

Resource Summary Report

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HOMOZYGOSITYMAPPER

RRID:SCR_001714

Type: Tool

Proper Citation

HOMOZYGOSITYMAPPER (RRID:SCR_001714)

Resource Information

URL: <http://www.homozygositymapper.org/>

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Description: A web-based approach of homozygosity mapping that can handle tens of thousands markers. User can upload their own SNP genotype files to the database. Intuitive graphic interface is provided to view the homozygous stretches, with the ability of zooming into single chromosomes or user-defined chromosome regions. The underlying genotypes in all samples are displayed. The software is also integrated with our candidate gene search engine, GeneDistiller, so that users can interactively determine the most promising gene. (entry from Genetic Analysis Software)

Abbreviations: HomozygosityMapper

Resource Type: data analysis service, production service resource, service resource, analysis service resource

Defining Citation: [PMID:19465395](#)

Keywords: gene, genetic, genomic, perl, genotype, homozygosity score, homozygosity, bio.tools, FASEB list

Availability: Free, Freely Available

Resource Name: HOMOZYGOSITYMAPPER

Resource ID: SCR_001714

Alternate IDs: nlx_154069, biotools:homozygositymapper, OMICS_00123

Alternate URLs: <https://bio.tools/homozygositymapper>

Record Creation Time: 20220129T080209+0000

Record Last Update: 20260806T054326+0000

Ratings and Alerts

No rating or validation information has been found for HOMOZYGOSITYMAPPER.

No alerts have been found for HOMOZYGOSITYMAPPER.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 121 mentions in open access literature.

Listed below are recent publications. The full list is available at [NIF](#) .

Sudhindar PD, et al. (2025) Urinary renal epithelial cells can be used for NPHP1 phenotyping and a personalized therapeutic strategy. *Journal of cell science*, 138(20).

Jean MM, et al. (2025) A ciliopathy combining Joubert syndrome and Oro-Facial-Digital syndrome caused by bi-allelic 5'-UTR loss-of-function CEP83 variant. *NPJ genomic medicine*, 10(1), 66.

Veerappa A, et al. (2024) Coordination among frequent genetic variants imparts substance use susceptibility and pathogenesis. *Frontiers in neuroscience*, 18, 1332419.

Khan H, et al. (2024) Biallelic variants identified in 36 Pakistani families and trios with autism spectrum disorder. *Scientific reports*, 14(1), 9230.

Khan J, et al. (2024) Mutational spectrum associated with oculocutaneous albinism and Hermansky-Pudlak syndrome in nine Pakistani families. *BMC ophthalmology*, 24(1), 345.

Nuzhat N, et al. (2023) CEP162 deficiency causes human retinal degeneration and reveals a dual role in ciliogenesis and neurogenesis. *The Journal of clinical investigation*, 133(8).

Sanga S, et al. (2023) Identification of a shared, common haplotype segregating with an SGCB c.544 T > G mutation in Indian patients affected with sarcoglycanopathy. *Scientific reports*, 13(1), 15095.

Aharoni S, et al. (2022) PSMC1 variant causes a novel neurological syndrome. *Clinical genetics*, 102(4), 324.

Halperin D, et al. (2022) A syndrome of severe intellectual disability, hypotonia, failure to thrive, dysmorphism, and thinning of corpus callosum maps to chromosome 7q21.13-q21.3. *Clinical genetics*, 102(2), 123.

Borg R, et al. (2021) Genetic analysis of ALS cases in the isolated island population of Malta. *European journal of human genetics : EJHG*, 29(4), 604.

Monfrini E, et al. (2021) A Novel Homozygous VPS11 Variant May Cause Generalized Dystonia. *Annals of neurology*, 89(4), 834.

Morgan NV, et al. (2021) Evidence that autosomal recessive spastic cerebral palsy-1 (CPSQ1) is caused by a missense variant in HPDL. *Brain communications*, 3(1), fcab002.

Kaiyrzhanov R, et al. (2021) A Novel Homozygous ADCY5 Variant is Associated with a Neurodevelopmental Disorder and Movement Abnormalities. *Movement disorders clinical practice*, 8(7), 1140.

Halperin D, et al. (2021) CDH2 mutation affecting N-cadherin function causes attention-deficit hyperactivity disorder in humans and mice. *Nature communications*, 12(1), 6187.

Al Alawi I, et al. (2021) The diagnostic yield of whole exome sequencing as a first approach in consanguineous Omani renal ciliopathy syndrome patients. *F1000Research*, 10, 207.

Vona B, et al. (2021) A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. *Human genetics*, 140(6), 915.

Guo L, et al. (2021) Deficiency of TMEM53 causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. *Nature communications*, 12(1), 2046.

Pastore SF, et al. (2021) Biallelic inheritance in a single Pakistani family with intellectual disability implicates new candidate gene RDH14. *Scientific reports*, 11(1), 23113.

Rasheed M, et al. (2021) Exome sequencing identifies novel and known mutations in families with intellectual disability. *BMC medical genomics*, 14(1), 211.

Pagnamenta AT, et al. (2021) An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. *Brain : a journal of neurology*, 144(2), 584.