Resource Summary Report

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PennCNV

RRID:SCR_002518
Type: Tool

Proper Citation

PennCNV (RRID:SCR_002518)

Resource Information

URL: http://www.nitrc.org/projects/penncnv

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Description: A free software tool for Copy Number Variation (CNV) detection from SNP genotyping arrays. Currently it can handle signal intensity data from Illumina and Affymetrix arrays. With appropriate preparation of file format, it can also handle other types of SNP arrays and oligonucleotide arrays. PennCNV implements a hidden Markov model (HMM) that integrates multiple sources of information to infer CNV calls for individual genotyped samples. It differs form segmentation-based algorithm in that it considered SNP allelic ratio distribution as well as other factors, in addition to signal intensity alone. In addition, PennCNV can optionally utilize family information to generate family-based CNV calls by several different algorithms. Furthermore, PennCNV can generate CNV calls given a specific set of candidate CNV regions, through a validation-calling algorithm.

Abbreviations: PennCNV

Synonyms: PennCNV: copy number variation detection

Resource Type: software resource

Defining Citation: PMID:17921354

Keywords: imaging genomics, copy number variation, snp, genotyping array, array, oligonucleotide, hidden markov model, genotype, genome

Funding: NIMH MH604687

Availability: Free

Resource Name: PennCNV

Resource ID: SCR_002518

Alternate IDs: OMICS_00729, nlx_155921

Alternate URLs: http://www.openbioinformatics.org/penncnv/

Old URLs: http://www.neurogenome.org/cnv/penncnv

License: Public domain

Record Creation Time: 20220129T080213+0000

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Ratings and Alerts

No rating or validation information has been found for PennCNV.

No alerts have been found for PennCNV.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 372 mentions in open access literature.

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

Wong H, et al. (2026) An Analytic Framework Characterizes the Biological Processes That Shape Copy Number-Based Genome Instability Patterns in Breast Cancer. Cancer research, 86(13), 3344.

de Oliveira LF, et al. (2026) Genome-Wide Detection of Copy Number Variation and Association Studies With Physiological and Anatomical Indicators of Heat Stress Response in Lactating Sows. Journal of animal breeding and genetics = Zeitschrift fur Tierzuchtung und Zuchtungsbiologie, 143(1), 183.

Ivankovic F, et al. (2026) MarkerMatch: a proximity-based probe-matching algorithm for joint analysis of copy-number variants from different genotyping arrays. Bioinformatics (Oxford, England), 42(6).

Montalbano S, et al. (2026) CNValidatron: accurate and efficient validation of PennCNV calls using computer vision. BMC bioinformatics, 27(1), 47.

Lodi MK, et al. (2026) Narrow Versus Broad Phenotype Definitions Affect Genetic Analysis of Language More than Other Broad Autism Phenotype Traits. Genes, 17(2).

Wakil SM, et al. (2026) Recurrent and Non-Recurrent Copy Number Variants in Native Americans and a Cosmopolitan Sample in Relation to Alcohol Use Disorder and Other Psychiatric Diseases. Molecular neurobiology, 63(1).

Ko SH, et al. (2026) ???The inverse relationship between copy number variation burden and age of onset of metabolic syndrome in Korean middle-aged adults. BMC medical genomics, 19(1).

Sacks MF, et al. (2026) Combinatorial effects of gene dosage, polygenic background and environment on complex traits. medRxiv : the preprint server for health sciences.

Tiwari HK, et al. (2026) Exploration of Regulatory Elements, MicroRNAs, and Copy Number Variation in Urogenital Chlamydia Reinfection in African American Women. International journal of molecular sciences, 27(12).

Liu F, et al. (2026) Integration of GWAS and eQTL analysis reveals regulatory CNVs for fatty acid traits in cattle. iScience, 29(6), 116180.

Dennison CA, et al. (2026) Childhood ADHD and autism spectrum disorder difficulties: exploring the impact of copy number variants on young adult outcomes. BJPsych open, 12(3), e108.

Harripaul R, et al. (2026) Autism spectrum disorder trios from consanguineous populations are enriched for rare homozygous variants, identifying 32 new candidate genes. Scientific reports, 16(1).

Landoulsi Z, et al. (2026) Genome-wide association study of copy number variations in Parkinson's disease. NPJ Parkinson's disease, 12(1).

Lee J, et al. (2026) Structural variants in human congenital heart disease disrupt distal genomic regulatory contacts of developmental genes. bioRxiv : the preprint server for biology.

Prapiadou S, et al. (2026) Copy Number Variants and Their Association With Intracerebral Hemorrhage Risk: A Case-Control Study. Annals of clinical and translational neurology, 13(3), 530.

Dong W, et al. (2026) Age at type 2 diabetes onset and risk of dementia: The modifying role of genetic susceptibility and mitochondrial function. The journal of nutrition, health & aging, 30(3), 100780.

Vaez M, et al. (2026) Recurrent Copy Number Variants and Psychiatric Outcomes in the Context of Polygenic Scores. JAMA psychiatry.

Krebs K, et al. (2026) Pharmacokinetic recall study of Estonian Biobank participants with novel genetic variants in CYP2C19 and CYP2D6. NPJ genomic medicine, 11(1), 10.

Snihirova Y, et al. (2026) The Association of Environmental Variables with Intelligence and Well-Being in Copy Number Variation (CNV) Carriers. Research square.

Hall JC, et al. (2026) Patient induced pluripotent stem cells identify specificities of a reticular pseudodrusen phenotype in age-related macular degeneration. Genome medicine, 18(1).