Resource Summary Report

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Human Genome Variation database of Genotype-to-Phenotype information

RRID:SCR_007709
Type: Tool

Proper Citation

Human Genome Variation database of Genotype-to-Phenotype information (RRID:SCR_007709)

Resource Information

URL: https://www.gwascentral.org/

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Description: It re-directs to the ""GWAS Central"" resource, https://www.gwascentral.org/. Centralized compilation of summary level findings from genetic association studies, both large and small. They actively gather datasets from public domain projects, and encourage direct data submission from the community. HGVbaseG2P is built upon a basal layer of Markers that comprises all known SNPs and other variants from public databases such as dbSNP and the DBGV. Allele and genotype frequency data, plus genetic association significance findings, are added on top of the Marker data, and organized the same way that investigations are reported in typical journal manuscripts. Critically, no individual level genotypes or phenotypes are presented in HGVbaseG2P - only group level aggregated (summary level) data. The largest unit in a data submission is a Study, which can be thought of as being equivalent to one journal article. This may contain one or more Experiments, one or more Sample Panels of test subjects, and one or more Phenotypes. Sample Panels may be characterized in terms of various Phenotypes, and they also may be combined and/or split into Assayed Panels. The Assayed Panels are used as the basis for reporting allele/genotype frequencies (in `Genotype Experiments`) and/or genetic association findings (in ""Analysis Experiments""). Environmental factors are handled as part of the Sample Panel and Assayed Panel data structures.

Abbreviations: HGVbaseG2P, GWAS Central

Resource Type: data repository, storage service resource, database, service resource, data or information resource

Keywords: genotype, phenotype, genetic association study, allele, genotype frequency data, genetic association

Availability: Free, Freely available

Resource Name: Human Genome Variation database of Genotype-to-Phenotype information

Resource ID: SCR_007709

Alternate IDs: nif-0000-02958

Old URLs: http://hgvbase.cgb.ki.se/, http://www.hgvbaseg2p.org/index

Record Creation Time: 20220129T080243+0000

Record Last Update: 20260903T235809+0000

Ratings and Alerts

No rating or validation information has been found for Human Genome Variation database of Genotype-to-Phenotype information.

No alerts have been found for Human Genome Variation database of Genotype-to-Phenotype information.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 25 mentions in open access literature.

Listed below are recent publications. The full list is available at kravitz2 .

Lu Q, et al. (2023) Sema7A protects against high-fat diet-induced obesity and hepatic steatosis by regulating adipo/lipogenesis. Molecular metabolism, 70, 101698.

Sollis E, et al. (2023) The NHGRI-EBI GWAS Catalog: knowledgebase and deposition resource. Nucleic acids research, 51(D1), D977.

Beck T, et al. (2023) GWAS Central: an expanding resource for finding and visualising genotype and phenotype data from genome-wide association studies. Nucleic acids research, 51(D1), D986.

Günes Günsel G, et al. (2022) The arginine methyltransferase PRMT7 promotes extravasation of monocytes resulting in tissue injury in COPD. Nature communications, 13(1), 1303.

Hameed BMZ, et al. (2021) Big Data Analytics in urology: the story so far and the road ahead. Therapeutic advances in urology, 13, 1756287221998134.

Lavinda O, et al. (2021) Biophysical Compatibility of a Heterotrimeric Tyrosinase-TYRP1-TYRP2 Metalloenzyme Complex. Frontiers in pharmacology, 12, 602206.

Dong C, et al. (2021) INFIMA leverages multi-omics model organism data to identify effector genes of human GWAS variants. Genome biology, 22(1), 241.

Forgacova N, et al. (2021) Repurposing non-invasive prenatal testing data: Population study of single nucleotide variants associated with colorectal cancer and Lynch syndrome. Oncology letters, 22(5), 779.

Gupta R, et al. (2021) Artificial intelligence to deep learning: machine intelligence approach for drug discovery. Molecular diversity, 25(3), 1315.

Levi G, et al. (2021) DLX5/6 GABAergic Expression Affects Social Vocalization: Implications for Human Evolution. Molecular biology and evolution, 38(11), 4748.

Ignatieva EV, et al. (2021) Disease-associated genetic variants in the regulatory regions of human genes: mechanisms of action on transcription and genomic resources for dissecting these mechanisms. Vavilovskii zhurnal genetiki i selektsii, 25(1), 18.

Suzuki Y, et al. (2021) Diacylglycerol kinase ? colocalizes and interacts with apoptosis signal-regulating kinase 3 in response to osmotic shock. Biochemistry and biophysics reports, 26, 101006.

Bonello-Palot N, et al. (2020) High prevalence of mutations in perilipin 1 in patients with precocious acute coronary syndrome. Atherosclerosis, 293, 86.

Rigden DJ, et al. (2020) The 27th annual Nucleic Acids Research database issue and molecular biology database collection. Nucleic acids research, 48(D1), D1.

Xu C, et al. (2020) Annotation of susceptibility SNPs associated with atrial fibrillation. Aging, 12(17), 16981.

Diegel CR, et al. (2020) An osteocalcin-deficient mouse strain without endocrine abnormalities. PLoS genetics, 16(5), e1008361.

Beck T, et al. (2020) GWAS Central: a comprehensive resource for the discovery and comparison of genotype and phenotype data from genome-wide association studies. Nucleic acids research, 48(D1), D933.

Xin J, et al. (2020) Chromatin accessibility landscape and regulatory network of high-altitude hypoxia adaptation. Nature communications, 11(1), 4928.

Jiang CL, et al. (2020) Glucose transporter 10 modulates adipogenesis via an ascorbic acid-mediated pathway to protect mice against diet-induced metabolic dysregulation. PLoS genetics, 16(5), e1008823.

Noronha A, et al. (2019) The Virtual Metabolic Human database: integrating human and gut microbiome metabolism with nutrition and disease. Nucleic acids research, 47(D1), D614.