# Resource Summary Report

Generated by NIF on May 23, 2025

## 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:** Public domain

**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

**Record Creation Time:** 20220129T080213+0000

**Record Last Update:** 20250519T203211+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 322 mentions in open access literature.

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

Littleton SH, et al. (2024) Variant-to-function analysis of the childhood obesity chr12q13 locus implicates rs7132908 as a causal variant within the 3' UTR of FAIM2. Cell genomics, 4(5), 100556.

Auwerx C, et al. (2024) Rare copy-number variants as modulators of common disease susceptibility. Genome medicine, 16(1), 5.

Halvorsen M, et al. (2024) A Burden of Rare Copy Number Variants in Obsessive-Compulsive Disorder. Research square.

Warren TL, et al. (2024) Association of neurotransmitter pathway polygenic risk with specific symptom profiles in psychosis. Molecular psychiatry, 29(8), 2389.

Sha Z, et al. (2024) The copy number variant architecture of psychopathology and cognitive development in the ABCD® study. medRxiv : the preprint server for health sciences.

Benfica LF, et al. (2024) Genome-wide association study between copy number variation and feeding behavior, feed efficiency, and growth traits in Nellore cattle. BMC genomics, 25(1), 54.

Ceroni F, et al. (2024) Deletion upstream of MAB21L2 highlights the importance of evolutionarily conserved non-coding sequences for eye development. Nature communications, 15(1), 9245.

Jensen M, et al. (2024) Genetic modifiers and ascertainment drive variable expressivity of complex disorders. medRxiv : the preprint server for health sciences.

Landoulsi Z, et al. (2024) Genome-wide association study of copy number variations in Parkinson's disease. medRxiv : the preprint server for health sciences.

Nyaga DM, et al. (2024) Leveraging multiple approaches for the detection of pathogenic deep intronic variants in developmental and epileptic encephalopathies: A case report. Epilepsia open, 9(2), 758.

Dinneen TJ, et al. (2024) Polygenic scores stratify neurodevelopmental copy number variant carrier cognitive outcomes in the UK Biobank. NPJ genomic medicine, 9(1), 43.

Gan P, et al. (2024) Development and validation of a pharmacogenomics reporting workflow based on the illumina global screening array chip. Frontiers in pharmacology, 15, 1349203.

Vaez M, et al. (2024) Population-Based Risk of Psychiatric Disorders Associated With Recurrent Copy Number Variants. JAMA psychiatry, 81(10), 957.

Subramanian K, et al. (2024) Landscape of genomic structural variations in Indian population-based cohorts: Deeper insights into their prevalence and clinical relevance. HGG advances, 5(3), 100285.

Huguet G, et al. (2024) Effects of gene dosage on cognitive ability: A function-based association study across brain and non-brain processes. Cell genomics, 4(12), 100721.

Viggiano M, et al. (2024) Genomic analysis of 116 autism families strengthens known risk genes and highlights promising candidates. NPJ genomic medicine, 9(1), 21.

Artaza H, et al. (2024) Rare copy number variation in autoimmune Addison's disease. Frontiers in immunology, 15, 1374499.

Sun Y, et al. (2024) Contribution of copy number variants on antipsychotic treatment response in Han Chinese patients with schizophrenia. EBioMedicine, 105, 105195.

Gao Z, et al. (2024) Copy number deletion of PLA2G4A affects the susceptibility and clinical phenotypes of schizophrenia. Schizophrenia (Heidelberg, Germany), 10(1), 55.

Littleton SH, et al. (2023) Variant-to-function analysis of the childhood obesity chr12q13 locus implicates rs7132908 as a causal variant within the 3' UTR of FAIM2. bioRxiv : the preprint server for biology.