Resource Summary Report

Generated by NIF on Aug 26, 2026

Phevor

RRID:SCR_002273
Type: Tool

Proper Citation

Phevor (RRID:SCR_002273)

Resource Information

URL: http://weatherby.genetics.utah.edu/cgi-bin/Phevor/PhevorWeb.html

Proper Citation: Phevor (RRID:SCR_002273)

Description: THIS RESOURCE IS NO LONGER IN SERVICE. Documented on October 28,2025. Tool that integrates phenotype, gene function, and disease information with personal genomic data for improved power to identify disease-causing alleles. It works by combining knowledge resident in multiple biomedical ontologies with the outputs of variant prioritization tools. It does so using an algorithm that propagates information across and between ontologies. This process enables Phevor to accurately reprioritize potentially damaging alleles identified by variant prioritization tools in light of gene function, disease, and phenotype knowledge. Phevor is especially useful for single exome and family trio-based diagnostic analyses, the most commonly occurring clinical scenarios, and ones for which existing personal-genomes diagnostic tools are most inaccurate and underpowered. Phevor not only improves diagnostic accuracy for individuals presenting with established disease phenotypes, but also for those with previously undescribed and atypical disease presentations. Importantly, Phevor is not limited to known diseases, or known disease-causing alleles.

Abbreviations: Phevor

Synonyms: Phenotype Driven Variant Ontological Re-Ranking Tool

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

Defining Citation: PMID:24702956

Keywords: genome interpretation, variant prioritization, disease gene prioritization, phenotype, gene function, disease, genomic, disease-causing allele, gene, function, allele

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: Phevor

Resource ID: SCR_002273

Alternate IDs: SciRes_000139

Record Creation Time: 20220129T080212+0000

Record Last Update: 20260821T042543+0000

Ratings and Alerts

No rating or validation information has been found for Phevor.

No alerts have been found for Phevor.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 9 mentions in open access literature.

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

Seaby EG, et al. (2021) Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. Frontiers in genetics, 12, 674295.

De La Vega FM, et al. (2021) Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. Genome medicine, 13(1), 153.

Sweeney NM, et al. (2021) Rapid whole genome sequencing impacts care and resource utilization in infants with congenital heart disease. NPJ genomic medicine, 6(1), 29.

James KN, et al. (2020) Partially automated whole-genome sequencing reanalysis of previously undiagnosed pediatric patients can efficiently yield new diagnoses. NPJ genomic medicine, 5, 33.

Pengelly RJ, et al. (2017) Evaluating phenotype-driven approaches for genetic diagnoses from exomes in a clinical setting. Scientific reports, 7(1), 13509.

Requena T, et al. (2017) A pipeline combining multiple strategies for prioritizing heterozygous variants for the identification of candidate genes in exome datasets. Human genomics, 11(1), 11.

Boudellioua I, et al. (2017) Semantic prioritization of novel causative genomic variants. PLoS computational biology, 13(4), e1005500.

Stelzer G, et al. (2016) VarElect: the phenotype-based variation prioritizer of the GeneCards Suite. BMC genomics, 17 Suppl 2(Suppl 2), 444.

Krøigård AB, et al. (2016) Bone structure in two adult subjects with impaired minor spliceosome function resulting from RNU4ATAC mutations causing microcephalic osteodysplastic primordial dwarfism type 1 (MOPD1). Bone, 92, 145.