

Resource Summary Report

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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, analysis service resource, software resource, production 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

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 [nidm-terms](#) .

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).

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.

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.

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.

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

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.

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).

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

Matentzoglou N, et al. (2024) The Unified Phenotype Ontology (uPheno): A framework for cross-species integrative phenomics. *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.

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*.

Wang C, et al. (2024) High-depth whole-genome sequencing identifies structure variants, copy number variants and short tandem repeats associated with Parkinson's disease. *NPJ Parkinson's disease*, 10(1), 134.

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.