Resource Summary Report

Generated by kravitz2 on Jul 31, 2026

Exomiser

RRID:SCR_002192
Type: Tool

Proper Citation

Exomiser (RRID:SCR_002192)

Resource Information

URL: http://www.sanger.ac.uk/resources/databases/exomiser/query/exomiser2

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Description: A Java program that functionally annotates variants from whole-exome sequencing data starting from a VCF (Variant Call Format) file (version 4). The functional annotation code is based on Annovar and uses UCSCKnownGene transcript definitions and hg19 genomic coordinates. Variants are prioritized according to user-defined criteria on variant frequency, pathogenicity, quality, inheritance pattern, phenotype data from human and model organisms, and proximity in the interactome to phenotypically similar genes.

Abbreviations: Exomiser, Exomiser2

Synonyms: The Exomiser2: Annotate and Filter Variants, Exomiser2: Annotate and Filter Variants, Exomiser 2.0

Resource Type: service resource, data analysis service, software resource, production service resource, analysis service resource

Defining Citation: PMID:24162188

Keywords: java, functional annotation, function, variant, whole-exome sequencing, gene, phenotype, model organism

Availability: Free, Freely available

Resource Name: Exomiser

Resource ID: SCR_002192

Alternate IDs: SciRes_000142

Record Creation Time: 20220129T080212+0000

Record Last Update: 20260729T044022+0000

Ratings and Alerts

No rating or validation information has been found for Exomiser.

No alerts have been found for Exomiser.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 60 mentions in open access literature.

Listed below are recent publications. The full list is available at kravitz2 .

Zou J, et al. (2025) Genetic and clinical insights into acute kidney injury in maturity-onset diabetes of the young caused by the ABCC8 c.2500C>T mutation: Potential risk factors associated with SGLT2 inhibitors. Experimental and therapeutic medicine, 30(2), 164.

Sudhakar DV, et al. (2025) Exome sequencing uncovers promising candidate genes for foetal structural malformations. The Indian journal of medical research, 161(5), 510.

Lam WKJ, et al. (2025) The implementation of genome sequencing in rare genetic diseases diagnosis: a pilot study from the Hong Kong genome project. The Lancet regional health. Western Pacific, 55, 101473.

Alonso-González A, et al. (2025) A tiered strategy to identify relevant genetic variants in familial pulmonary fibrosis: a proof of concept for the clinical practice. European journal of human genetics : EJHG, 33(11), 1509.

Yu HP, et al. (2025) Diagnostic journey and genetic analysis of a novel homozygous CYP2U1 mutation causing autosomal recessive spastic paraplegia type 56 (SPG56) in a consanguineous family. BMC neurology, 25(1), 207.

Xi AMC, et al. (2025) Clinical and data-driven optimization of Genomiser for rare disease patients: experience from the Hong Kong Genome Project. Briefings in bioinformatics, 26(5).

Matentzoglu N, et al. (2025) The Unified Phenotype Ontology : a framework for cross-species integrative phenomics. Genetics, 229(3).

Zhu J, et al. (2025) Clinical genetic analysis of an adult polyglucosan body disease (APBD) family caused by the compound heterozygous variant of GBE1 p.R156C and deletion exon 3-7. Frontiers in genetics, 16, 1514610.

Kafkas ?, et al. (2025) The application of Large Language Models to the phenotype-based prioritization of causative genes in rare disease patients. Scientific reports, 15(1), 15093.

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.

Gold JI, et al. (2025) Exclusion-based exome sequencing in critically ill adults 18-40 years old has a 24% diagnostic rate and finds racial disparities in access to genetic testing. American journal of human genetics, 112(8), 1792.

Godinez-Zamora GF, et al. (2025) Contribution of next generation sequencing to the diagnosis of inborn errors of immunity in a pediatric cohort. Frontiers in immunology, 16, 1638544.

Gold J, et al. (2024) Universal Exome Sequencing in Critically Ill Adults: A Diagnostic Yield of 25% and Race-Based Disparities in Access to Genetic Testing. medRxiv : the preprint server for health sciences.

Gravel B, et al. (2024) Prioritization of oligogenic variant combinations in whole exomes. Bioinformatics (Oxford, England), 40(4).

Matentzoglu N, et al. (2024) The Unified Phenotype Ontology (uPheno): A framework for cross-species integrative phenomics. bioRxiv : the preprint server for biology.

Wang X, et al. (2024) DNAH3 deficiency causes flagellar inner dynein arm loss and male infertility in humans and mice. eLife, 13.

Werren EA, et al. (2024) A de novo variant in PAK2 detected in an individual with Knobloch type 2 syndrome. bioRxiv : the preprint server for biology.

Yuan X, et al. (2024) Refined preferences of prioritizers improve intelligent diagnosis for Mendelian diseases. Scientific reports, 14(1), 2845.

Odrzywolski A, et al. (2024) Gollop-Wolfgang Complex Is Associated with a Monoallelic Variation in WNT11. Genes, 15(1).

Sheth H, et al. (2024) Development, validation and application of single molecule molecular inversion probe based novel integrated genetic screening method for 29 common lysosomal storage disorders in India. Human genomics, 18(1), 46.