Resource Summary Report

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Human Genome Variation Society

RRID:SCR_012989
Type: Tool

Proper Citation

Human Genome Variation Society (RRID:SCR_012989)

Resource Information

URL: http://www.hgvs.org

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Description: The Society aims to foster discovery and characterization of genomic variations including population distribution and phenotypic associations. We promote collection, documentation and free distribution of genomic variation information and associated clinical variations and endeavor to foster the development of the necessary methodology and informatics. Mission Statement To enhance human health through identification and characterization of changes in the genome that lead to susceptibility to illness. To this end, to collate the genomic information necessary for molecular diagnosis, research on basic mechanisms and design of treatments of human ailments. Society Journal Human Mutation is the Society journal. Members will receive a reduced subscription to the journal if they choose to subscribe. Meetings The Society holds two scientific meetings per year. One as a satellite to either the HUGO (Human Genome Organization) annual meeting or the ESHG (European Society of Human Genetics) annual meeting and one meeting is a satellite to the ASHG (American Society of Human Genetics annual meeting. The meetings are a forum for scientists to exchange ideas and form collaborations. Prominent speakers in the field are invited as well as a call for abstracts at large. The meetings are designed to update and increase knowledge of human genome variation and generally attract a stimulating and interesting collection of abstracts in all fields of human genome variation making it an ideal forum to share information and results. Past themes include: copy number variation, pathogenic or not?, pharmacogenomics, new DNA sequencing technologies, and genotype to pheontype relationships. We invite members and non-members alike to attend these meetings. The Society holds the Annual General Meeting of the members after the scientific meeting that is a satellite of the ASHG. Exhibitor''s booths The Society usually takes out an Exhibitor''s booth at the American & European Societies of Human Genetics annual meetings and sometimes the HUGO HGM meeting. GUIDELINES & RECOMMENDATIONS Members of the Society have formulated Guidelines & Recommendations on a number of topics, but especially for nomenclature of gene variations and guidelines on variation databases.

Abbreviations: HGVS

Resource Type: journal article, meeting resource, data or information resource, portal, community building portal, knowledge environment, training resource

Keywords: genetic variation, genome, homo sapiens genome, human, mutation, nomenclature, phenotypic associations, population distribution

Resource Name: Human Genome Variation Society

Resource ID: SCR_012989

Alternate IDs: nif-0000-23953

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Ratings and Alerts

No rating or validation information has been found for Human Genome Variation Society.

No alerts have been found for Human Genome Variation Society.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 291 mentions in open access literature.

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

Simonati A, et al. (2026) Age at onset and gene variants predict lifespan and disease duration in childhood neuronal ceroid lipofuscinoses. Developmental medicine and child neurology, 68(2), 276.

van der Leest EC, et al. (2025) Genotype-Phenotype Correlations, Treatment, and Prognosis of Children With Early-Onset (Neonatal) Marfan Syndrome. Clinical genetics, 108(2), 134.

Zhao P, et al. (2025) Clinical and genetic characterization of congenital disorders of glycosylation in 20 Chinese patients. Orphanet journal of rare diseases, 20(1), 625.

Abdelmaksoud-Dammak R, et al. (2025) Genetic profiling of inherited colorectal cancer syndromes in Tunisian patients. PloS one, 20(6), e0326343.

Littler S, et al. (2025) Targeting SUMOylation in ovarian cancer: Sensitivity, resistance, and the role of MYC. iScience, 28(6), 112555.

Han E, et al. (2025) Impact of HOMER2 frameshift extension variant on auditory function and development. Journal of molecular medicine (Berlin, Germany), 103(8), 975.

Aranaga-Decori AC, et al. (2025) Association of germline variants in the ZFX gene with primary hyperparathyroidism. PloS one, 20(8), e0329388.

Ammous-Boukhris N, et al. (2025) Germline and somatic mutational variants of Tunisian high grade serous ovarian cancer identified by next-generation sequencing. BMC cancer, 25(1), 1542.

Miloševi? V, et al. (2025) Genetics of Frontotemporal Dementia in the Serbian Population: Findings from a Hospital-Based Cohort. Neurology international, 17(10).

Hua R, et al. (2025) Clinical and functional characterization of the GABRB3 p.Met80Val variant in early-onset epilepsy with long-term follow-up. Italian journal of pediatrics, 51(1), 195.

Tighe A, et al. (2025) Screening a living biobank identifies cabazitaxel as a strategy to combat acquired taxol resistance in high-grade serous ovarian cancer. Cell reports. Medicine, 6(6), 102160.

Noorian S, et al. (2025) Isolated Growth Hormone Deficiency IA due to a Novel Homozygous Large Deletion ?1.6 kb Spanning Exons 1-4 of GH1 Gene: A Case Report. Clinical case reports, 13(2), e70234.

Fathi M, et al. (2025) Overview of genetic variants in a cohort of Iranian patients with leukodystrophy. Scientific reports, 15(1), 21898.

Alonso-García S, et al. (2025) Regulation of Stemness by NR1D2 in Colorectal Cancer. Biomedicines, 13(6).

Nikitin S, et al. (2024) Case Report: Exploring the clinical spectrum of LGMD R27: insights from a case study with homozygous pathogenic variant in the JAG2 gene. Frontiers in pediatrics, 12, 1414465.

Murtazina A, et al. (2024) Mild phenotype of CHAT-associated congenital myasthenic syndrome: case series. Frontiers in pediatrics, 12, 1280394.

He QB, et al. (2024) Functional assessment of a novel biallelic MYH3 variation causing CPSKF1B (contractures, pterygia, and spondylocarpotarsal fusion syndrome1B). Molecular genetics & genomic medicine, 12(3), e2401.

Asla Q, et al. (2024) Clinical and outcome comparison of genetically positive vs. negative patients in a large cohort of suspected familial hypocalciuric hypercalcemia. Endocrine, 83(3), 747.

Gao A, et al. (2024) Homologous recombination deficiency status predicts response to immunotherapy-based treatment in non-small cell lung cancer patients. Thoracic cancer, 15(25), 1842.

Chakrabarti I, et al. (2024) What Changed in CNS5? A Mini-Review on General Changes and Adult Diffuse Gliomas. Annals of African medicine, 23(3), 255.