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: analysis service resource, data analysis service, production service resource, 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: 20260905T133114+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 125 mentions in open access literature.

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

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).

Zang L, et al. (2025) Bi-Allelic Loss-of-Function Variant in MAN1B1 Cause Rafiq Syndrome and Developmental Delay. International journal of molecular sciences, 26(16).

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.

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

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

Yousaf A, et al. (2024) Genome-Wide Mapping of Consanguineous Families Confirms Previously Implicated Gene Loci and Suggests New Loci in Specific Language Impairment (SLI). Children (Basel, Switzerland), 11(9).

Yasmin T, et al. (2024) Whole Genome Analysis in Consanguineous Families Reveals New Loci for Speech Sound Disorder (SSD). Genes, 15(8).

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.

Wang X, et al. (2022) A novel homozygous mutation in the PADI6 gene causes early embryo arrest. Reproductive health, 19(1), 190.

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.

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.

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.

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.

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.

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