# Resource Summary Report

Generated by FDI Lab - SciCrunch.org on Apr 21, 2025

## FishingCNV

RRID:SCR_013038
Type: Tool

### Proper Citation

FishingCNV (RRID:SCR_013038)

### Resource Information

**URL:** http://sourceforge.net/projects/fishingcnv/

**Proper Citation:** FishingCNV (RRID:SCR_013038)

**Description:** A software tool developed at McGill University, is a tool for comprehensive analysis of rare copy number variations in high-throughput exome sequencing data.

**Abbreviations:** FishingCNV

**Synonyms:** FishingCNV - Copy number variation detection in exome sequencing data, FishingCNV - CNV detection in exome sequencing data, FishingCNV - Copy number variation (CNV) detection in exome sequencing data

**Resource Type:** software resource

**Defining Citation:** PMID:23539306

**Funding:**

**Availability:** Commercial license

**Resource Name:** FishingCNV

**Resource ID:** SCR_013038

**Alternate IDs:** OMICS_00334

**Record Creation Time:** 20220129T080313+0000

**Record Last Update:** 20250420T014630+0000

## Ratings and Alerts

No rating or validation information has been found for FishingCNV.

No alerts have been found for FishingCNV.

## Data and Source Information

**Source:** SciCrunch Registry

## Usage and Citation Metrics

We found 11 mentions in open access literature.

**Listed below are recent publications.** The full list is available at FDI Lab - SciCrunch.org.


Aamir A, et al. (2021) Discordant phenotypes in twins with infantile nystagmus. Scientific reports, 11(1), 2826.

Kim YJ, et al. (2021) Identification of a Homozygous PEX26 Mutation in a Heimler Syndrome Patient. Genes, 12(5).

Gordeeva V, et al. (2021) Benchmarking germline CNV calling tools from exome sequencing data. Scientific reports, 11(1), 14416.

De Vilder EYG, et al. (2021) Rare Modifier Variants Alter the Severity of Cardiovascular Disease in Pseudoxanthoma Elasticum: Identification of Novel Candidate Modifier Genes and Disease Pathways Through Mixture of Effects Analysis. Frontiers in cell and developmental biology, 9, 612581.

Sadler B, et al. (2020) Rare and de novo duplications containing SHOX in clubfoot. Journal of medical genetics, 57(12), 851.

Salloum R, et al. (2017) Characterizing temporal genomic heterogeneity in pediatric high-grade gliomas. Acta neuropathologica communications, 5(1), 78.

Thomas MG, et al. (2017) Development and clinical utility of a novel diagnostic nystagmus gene panel using targeted next-generation sequencing. European journal of human genetics : EJHG, 25(6), 725.

Watson CM, et al. (2016) Enhanced diagnostic yield in Meckel-Gruber and Joubert syndrome through exome sequencing supplemented with split-read mapping. BMC medical genetics, 17, 1.

Nikbakht H, et al. (2016) Spatial and temporal homogeneity of driver mutations in diffuse intrinsic pontine glioma. Nature communications, 7, 11185.

Poulter JA, et al. (2015) A distinctive oral phenotype points to FAM20A mutations not identified by Sanger sequencing. Molecular genetics & genomic medicine, 3(6), 543.

Barclay SF, et al. (2015) Rapid-Onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD): exome sequencing of trios, monozygotic twins and tumours. Orphanet journal of rare diseases, 10, 103.