Resource Summary Report

Generated by ASWG on Aug 2, 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 ASWG .

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.

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.

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.

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

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.

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.

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.

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

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

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

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.

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 TGFBR2 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.