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 or information resource, data processing software, data repository, data storage software, database, service resource, software application, software resource, 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: 20260905T132556+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 315 mentions in open access literature.

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

Takeuchi F, et al. (2026) Simultaneous Assessment of Genetic and Epigenetic Contributions to the Plasma Lipid Levels with Respect to Cardiovascular Risk. Journal of atherosclerosis and thrombosis, 33(6), 717.

Cha?upczy?ska B, et al. (2026) Molecular Profiling of Polish Pediatric Patients with Epilepsy: A Single-Center Diagnostic Experience Using Next-Generation Sequencing. Genes, 17(2).

Serpieri V, et al. (2026) Bi-allelic variants in FSD1L cause a neurodevelopmental disorder overlapping with L1 syndrome. American journal of human genetics, 113(3), 600.

Lyu P, et al. (2026) Database of CLCN5 Pathogenic Variants Causing Dent Disease. Kidney international reports, 11(5), 106475.

Hu J, et al. (2026) Evaluation and management of DMD gene copy number variations detected by prenatal SNP-array testing. BMC medical genomics, 19(1).

Anzoom N, et al. (2026) Modeling the Effects of Single Nucleotide Polymorphisms (SNPs) on the Structure and Function of the Human RET Gene: An In Silico Study. Human mutation, 2026, 8848146.

Quinodoz M, et al. (2026) De novo and inherited dominant variants in U4 and U6 snRNA genes cause retinitis pigmentosa. Nature genetics, 58(1), 169.

Dell'Oca M, et al. (2026) NEDAMSS syndrome-related truncating and missense mutations are associated with aberrant liquid-liquid phase separation of IRF2BPL. Nature communications, 17(1).

Beljan JC, et al. (2026) U7snRNA-mediated skipping of intron-derived pseudoexons restores full-length DMD expression in patient-derived cell lines. Molecular therapy. Advances, 34(2), 201750.

Khandelwal MH, et al. (2026) Familial WT1-associated nephropathy - 46, XY Frasier syndrome and 46, XX steroid-resistant nephrotic syndrome in female siblings: A case report and review of literature. World journal of nephrology, 15(1), 110867.

Reuter P, et al. (2026) Systematic functional evaluation of CNGA1 missense variants associated with retinitis pigmentosa. Molecular medicine (Cambridge, Mass.), 32(1).

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).

Shekfeh M, et al. (2025) Prevalence and impact of molecular variation in the three-prime repair exonuclease 1 TREX1 and its implications for oncology. Human genomics, 19(1), 73.

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.

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.

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.

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

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.

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.

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.