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: community building portal, data or information resource, journal article, knowledge environment, meeting resource, portal, 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 337 mentions in open access literature.

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

Albokhari D, et al. (2026) Rare genetic causes of primary microcephaly in two Saudi families identified via whole-exome sequencing: Genomic and phenotypic delineation of pathogenic CDK5RAP2 and CIT variants. Molecular genetics and metabolism reports, 47, 101315.

Han SY, et al. (2026) Hearing loss phenotypes in Alport syndrome: experience in a tertiary referral center. Kidney research and clinical practice, 45(3), 357.

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.

Owrang D, et al. (2026) Uncovering dual molecular diagnoses in families with complex phenotypes through structural and clinical studies of novel COL4A6 variants. QJM : monthly journal of the Association of Physicians, 119(3), 187.

Huang YC, et al. (2026) Genotype-Specific Postural Control Deficits in Hemophilia A: Insights from Center of Pressure Analysis Beyond Radiographic Arthropathy. International journal of molecular sciences, 27(5).

Armengol-Alsina M, et al. (2026) Placental insufficiency markers to assess the risk of non-chromosomal genetic conditions in early-onset fetal growth restriction. Ultrasound in obstetrics & gynecology : the official journal of the International Society of Ultrasound in Obstetrics and Gynecology, 67(4), 492.

Rahmuni Y, et al. (2026) Implementing a Tiered Genetic Testing Strategy for Muscular Dystrophies in Morocco: From Targeted Assays to Exome Sequencing. Molecular genetics & genomic medicine, 14(3), e70192.

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.

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.

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

Naeemi H, et al. (2025) Contribution of MLH1, MSH2, and MSH6 large genomic rearrangements to Pakistani colorectal cancer patients. Hereditary cancer in clinical practice, 23(1), 26.

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

Di Muro E, et al. (2025) Long-Term Phenotypic Evolution in GRIN2A-Related Disorders: Electroclinical and Genetic Insights from Two Families with Extended Follow-Up. Genes, 16(3).

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.

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.

Rodríguez MR, et al. (2025) Elucidating the Role of KRAS, NRAS, and BRAF Mutations and Microsatellite Instability in Colorectal Cancer via Next-Generation Sequencing. Cancers, 17(13).

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.