Resource Summary Report

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HGVS Locus Specific Mutation Databases

RRID:SCR_006730
Type: Tool

Proper Citation

HGVS Locus Specific Mutation Databases (RRID:SCR_006730)

Resource Information

URL: http://www.hgvs.org/dblist/glsdb.htm

Proper Citation: HGVS Locus Specific Mutation Databases (RRID:SCR_006730)

Description: Database of Locus Specific Mutation Databases. Fields include HGNC Gene symbol / OMIM No., Database name / Internet address, and Curators. If you wish to add an LSDB please go to the LSDB Submission Page.

Abbreviations: LSMD

Synonyms: Locus Specific Mutation Databases

Resource Type: database, data or information resource

Keywords: mutation, locus, registry, catalog, genome variation, gene

Availability: The community can contribute to this resource

Resource Name: HGVS Locus Specific Mutation Databases

Resource ID: SCR_006730

Alternate IDs: nlx_153903

Record Creation Time: 20220129T080237+0000

Record Last Update: 20260803T040445+0000

Ratings and Alerts

No rating or validation information has been found for HGVS Locus Specific Mutation Databases.

No alerts have been found for HGVS Locus Specific Mutation Databases.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 138 mentions in open access literature.

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

Xue H, et al. (2024) Genetic testing for fetal loss of heterozygosity using single nucleotide polymorphism array and whole-exome sequencing. Scientific reports, 14(1), 2190.

Xiao F, et al. (2022) Dissecting mutational allosteric effects in alkaline phosphatases associated with different Hypophosphatasia phenotypes: An integrative computational investigation. PLoS computational biology, 18(3), e1010009.

Ullah S, et al. (2022) The Cancer Research Database (CRDB): Integrated Platform to Gain Statistical Insight Into the Correlation Between Cancer and COVID-19. JMIR cancer, 8(2), e35020.

Yu R, et al. (2020) Genetic and clinical phenotypic analysis of familial stapes sclerosis caused by an NOG mutation. BMC medical genomics, 13(1), 187.

Liu J, et al. (2019) Clinical value of genetic analysis in prenatal diagnosis of short femur. Molecular genetics & genomic medicine, 7(11), e978.

Jeannin G, et al. (2014) Recurrent exercise-induced acute renal failure in a young Pakistani man with severe renal hypouricemia and SLC2A9 compound heterozygosity. BMC medical genetics, 15, 3.

Kohlhase S, et al. (2014) Mutation analysis of the ERCC4/FANCQ gene in hereditary breast cancer. PloS one, 9(1), e85334.

Yan W, et al. (2014) Identification of more than two paternal haplotypes of the ovine fatty acid-binding protein 4 (FABP4) gene in half-sib families: evidence of intragenic meiotic recombination. PloS one, 9(2), e88691.

Farhan SM, et al. (2014) Exome sequencing identifies NFS1 deficiency in a novel Fe-S cluster disease, infantile mitochondrial complex II/III deficiency. Molecular genetics & genomic medicine, 2(1), 73.

Pangrazio A, et al. (2014) Exome sequencing identifies CTSK mutations in patients originally diagnosed as intermediate osteopetrosis. Bone, 59, 122.

Gong S, et al. (2014) NECTAR: a database of codon-centric missense variant annotations. Nucleic acids research, 42(Database issue), D1013.

Nguyen NM, et al. (2014) Genetics and Epigenetics of Recurrent Hydatidiform Moles: Basic Science and Genetic Counselling. Current obstetrics and gynecology reports, 3(1), 55.

Stenson PD, et al. (2014) The Human Gene Mutation Database: building a comprehensive mutation repository for clinical and molecular genetics, diagnostic testing and personalized genomic medicine. Human genetics, 133(1), 1.

Gottlieb B, et al. (2014) Changing genetic paradigms: creating next-generation genetic databases as tools to understand the emerging complexities of genotype/phenotype relationships. Human genomics, 8(1), 9.

Wu JY, et al. (2013) A novel NR5A1 variant in an infant with elevated testosterone from an Australasian cohort of 46,XY patients with disorders of sex development. Clinical endocrinology, 78(4), 545.

Pongor LS, et al. (2013) HeurAA: accurate and fast detection of genetic variations with a novel heuristic amplicon aligner program for next generation sequencing. PloS one, 8(1), e54294.

Jiao X, et al. (2013) Novel NR5A1 missense mutation in premature ovarian failure: detection in han chinese indicates causation in different ethnic groups. PloS one, 8(9), e74759.

van der Woerd WL, et al. (2013) Mutational analysis of ATP8B1 in patients with chronic pancreatitis. PloS one, 8(11), e80553.

Mellai M, et al. (2013) Promoter hypermethylation of the EMP3 gene in a series of 229 human gliomas. BioMed research international, 2013, 756302.

Hoefsloot LH, et al. (2013) EMQN Best Practice guidelines for diagnostic testing of mutations causing non-syndromic hearing impairment at the DFNB1 locus. European journal of human genetics : EJHG, 21(11), 1325.