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

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

Funding:

Resource Name: Human Genome Variation Society

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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 277 mentions in open access literature.

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

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.

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.

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.

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.

Gong B, et al. (2024) Towards accurate indel calling for oncopanel sequencing through an international pipeline competition at precisionFDA. Scientific reports, 14(1), 8165.

Abdelkhalek ZS, et al. (2024) Expanding the genotypic and phenotypic spectrum of Egyptian children with maple syrup urine disease. Scientific reports, 14(1), 28391.

Tong KY, et al. (2024) Novel PLCZ1 mutation caused polyspermy during in vitro fertilization. Asian journal of andrology, 26(4), 389.

Nawaz H, et al. (2024) Brachyolmia, dental anomalies and short stature (DASS): Phenotype and genotype analyses of Egyptian and Pakistani patients. Heliyon, 10(1), e23688.

Fernández-Cancio M, et al. (2024) Clinical and molecular study of patients with thyroid dyshormogenesis and variants in the thyroglobulin gene. Frontiers in endocrinology, 15, 1367808.

?im?ek E, et al. (2024) Screening of Mutations in Maturity-onset Diabetes of the Young-related Genes and RFX6 in Children with Autoantibody-negative Type 1 Diabetes Mellitus. Journal of clinical research in pediatric endocrinology, 16(2), 137.

Guidorizzi NR, et al. (2024) Comprehensive analysis of morbidity and mortality patterns in familial partial lipodystrophy patients: insights from a population study. Frontiers in endocrinology, 15, 1359211.

Herencia-Ropero A, et al. (2024) The PARP1 selective inhibitor saruparib (AZD5305) elicits potent and durable antitumor activity in patient-derived BRCA1/2-associated cancer models. Genome medicine, 16(1), 107.

Manotas MC, et al. (2023) Variant curation and interpretation in hereditary cancer genes: An institutional experience in Latin America. Molecular genetics & genomic medicine, 11(5), e2141.

Lee S, et al. (2023) Phenotypic and molecular basis of SIX1 variants linked to non-syndromic deafness and atypical branchio-otic syndrome in South Korea. Scientific reports, 13(1), 11776.

Mohammed I, et al. (2023) Understanding the Genetics of Early-Onset Obesity in a Cohort of Children From Qatar. The Journal of clinical endocrinology and metabolism, 108(12), 3201.

Yang K, et al. (2023) Assessment of a novel variation in DHODH gene causing Miller syndrome: The first report in Chinese population. Molecular genetics & genomic medicine, 11(7), e2186.

Lee SY, et al. (2023) Ramifications of POU4F3 variants associated with autosomal dominant hearing loss in various molecular aspects. Scientific reports, 13(1), 12584.

Marchi M, et al. (2023) TRPA1 rare variants in chronic neuropathic and nociplastic pain patients. Pain, 164(9), 2048.