Resource Summary Report

Generated by NIF on Apr 29, 2025

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, production service resource, data analysis service, analysis service resource, software resource

Defining Citation: PMID:24162188

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

Funding:

Resource Name: Exomiser

Resource ID: SCR_002192

Alternate IDs: SciRes_000142

Record Creation Time: 20220129T080212+0000

Record Last Update: 20250429T054718+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 48 mentions in open access literature.

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

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

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.

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.

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

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.

Burleigh A, et al. (2024) Genetic testing of Behçet's disease using next-generation sequencing to identify monogenic mimics and HLA-B*51. Rheumatology (Oxford, England), 63(12), 3457.

Ruan DD, et al. (2024) Clinical phenotype and genetic function analysis of a family with hypomyelinating leukodystrophy-7 caused by POLR3A mutation. Scientific reports, 14(1), 7638.

Kim HH, et al. (2024) Explicable prioritization of genetic variants by integration of rule-based and machine learning algorithms for diagnosis of rare Mendelian disorders. Human genomics, 18(1), 28.

Vestito L, et al. (2024) Efficient reinterpretation of rare disease cases using Exomiser. NPJ genomic medicine, 9(1), 65.

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.

Danzi MC, et al. (2023) Deep structured learning for variant prioritization in Mendelian diseases. Nature communications, 14(1), 4167.

Lenassi E, et al. (2023) EyeG2P: an automated variant filtering approach improves efficiency of diagnostic genomic testing for inherited ophthalmic disorders. Journal of medical genetics, 60(8), 810.

Sundaramurthi JC, et al. (2023) De novo TRPM3 missense variant associated with neurodevelopmental delay and manifestations of cerebral palsy. Cold Spring Harbor molecular case studies, 9(4).

Ververi A, et al. (2023) Germline homozygous missense DEPDC5 variants cause severe refractory early-onset epilepsy, macrocephaly and bilateral polymicrogyria. Human molecular genetics, 32(4), 580.

Pagnamenta AT, et al. (2023) Structural and non-coding variants increase the diagnostic yield of clinical whole genome sequencing for rare diseases. Genome medicine, 15(1), 94.

Nguyen Q, et al. (2023) Can artificial intelligence accelerate the diagnosis of inherited retinal diseases? Protocol for a data-only retrospective cohort study (Eye2Gene). BMJ open, 13(3), e071043.

Alsentzer E, et al. (2023) Simulation of undiagnosed patients with novel genetic conditions. Nature communications, 14(1), 6403.