Resource Summary Report

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Human Variome Project

RRID:SCR_003492
Type: Tool

Proper Citation

Human Variome Project (RRID:SCR_003492)

Resource Information

URL: http://www.humanvariomeproject.org/

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Description: Project facilitating the establishment and maintenance of standards systems and infrastructure for the worldwide collection and sharing of all genetic variations effecting human disease. The Human Variome Project produces two categories of recommendations: HVP Standards and HVP Guidelines. HVP Standards are those systems, procedures and technologies that the Human Variome Project Consortium has determined should be used by the community. These carry more weight than the less prescriptive HVP Guidelines, which cover those systems, procedures and technologies that the Human Variome Project Consortium has determined would be beneficial for the community to adopt. HVP Standards and Guidelines are central to supporting the work of the Human Variome Project Consortium and cover a wide range of fields and disciplines, from ethics to nomenclature, data transfer protocols to collection protocols from clinics. They can be thought of as both technical manuals and scientific documents, and while the impact of HVP Standards and Guidelines differ, they are both generated in a similar fashion. A document has been generated both as a guide for those collecting and distributing data and for those developing policy. Items should include those generated by HGVS/HVP collaborators as well as those generated by groups of individual Societies and Standards bodies in all relevant fields worldwide.

Abbreviations: HVP

Synonyms: The Human Variome Project

Resource Type: data or information resource, knowledge environment, standard specification, international standard specification, narrative resource

Keywords: genetics, genomics, clinical, diagnosis, disease, human, genetic variation,

variome, data sharing

Related Condition: Genetic disease

Funding: Genomic Disorders Research Center ;
Howard Florey Institute ;
Human Genome Variation Society ;
University of Melbourne; Victoria; Australia ;
Victorian State Government ;
CASS Foundation ;
Gandel Foundation ;
Pierce Armstrong Foundation ;
Helen MacPherson Trust ;
UNESCO

Resource Name: Human Variome Project

Resource ID: SCR_003492

Alternate IDs: nif-0000-36300, OMICS_00282

Record Creation Time: 20220129T080219+0000

Record Last Update: 20250429T054831+0000

Ratings and Alerts

No rating or validation information has been found for Human Variome Project.

No alerts have been found for Human Variome Project.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 29 mentions in open access literature.

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

Hart RK, et al. (2024) HGVS Nomenclature 2024: improvements to community engagement, usability, and computability. Genome medicine, 16(1), 149.

Dy Closas AMF, et al. (2023) A KMT2B Frameshift Variant Causing Focal Dystonia Restricted to the Oromandibular Region After Long-Term Follow-up. Journal of movement disorders, 16(1), 91.

Korhonen PK, et al. (2022) "Escalibur"-A practical pipeline for the de novo analysis of nucleotide variation in nonmodel eukaryotes. Molecular ecology resources, 22(5), 2120.

Fanale D, et al. (2021) Prevalence and Spectrum of Germline BRCA1 and BRCA2 Variants of Uncertain Significance in Breast/Ovarian Cancer: Mysterious Signals From the Genome. Frontiers in oncology, 11, 682445.

McInerney-Leo AM, et al. (2020) Massively Parallel Sequencing for Rare Genetic Disorders: Potential and Pitfalls. Frontiers in endocrinology, 11, 628946.

Fanale D, et al. (2020) Detection of Germline Mutations in a Cohort of 139 Patients with Bilateral Breast Cancer by Multi-Gene Panel Testing: Impact of Pathogenic Variants in Other Genes beyond BRCA1/2. Cancers, 12(9).

Incorvaia L, et al. (2020) BRCA1/2 pathogenic variants in triple-negative versus luminal-like breast cancers: genotype-phenotype correlation in a cohort of 531 patients. Therapeutic advances in medical oncology, 12, 1758835920975326.

Incorvaia L, et al. (2020) Hereditary Breast and Ovarian Cancer in Families from Southern Italy (Sicily)-Prevalence and Geographic Distribution of Pathogenic Variants in BRCA1/2 Genes. Cancers, 12(5).

Kanzi AM, et al. (2020) Next Generation Sequencing and Bioinformatics Analysis of Family Genetic Inheritance. Frontiers in genetics, 11, 544162.

Chakrabarti S, et al. (2020) Phosphorylation of unique C-terminal sites of the mu-opioid receptor variants 1B2 and 1C1 influences their Gs association following chronic morphine. Journal of neurochemistry, 152(4), 449.

Katara P, et al. (2019) Pharmacogenes (PGx-genes): Current understanding and future directions. Gene, 718, 144050.

Boycott KM, et al. (2019) International collaborative actions and transparency to understand, diagnose, and develop therapies for rare diseases. EMBO molecular medicine, 11(5).

Cline MS, et al. (2018) BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS genetics, 14(12), e1007752.

Jonsson F, et al. (2018) Non-homologous recombination between Alu and LINE-1 repeats results in a 91 kb deletion in MERTK causing severe retinitis pigmentosa. Molecular vision, 24, 667.

Boycott KM, et al. (2017) International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American journal of human genetics, 100(5), 695.

Smith TD, et al. (2015) Standard development at the Human Variome Project. Database : the journal of biological databases and curation, 2015.

Cutting GR, et al. (2014) Annotating DNA variants is the next major goal for human genetics. American journal of human genetics, 94(1), 5.

van Schaik TA, et al. (2014) The need to redefine genomic data sharing: A focus on data accessibility. Applied & translational genomics, 3(4), 100.

Jimeno Yepes A, et al. (2014) Mutation extraction tools can be combined for robust recognition of genetic variants in the literature. F1000Research, 3, 18.

Barash CI, et al. (2014) The new Journal of Applied & Translational Genomics: No, not just another new journal. Applied & translational genomics, 3(3), 41.