Resource Summary Report

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Leiden Open Variation Database

RRID:SCR_006566
Type: Tool

Proper Citation

Leiden Open Variation Database (RRID:SCR_006566)

Resource Information

URL: http://www.LOVD.nl/

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Description: Freely available tool for Gene-centered collection and display of DNA variations. It also provides patient-centered data storage and storage of Next Generation Sequencing (NGS) data, even of variants outside of genes. Please note that LOVD provides a system for storage of information on genes and allelic variants. To obtain information about any genes or variants, do not download the LOVD package. This information should be obtained from the respective databases, http://www.lovd.nl/2.0/index_list.php In total: 2,507,027 variants (2,208,937 unique) in 170,935 individuals in 62619 genes in 88 LOVD installations. (Aug. 2013) LOVD 3.0 shared installation, http://databases.lovd.nl/shared/genes To maintain a high quality of the data stored, LOVD connects with various resources, like HGNC, NCBI, EBI and Mutalyzer. You can download LOVD in ZIP and GZIPped TARball formats.

Abbreviations: LOVD

Synonyms: Leiden Open Variation Database (LOVD)

Resource Type: data processing software, service resource, data repository, software application, software resource, data storage software, data or information resource, database, storage service resource

Defining Citation: PMID:21520333, PMID:15977173

Keywords: genetic variation, genomic variant, gene, transcript, disease, next generation sequencing, dna variation, variant, clinical, screening, locus, phenotype, sequence variation, allelic variant, data sharing, FASEB list

Funding: European Union FP7 GEN2PHEN 200754

Availability: The community can contribute to this resource, Clearance to contribute required, GNU General Public License, Acknowledgement requested

Resource Name: Leiden Open Variation Database

Resource ID: SCR_006566

Alternate IDs: nif-0000-02998, OMICS_00275, r3d100011905

Alternate URLs: https://doi.org/10.17616/R3993T

Record Creation Time: 20220129T080236+0000

Record Last Update: 20260731T042726+0000

Ratings and Alerts

No rating or validation information has been found for Leiden Open Variation Database.

No alerts have been found for Leiden Open Variation Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 238 mentions in open access literature.

Listed below are recent publications. The full list is available at kravitz2 .

Shi Q, et al. (2025) A novel targeted long-read sequencing-based preimplantation genetic testing method for ??-thalassemia (tlrPGT-??-thal). Journal of assisted reproduction and genetics, 42(8), 2565.

Bayam E, et al. (2025) Bi-allelic variants in WDR47 cause a complex neurodevelopmental syndrome. EMBO molecular medicine, 17(1), 129.

Smith TB, et al. (2025) Bi-allelic variants in DAP3 result in reduced assembly of the mitoribosomal small subunit with altered apoptosis and a Perrault-syndrome-spectrum phenotype. American journal of human genetics, 112(1), 59.

Yang M, et al. (2025) Uncovering the genetic variation spectrum of colorectal polyposis from a multicentre cohort in China. NPJ precision oncology, 9(1), 75.

Yalcin Z, et al. (2025) Two Novel Protein-Truncating Variants in NLRP2 and Their Functional Impacts on the Subcortical Maternal Complex. Clinical genetics, 108(2), 179.

Wu J, et al. (2025) Expanding the Clinical and Genetic Spectrum of Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency: Insights From Two Unrelated Chinese Families. Molecular genetics & genomic medicine, 13(4), e70097.

De Marchis L, et al. (2025) BRCA Screening and Identification of a Common Haplotype in the Jewish Community of Rome Reveal a Founder Effect for the c.7007G>C, p. (Arg2336Pro) BRCA2 Variant. Cancers, 17(12).

Ng HY, et al. (2025) Identification of technically challenging variants: Whole-genome sequencing improves diagnostic yield in patients with high clinical suspicion of rare diseases. HGG advances, 6(3), 100469.

Garrigue A, et al. (2025) Human oncostatin M deficiency underlies an inherited severe bone marrow failure syndrome. The Journal of clinical investigation, 135(6).

Gambardella A, et al. (2025) PAK3 pathogenic variant associated with sleep-related hypermotor epilepsy in a family with parental mosaicism. Epilepsia open, 10(2), 593.

Singh S, et al. (2025) Biallelic variants in CCN2 underlie an autosomal recessive kyphomelic dysplasia. European journal of human genetics : EJHG, 33(1), 30.

Thomas HB, et al. (2025) Bi-allelic variants in MRPL49 cause variable clinical presentations, including sensorineural hearing loss, leukodystrophy, and ovarian insufficiency. American journal of human genetics, 112(4), 952.

Oaxaca-Castillo D, et al. (2025) A Novel 1259 bp Intragenic Deletion in the GJB2 Gene in a Mexican Family with Congenital Profound Hearing Loss. Audiology research, 15(5).

Blandin C, et al. (2025) Genetics and Bone Mineral Density Predict the Fractures in Adults With Osteogenesis Imperfecta: A Prospective Study. The Journal of clinical endocrinology and metabolism, 110(8), 2307.

Chetta M, et al. (2025) Lost in .*VCF Translation. From Data Fragmentation to Precision Genomics: Technical, Ethical, and Interpretive Challenges in the Post-Sequencing Era. Journal of personalized medicine, 15(8).

Mikhaylenko DS, et al. (2025) Patients with Papillary Renal Cancer and Germline Duplication of MET Exons 5-21. Biomedicines, 13(6).

Campobasso G, et al. (2025) Investigating the Role of B9D1 in Meckel-Gruber Syndrome: A Case Report and Comprehensive Literature Review. Genes, 16(6).

Rawnsley K, et al. (2025) Comprehensive functional splicing analysis of non-canonical CNGB3 variants using in vitro minigene splice assays. The Journal of pathology, 266(3), 322.

Quinodoz M, et al. (2025) De novo and inherited dominant variants in U4 and U6 snRNAs cause retinitis pigmentosa. medRxiv : the preprint server for health sciences.

Swetha J, et al. (2025) A 250-kb Microdeletion Identified in Chromosome 16 Is Associated With Non-Syndromic Sensorineural Hearing Loss in a South Indian Consanguineous Family. Journal of audiology & otology, 29(1), 31.