Resource Summary Report

Generated by <u>dkNET</u> on Apr 15, 2025

<u>dbVar</u>

RRID:SCR_003219 Type: Tool

Proper Citation

dbVar (RRID:SCR_003219)

Resource Information

URL: http://www.ncbi.nlm.nih.gov/dbvar/

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Description: Structural variation database designed to store data on variant DNA > / = 1 bp in size from all organisms. Associations of defined variants with phenotype information is also provided. Users can browse data containing number of variant cells from each study, and filter studies by organism, study type, method and genomic variant. Organisms include human, mouse, cattle and several additional animals.

Abbreviations: dbVar

Synonyms: dbVar, Database of Genomic Structural Variation, NCBI dbVar

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

Defining Citation: PMID:23193291

Keywords: structure, variation, structural variation, genetics, insertion, deletion, copy number variant, inversion, translocation, genomic imbalance, genotype, gene expression, dna, genomics, phenotype, genetic code

Funding:

Availability: Free, Freely available

Resource Name: dbVar

Resource ID: SCR_003219

Alternate IDs: nlx_157217

Record Creation Time: 20220129T080217+0000

Record Last Update: 20250412T054811+0000

Ratings and Alerts

No rating or validation information has been found for dbVar.

No alerts have been found for dbVar.

Data and Source Information

Source: <u>SciCrunch Registry</u>

Usage and Citation Metrics

We found 180 mentions in open access literature.

Listed below are recent publications. The full list is available at <u>dkNET</u>.

Wang Z, et al. (2024) VarCards2: an integrated genetic and clinical database for ACMG-AMP variant-interpretation guidelines in the human whole genome. Nucleic acids research, 52(D1), D1478.

Rodrigues Alves Barbosa V, et al. (2024) Single variant, yet "double trouble": TSC and KBG syndrome because of a large de novo inversion. Life science alliance, 7(4).

Hayes V, et al. (2024) Rare pathogenic structural variants show potential to enhance prostate cancer germline testing for African men. Research square.

Schrauwen I, et al. (2024) Optical genome mapping unveils hidden structural variants in neurodevelopmental disorders. Scientific reports, 14(1), 11239.

Ma Y, et al. (2024) Complete F9 Gene Deletion, Duplication, and Triplication Rearrangements: Implications for Factor IX Expression and Clinical Phenotypes. Thrombosis and haemostasis, 124(4), 374.

Yuan N, et al. (2024) Comprehensive assessment of long-read sequencing platforms and calling algorithms for detection of copy number variation. Briefings in bioinformatics, 25(5).

Järvelä I, et al. (2024) Heterogeneous genetic patterns in bilateral perisylvian polymicrogyria: insights from a Finnish family cohort. Brain communications, 6(3), fcae142.

Guner Yilmaz B, et al. (2024) Rapid genome sequencing for critically ill infants: an inaugural pilot study from Turkey. Frontiers in pediatrics, 12, 1412880.

Wang Y, et al. (2024) Quantifying the regulatory potential of genetic variants via a hybrid sequence-oriented model with SVEN. Nature communications, 15(1), 10917.

Du H, et al. (2024) HMZDupFinder: a robust computational approach for detecting intragenic homozygous duplications from exome sequencing data. Nucleic acids research, 52(4), e18.

Geoffroy V, et al. (2023) The AnnotSV webserver in 2023: updated visualization and ranking. Nucleic acids research, 51(W1), W39.

Lee WP, et al. (2023) Structural Variation Detection and Association Analysis of Whole-Genome-Sequence Data from 16,905 Alzheimer's Diseases Sequencing Project Subjects. Research square.

Lee S, et al. (2023) Exploring quantitative traits-associated copy number deletions through reanalysis of UK10K consortium whole genome sequencing cohorts. BMC genomics, 24(1), 787.

Xu Z, et al. (2023) PhenoSV: interpretable phenotype-aware model for the prioritization of genes affected by structural variants. Nature communications, 14(1), 7805.

Jun G, et al. (2023) Structural variation across 138,134 samples in the TOPMed consortium. bioRxiv : the preprint server for biology.

Hussain M, et al. (2023) A Novel CRYBB2 Silent Variant in Autosomal Dominant Congenital Cataracts (ADCC) in Pakistani families. Pakistan journal of medical sciences, 39(5), 1399.

Wang H, et al. (2023) Structural Variation Detection and Association Analysis of Whole-Genome-Sequence Data from 16,905 Alzheimer's Diseases Sequencing Project Subjects. medRxiv : the preprint server for health sciences.

Kim P, et al. (2023) Systematic investigation of the homology sequences around the human fusion gene breakpoints in pan-cancer - bioinformatics study for a potential link to MMEJ. Briefings in bioinformatics, 24(5).

Lv K, et al. (2023) dbCNV: deleteriousness-based model to predict pathogenicity of copy number variations. BMC genomics, 24(1), 131.

Fei Y, et al. (2023) Whole-genome sequencing revealed a novel long-range deletion mutation spanning GNAS in familial pseudohypoparathyroidism. Molecular genetics & genomic medicine, 11(5), e2144.