Resource Summary Report

Generated by T1D on Aug 5, 2026

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

Record Last Update: 20260801T190211+0000

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 349 mentions in open access literature.

Listed below are recent publications. The full list is available at T1D .

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.

Kariis HM, et al. (2025) Genetic influences on antidepressant side effects: a CYP2C19 gene variation and polygenic risk study in the Estonian Biobank. European journal of human genetics : EJHG, 33(10), 1376.

Fernández-Suárez E, et al. (2025) New genetic diagnoses for inherited retinal dystrophies by integrating splicing tools into NGS pipelines. NPJ genomic medicine, 10(1), 52.

Xing J, et al. (2025) Narrow Versus Broad Phenotype Definitions Affect Genetic Analysis of Language More Than Other Broad Autism Phenotype Traits. Research square.

Montalbano S, et al. (2025) CNValidatron: Accurate And Efficient Validation of PennCNV Calls Using Computer Vision. bioRxiv : the preprint server for biology.

Sharma NK, et al. (2025) Genome wide landscaping of copy number variations for horse inter-breed variability. Animal biotechnology, 36(1), 2446251.

Xiong Y, et al. (2025) Genome-wide association meta-analysis and rare copy number variant analysis of treatment-resistant depression. Molecular psychiatry, 30(11), 5024.

Ivankovic F, et al. (2025) MarkerMatch: A Proximity-Based Probe-Matching Algorithm for Joint Analysis of Copy-Number Variants from Different Genotyping Arrays. bioRxiv : the preprint server for biology.

Li D, et al. (2025) Modeling the long-range effect of an inversion downstream of EFNB1 concludes a 43-year molecular diagnostic odyssey for craniofrontonasal syndrome. European journal of human genetics : EJHG, 33(12), 1684.

Rammos A, et al. (2025) Copy number variants and their implications for developmental and behavioural problems in cleft lip and/or palate. Human molecular genetics, 34(18), 1563.

Gijsbertsen M, et al. (2025) Generation of human induced pluripotent stem cell lines from patients with FGFR2-linked syndromic craniosynostosis. Disease models & mechanisms, 18(10).

Liao Z, et al. (2025) Copy number variants and the tangential expansion of the cerebral cortex. Nature communications, 16(1), 1697.

Flanagan SE, et al. (2025) Large copy number variants are an important cause of congenital hyperinsulinism that should be screened for during routine testing. Frontiers in endocrinology, 16, 1514916.

Landoulsi Z, et al. (2025) Large-scale copy number variant analysis in genes linked to Parkinson´s disease. NPJ Parkinson's disease, 11(1), 225.

Milo Rasouly H, et al. (2025) Exome analysis links kidney malformations to developmental disorders and reveals causal genes. Nature communications, 16(1), 7290.

Engchuan W, et al. (2025) Psychiatric disorders converge on common pathways but diverge in cellular context, spatial distribution, and directionality of genetic effects. medRxiv : the preprint server for health sciences.

Cruz T, et al. (2025) Clinical, laboratory and genetic factors associated with smoking in a Brazilian Sickle Cell Disease (SCD) cohort. PloS one, 20(9), e0332305.

Halvorsen M, et al. (2025) Persistent Tic Disorders Are Associated With 17q12 Duplications. Research square.

Poulain C, et al. (2025) The interplay between genomic copy number variants, sleep, and cognition in the general population. Translational psychiatry, 15(1), 351.

Walker A, et al. (2025) Genome-wide copy number variation association study in anorexia nervosa. Molecular psychiatry, 30(5), 2009.