Resource Summary Report

Generated by PRECISE-TBI on Sep 8, 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

Proper Citation: Exomiser (RRID:SCR_002192)

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: analysis service resource, data analysis service, production service resource, service resource, software 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: 20260905T132954+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 71 mentions in open access literature.

Listed below are recent publications. The full list is available at PRECISE-TBI .

Sheth J, et al. (2026) Development of national biobank for lysosomal storage disorders in India- a step towards advancing research and precision medicine. Orphanet journal of rare diseases, 21(1), 65.

Smits DJ, et al. (2026) Nanopore long-read sequencing for the critically ill facilitates ultrarapid diagnostics and urgent clinical decision making. European journal of human genetics : EJHG, 34(1), 108.

Lawless D, et al. (2026) Application of qualifying variants for genomic analysis. Bioinformatics (Oxford, England), 42(2).

Ratnaike TE, et al. (2026) A systematic analysis of mitochondrial aminoacyl tRNA synthetase variants in a rare disease cohort. European journal of human genetics : EJHG, 34(3), 395.

Zhao W, et al. (2026) An agentic system for rare disease diagnosis with traceable reasoning. Nature, 651(8106), 775.

Reese JT, et al. (2026) Systematic benchmarking demonstrates large language models have not reached the diagnostic accuracy of traditional rare-disease decision support tools. European journal of human genetics : EJHG, 34(4), 498.

Ma J, et al. (2026) Identification of a Novel Biallelic CFAP119 Variant in an Infertile Man with Asthenoteratozoospermia. The world journal of men's health, 44(1), 203.

Nicholls HL, et al. (2026) Genome-wide analysis of cardiac ventricular phenotypes reveals novel loci and therapeutic targets for heart failure. Nature communications, 17(1).

Lee J, et al. (2026) LA-MARRVEL: A Knowledge-Grounded, Language-Aware LLM Framework for Clinically Robust Rare Disease Gene Prioritization. ArXiv.

Kong F, et al. (2026) A Genetic Landscape of Euploid Miscarriages From Couples With Recurrent Pregnancy Loss Through Whole Exome Sequencing. Molecular genetics & genomic medicine, 14(4), e70220.

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

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.

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.

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.

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.

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.

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

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.

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.

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.