Resource Summary Report

Generated by dkNET on Apr 25, 2025

exomeCopy

RRID:SCR_001276
Type: Tool

Proper Citation

exomeCopy (RRID:SCR_001276)

Resource Information

URL: http://www.bioconductor.org/packages/release/bioc/html/exomeCopy.html

Proper Citation: exomeCopy (RRID:SCR_001276)

Description: Software package for detection of copy number variants (CNV) from exome sequencing samples, including unpaired samples. The package implements a hidden Markov model which uses positional covariates, such as background read depth and GC-content, to simultaneously normalize and segment the samples into regions of constant copy count.

Abbreviations: exomeCopy

Synonyms: exomeCopy - Copy number variant detection from exome sequencing read depth

Resource Type: software resource

Defining Citation: PMID:23089826

Keywords: copy number variation, genetics, sequencing, exome

Funding:

Availability: GNU General Public License, v2 or newer

Resource Name: exomeCopy

Resource ID: SCR_001276

Alternate IDs: OMICS_02062

Record Creation Time: 20220129T080206+0000

Record Last Update: 20250420T014025+0000

Ratings and Alerts

No rating or validation information has been found for exomeCopy.

No alerts have been found for exomeCopy.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 15 mentions in open access literature.

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

Broghammer F, et al. (2024) Resistance of HNSCC cell models to pan-FGFR inhibition depends on the EMT phenotype associating with clinical outcome. Molecular cancer, 23(1), 39.

Mamontova V, et al. (2024) NEAT1 promotes genome stability via m6A methylation-dependent regulation of CHD4. Genes & development, 38(17-20), 915.

Trifault B, et al. (2024) Nucleolar detention of NONO shields DNA double-strand breaks from aberrant transcripts. Nucleic acids research, 52(6), 3050.

Gaballa A, et al. (2024) PAF1c links S-phase progression to immune evasion and MYC function in pancreatic carcinoma. Nature communications, 15(1), 1446.

Redfield SE, et al. (2024) PKHD1L1, a gene involved in the stereocilia coat, causes autosomal recessive nonsyndromic hearing loss. Human genetics, 143(3), 311.

Sen A, et al. (2022) Leveraging Allele-Specific Expression for Therapeutic Response Gene Discovery in Glioblastoma. Cancer research, 82(3), 377.

Qi X, et al. (2021) Uncovering potential single nucleotide polymorphisms, copy number variations and related signaling pathways in primary Sjogren's syndrome. Bioengineered, 12(2), 9313.

Mansilla MA, et al. (2021) Targeted broad-based genetic testing by next-generation

sequencing informs diagnosis and facilitates management in patients with kidney diseases. Nephrology, dialysis, transplantation : official publication of the European Dialysis and Transplant Association - European Renal Association, 36(2), 295.

Zhao L, et al. (2020) Comparative study of whole exome sequencing-based copy number variation detection tools. BMC bioinformatics, 21(1), 97.

Ku?mirek W, et al. (2019) Comparison of kNN and k-means optimization methods of reference set selection for improved CNV callers performance. BMC bioinformatics, 20(1), 266.

Tessoulin B, et al. (2018) Whole-exon sequencing of human myeloma cell lines shows mutations related to myeloma patients at relapse with major hits in the DNA regulation and repair pathways. Journal of hematology & oncology, 11(1), 137.

Povysil G, et al. (2017) panelcn.MOPS: Copy-number detection in targeted NGS panel data for clinical diagnostics. Human mutation, 38(7), 889.

Vesely C, et al. (2017) Genomic and transcriptional landscape of P2RY8-CRLF2-positive childhood acute lymphoblastic leukemia. Leukemia, 31(7), 1491.

Samarakoon PS, et al. (2016) cnvScan: a CNV screening and annotation tool to improve the clinical utility of computational CNV prediction from exome sequencing data. BMC genomics, 17, 51.

Kalscheuer VM, et al. (2015) Novel Missense Mutation A789V in IQSEC2 Underlies X-Linked Intellectual Disability in the MRX78 Family. Frontiers in molecular neuroscience, 8, 85.