

Resource Summary Report

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HaploReg

RRID:SCR_006796

Type: Tool

Proper Citation

HaploReg (RRID:SCR_006796)

Resource Information

URL: <http://www.broadinstitute.org/mammals/haploreg/haploreg.php>

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Description: HaploReg is a tool for exploring annotations of the noncoding genome at variants on haplotype blocks, such as candidate regulatory SNPs at disease-associated loci. Using linkage disequilibrium (LD) information from the 1000 Genomes Project, linked SNPs and small indels can be visualized along with their predicted chromatin state in nine cell types, conservation across mammals, and their effect on regulatory motifs. HaploReg is designed for researchers developing mechanistic hypotheses of the impact of non-coding variants on clinical phenotypes and normal variation.

Abbreviations: HaploReg

Resource Type: data or information resource, database

Defining Citation: [PMID:22064851](#)

Keywords: chromatin state, conservation, regulatory motif, alteration, variant, chromatin, motif, annotation, genome, variation, genome-wide association study, refsnps, refseq gene, snp, bio.tools, FASEB list

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Resource Name: HaploReg

Resource ID: SCR_006796

Alternate IDs: biotools:HaploReg, nlx_151407

Alternate URLs: <http://compbio.mit.edu/HaploReg>, <https://bio.tools/HaploReg>

Record Creation Time: 20220129T080238+0000

Record Last Update: 20260804T043310+0000

Ratings and Alerts

No rating or validation information has been found for HaploReg.

No alerts have been found for HaploReg.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 1004 mentions in open access literature.

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

Liu Y, et al. (2025) Novel genetic variants in the NLRP3 inflammasome-related PANX1 and APP genes predict survival of patients with hepatitis B virus-related hepatocellular carcinoma. Clinical & translational oncology : official publication of the Federation of Spanish Oncology Societies and of the National Cancer Institute of Mexico, 27(2), 630.

Akhiiarova K, et al. (2025) Molecular Pathogenesis of Joint Hypermobility: The Role of Intergenic Interactions. Medical sciences (Basel, Switzerland), 13(4).

Chen YT, et al. (2025) Angiogenic Edge of ANGPT2: Genetic Variants Shape Prostate Cancer Prognosis on Androgen Deprivation Therapy. Cancer genomics & proteomics, 22(6), 991.

Cruz T, et al. (2025) Clinical, laboratory and genetic factors associated with smoking in a Brazilian Sick Cell Disease (SCD) cohort. PloS one, 20(9), e0332305.

Díaz-Belloso R, et al. (2025) HMGCR genetic variability in Parkinson's disease in a Spanish cohort: associations with lipid metabolism and early onset. Journal of neurology, 272(10), 671.

Lawrence ES, et al. (2025) Adaptive PRKAA1 variant in Andeans is associated with improved ventilation and sleep phenotypes. iScience, 28(8), 112911.

Shriner D, et al. (2025) Three Loci Affecting Variance of Body Mass Index in African Americans and Sub-Saharan Africans. Genetic epidemiology, 49(4), e70009.

Tchio C, et al. (2025) An integrative approach prioritizes the orphan GPR61 genomic region in tissue-specific regulation of chronotype. Sleep advances : a journal of the Sleep Research Society, 6(2), zpaf030.

Gálvez-Montosa F, et al. (2025) Polymorphisms within autophagy-related genes as susceptibility biomarkers for pancreatic cancer: A meta-analysis of three large European cohorts and functional characterization. *International journal of cancer*, 156(2), 339.

Shilenok VV, et al. (2025) Bioinformatic analysis of the regulatory potential of tagging SNPs provides evidence of the involvement of genes encoding the heat-resistant obscure (Hero) proteins in the pathogenesis of cardiovascular diseases. *Journal of integrative bioinformatics*, 22(1).

Meng H, et al. (2025) Genetic Predisposition and Mitochondrial Dysfunction in Sudden Cardiac Death: Role of MCU Complex Genetic Variations. *Cells*, 14(10).

Mogire RM, et al. (2025) OSBPL11 is an African-specific locus associated with 25-hydroxyvitamin D concentrations and cardiometabolic health. *medRxiv : the preprint server for health sciences*.

Li X, et al. (2025) Genetic modulation of IncPSMB1 confers non-syndromic cleft lip with or without cleft palate susceptibility by promoting cell apoptosis. *Communications biology*, 8(1), 1123.

Fabo TN, et al. (2025) Interactions Between Dietary Metabolites and Regulatory Risk Variants for Human Colon Cancer. *bioRxiv : the preprint server for biology*.

Ji G, et al. (2025) Genetic variants in m6A regulator genes confer susceptibility and progression of HCC in a Northern Chinese population. *Discover oncology*, 16(1), 526.

You D, et al. (2025) A genome-wide cross-trait analysis characterizes the shared genetic architecture between lung and gastrointestinal diseases. *Nature communications*, 16(1), 3032.

Hatzikotoulas K, et al. (2025) Translational genomics of osteoarthritis in 1,962,069 individuals. *Nature*, 641(8065), 1217.

Ping J, et al. (2025) A highland-adaptation variant near MCUR1 reduces its transcription and attenuates erythropoiesis in Tibetans. *Cell genomics*, 5(3), 100782.

Menon S, et al. (2025) Meta-analysis and in-silico functional characterization of the SNCA variant rs356220 in Parkinson's disease. *Scientific reports*, 15(1), 23358.

Satria RD, et al. (2025) Bioinformatic and genomic analysis identifies C allele of APOE rs7412 as the most prominent variant limiting extreme human longevity. *Scientific reports*, 15(1), 21688.