

Resource Summary Report

Generated by SPARC SAWG on Jul 28, 2026

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](#)

Availability: Commercial license

Resource Name: FishingCNV

Resource ID: SCR_013038

Alternate IDs: OMICS_00334

Record Creation Time: 20220129T080313+0000

Record Last Update: 20260725T190744+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 [SPARC SAWG](#) .

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

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

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.

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.

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.