

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: data or information resource, database

Defining Citation: [PMID:21965822](https://pubmed.ncbi.nlm.nih.gov/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: 20250331T061208+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 [FDI Lab - SciCrunch.org](#).

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) Hoyeraal-Hreidarsson syndrome caused by a germline mutation in the TEL patch of the telomere protein TPP1. *Genes & development*, 28(19), 2090.