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: production service resource, analysis service resource, data analysis service, service resource

Defining Citation: PMID:19465395

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

Funding:

Availability: Acknowledgement requested

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: 20250516T053619+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 119 mentions in open access literature.

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

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.

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.

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

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.

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.

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.

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.

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.

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.

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.

Salehi Z, et al. (2021) Exome sequencing reveals novel rare variants in Iranian familial multiple sclerosis: The importance of POLD2 in the disease pathogenesis. Genomics, 113(4), 2645.

Zhang Z, et al. (2020) Bi-allelic Missense Pathogenic Variants in TRIP13 Cause Female Infertility Characterized by Oocyte Maturation Arrest. American journal of human genetics, 107(1), 15.