy in 83 affected individuals. *Research square*.

Hammann N, et al. (2025) Hepatic Form of Dihydrolipoamide Dehydrogenase Deficiency (DLDD): Phenotypic Spectrum, Laboratory Findings, and Therapeutic Approaches in 52 Patients. *Journal of inherited metabolic disease*, 48(3), e70035.

Sheth J, et al. (2025) Prenatal diagnosis of rare genetic disorders: fourteen years' experience of a tertiary genetic centre from India. *Orphanet journal of rare diseases*, 20(1), 471.

Chen Q, et al. (2025) A novel frameshift variation of PKD1 in familial autosomal dominant polycystic kidney diseases: expanding the clinical phenotype and genetic spectrum of PKD1 disorders. *BMC medical genomics*, 18(1), 170.

Stangner K, et al. (2025) Apremilast improves cardiomyocyte cohesion and arrhythmia in different models for arrhythmogenic cardiomyopathy. *Stem cell research & therapy*, 16(1), 609.

Tejero E, et al. (2025) Ending diagnostic odyssey by reanalysis of whole exome sequencing data: reclassification of suspected Fanconi anemia cases to dyskeratosis congenita and Diamond-Blackfan anemia. *Orphanet journal of rare diseases*, 20(1), 511.

Boujemaa M, et al. (2025) Exome sequencing reveals new insights into the germline landscape of inflammatory breast cancer among Tunisian patients. *Journal of translational medicine*, 23(1), 1089.

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.

D'Abrusco F, et al. (2025) Pathogenic cryptic variants detectable through exome data reanalysis significantly increase the diagnostic yield in Joubert syndrome. *European journal of human genetics : EJHG*, 33(1), 72.

Cheng C, et al. (2025) Identifying novel heterozygous PI4KA variants in fetal abnormalities. *BMC medical genomics*, 18(1), 23.

Mirinezhad MR, et al. (2025) Reporting a Homozygous Case of Neurodevelopmental Disorder Associated With a Novel PRPF8 Variant. *Molecular genetics & genomic medicine*, 13(3), e70084.

Bregant E, et al. (2025) The molecular landscape of hereditary ataxia: a single-center study. *Human genetics*, 144(5), 545.

He D, et al. (2025) A novel splice-altering frameshift variant in the COL1A1 gene underlies osteogenesis imperfecta type I: molecular characterization of a four-generation Chinese pedigree and literature review. *Human genomics*, 19(1), 103.

Banning A, et al. (2025) Structural and Functional Characterization of N-Glycanase-1 Pathogenic Variants. *Cells*, 14(13).

Aguilar-Soto M, et al. (2025) Association of pituitary neuroendocrine tumors and neurofibromatosis type 1: assessing causation versus coincidence. Case report. *Frontiers in endocrinology*, 16, 1483305.

Li M, et al. (2025) Identification and functional analysis of a novel SMARCC2 splicing variant in a family with syndromic neurodevelopmental disorder. *Orphanet journal of rare diseases*, 20(1), 48.

Liu X, et al. (2025) A novel TGFB2 mutation causes Loeys-Dietz syndrome in a Chinese infant: A case report. *Heliyon*, 11(2), e42116.

Ridd S, et al. (2025) Myelodysplastic syndrome diagnosed by genetic testing for hereditary cancer: a case report. *NPJ genomic medicine*, 10(1), 39.