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 storage software, service resource, database, storage service resource, software resource, data repository, software application, data processing software

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

Record Creation Time: 20220129T080236+0000

Record Last Update: 20250521T061116+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 210 mentions in open access literature.

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

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.

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

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

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.

Deitch I, et al. (2024) Autosomal Recessive Rod-Cone Dystrophy with Mild Extra-Ocular Manifestations Due to a Splice-Affecting Variant in BBS9. Current issues in molecular biology, 46(3), 2566.

Riedhammer KM, et al. (2024) Implication of transcription factor FOXD2 dysfunction in syndromic congenital anomalies of the kidney and urinary tract (CAKUT). Kidney international, 105(4), 844.

Puapatanakul P, et al. (2024) Alport syndrome and Alport kidney diseases - elucidating the disease spectrum. Current opinion in nephrology and hypertension, 33(3), 283.

Andhika NS, et al. (2024) Using computational approaches to enhance the interpretation of missense variants in the PAX6 gene. European journal of human genetics : EJHG, 32(8), 1005.

Günbey C, et al. (2024) Horizontal gaze palsy with progressive scoliosis: Further expanding the ROBO3 spectrum. Annals of clinical and translational neurology, 11(8), 2088.

Donato L, et al. (2024) The genomic mosaic of mitochondrial dysfunction: Decoding nuclear and mitochondrial epigenetic contributions to maternally inherited diabetes and deafness pathogenesis. Heliyon, 10(14), e34756.

Buonfiglio PI, et al. (2024) In silico and in vivo analyses of a novel variant in MYO6 identified in a family with postlingual non-syndromic hearing loss from Argentina. NAR genomics and bioinformatics, 6(4), lqae162.

Efthymiou S, et al. (2024) Novel loss-of-function variants expand ABCC9-related intellectual disability and myopathy syndrome. Brain : a journal of neurology, 147(5), 1822.

Akhtar Z, et al. (2024) Phenotypic and Genetic Heterogeneity of a Pakistani Cohort of 15 Consanguineous Families Segregating Variants in Leber Congenital Amaurosis-Associated Genes. Genes, 15(12).

Shen C, et al. (2024) A novel homozygous RSPH4A variant in a family with primary ciliary dyskinesia and literature review. Frontiers in genetics, 15, 1364476.

Martinez-Montoya V, et al. (2024) Mutational spectrum and genotype-phenotype correlation in Mexican patients with infantile-onset and late-onset Pompe disease. Molecular genetics & genomic medicine, 12(7), e2480.

Coetzer KC, et al. (2024) Genetic basis of osteogenesis imperfecta from a single tertiary centre in South Africa. European journal of human genetics : EJHG, 32(10), 1285.

Huang B, et al. (2024) A novel GATA3 frameshift mutation causes hypoparathyroidism, sensorineural deafness, and renal dysplasia syndrome. Molecular genetics and metabolism reports, 38, 101063.

De Paolis E, et al. (2024) The novel CFTR haplotype E583G/F508del in CFTR-related disorder. Molecular biology reports, 51(1), 849.

Erinç A, et al. (2024) Birt-Hogg-Dubé syndrome with novel FLCN gene mutations and different clinical presentations: Case series. Tuberkuloz ve toraks, 72(1), 91.

Ibrahim M, et al. (2024) ABCA4-related retinopathies in Lebanon. Heliyon, 10(9), e30304.