

Resource Summary Report

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KAVIAR

RRID:SCR_013737

Type: Tool

Proper Citation

KAVIAR (RRID:SCR_013737)

Resource Information

URL: <http://db.systemsbiology.net/kaviar/>

Proper Citation: KAVIAR (RRID:SCR_013737)

Description: A database containing a compilation of SNVs, indels, and complex variants observed in humans, designed to facilitate testing for the novelty and frequency of observed variants.

Synonyms: queryable database of known variants, Known VARiants

Resource Type: database, data or information resource

Defining Citation: [PMID:21965822](#)

Keywords: SNV, single nucleotide variant, database, indel, bio.tools

Funding: Inova Translational Medicine Institute

Availability: Free, Public

Resource Name: KAVIAR

Resource ID: SCR_013737

Alternate IDs: biotools:kaviar

Alternate URLs: <https://bio.tools/kaviar>

Record Creation Time: 20220129T080317+0000

Record Last Update: 20260725T191206+0000

Ratings and Alerts

No rating or validation information has been found for KAVIAR.

No alerts have been found for KAVIAR.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 17 mentions in open access literature.

Listed below are recent publications. The full list is available at [nidm-terms](#) .

Dong J, et al. (2025) A Case Study Identified a New Mutation in the TTN Gene for Inherited Hypertrophic Cardiomyopathy. International journal of general medicine, 18, 447.

Yin JA, et al. (2025) Arrayed CRISPR libraries for the genome-wide activation, deletion and silencing of human protein-coding genes. Nature biomedical engineering, 9(1), 127.

Berry V, et al. (2024) A novel frameshift variant in BCOR causes congenital nuclear cataract. Ophthalmic genetics, 45(6), 591.

Rich J, et al. (2024) DiPRO1 distinctly reprograms muscle and mesenchymal cancer cells. EMBO molecular medicine, 16(8), 1840.

Zhao Z, et al. (2023) Whole exome sequencing of well-differentiated liposarcoma and dedifferentiated liposarcoma in older woman: a case report. Frontiers in medicine, 10, 1237246.

Berry V, et al. (2022) Variants in PAX6, PITX3 and HSF4 causing autosomal dominant congenital cataracts. Eye (London, England), 36(8), 1694.

Lin CW, et al. (2021) Genetic Spectrum and Characteristics of Hereditary Optic Neuropathy in Taiwan. Genes, 12(9).

Chen TC, et al. (2021) Genetic characteristics and epidemiology of inherited retinal degeneration in Taiwan. NPJ genomic medicine, 6(1), 16.

Qiao X, et al. (2021) Pathogenesis and characteristics of large ameloblastoma of the jaw: a report of two rare cases. The Journal of international medical research, 49(5), 3000605211014803.

Marchetti C, et al. (2020) Feasibility of tumor testing for BRCA status in high-grade serous ovarian cancer using fresh-frozen tissue based approach. Gynecologic oncology, 158(3), 740.

Felipe-Medina N, et al. (2020) A missense in HSF2BP causing primary ovarian insufficiency affects meiotic recombination by its novel interactor C19ORF57/BRME1. eLife, 9.

Codina-Sola M, et al. (2019) Genetic factors contributing to autism spectrum disorder in Williams-Beuren syndrome. *Journal of medical genetics*, 56(12), 801.

Caburet S, et al. (2019) A truncating MEIOB mutation responsible for familial primary ovarian insufficiency abolishes its interaction with its partner SPATA22 and their recruitment to DNA double-strand breaks. *EBioMedicine*, 42, 524.

Klemann C, et al. (2019) Clinical and Immunological Phenotype of Patients With Primary Immunodeficiency Due to Damaging Mutations in NFKB2. *Frontiers in immunology*, 10, 297.

Liskova P, et al. (2018) Ectopic GRHL2 Expression Due to Non-coding Mutations Promotes Cell State Transition and Causes Posterior Polymorphous Corneal Dystrophy 4. *American journal of human genetics*, 102(3), 447.

Traficante G, et al. (2015) Prenatal diagnosis of X-linked adrenoleukodystrophy associated with isolated pericardial effusion. *Clinical case reports*, 3(7), 643.

Kocak H, et al. (2014) Hoyerdal-Hreidarsson syndrome caused by a germline mutation in the TEL patch of the telomere protein TPP1. *Genes & development*, 28(19), 2090.