Resource Summary Report

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PolyPhen: Polymorphism Phenotyping

RRID:SCR_013189
Type: Tool

Proper Citation

PolyPhen: Polymorphism Phenotyping (RRID:SCR_013189)

Resource Information

URL: http://genetics.bwh.harvard.edu/pph2/

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Description: Software tool which predicts possible impact of amino acid substitution on structure and function of human protein using straightforward physical and comparative considerations. PolyPhen-2 is new development of PolyPhen tool for annotating coding nonsynonymous SNPs.

Abbreviations: PolyPhen, PolyPhen-2, POLYPHEN

Synonyms: PolyPhen, POLYPHEN, PolyPhen-2, Polymorphism Phenotyping, Polymorphism Phenotyping v2

Resource Type: simulation software, data processing software, data analysis software, software resource, software application

Defining Citation: PMID:20354512, PMID:23315928

Keywords: annotate, nonsynonymous, SNP, predict, coding, damaging, effect, missense, mutation, sequence, variant, phenotype, genetic, disease, exon, protein, coding, fraction, genome, bio.tools

Resource Name: PolyPhen: Polymorphism Phenotyping

Resource ID: SCR_013189

Alternate IDs: SCR_013200, OMICS_00136, nlx_154540, nif-0000-21329, biotools:polyphen, SCR_013238

Alternate URLs: https://bio.tools/polyphen

Old URLs: http://www.bork.embl-heidelberg.de/PolyPhen/

Record Creation Time: 20220129T080314+0000

Record Last Update: 20260805T044309+0000

Ratings and Alerts

No rating or validation information has been found for PolyPhen: Polymorphism Phenotyping.

No alerts have been found for PolyPhen: Polymorphism Phenotyping.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 4151 mentions in open access literature.

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

Watanabe T, et al. (2026) Whole exome sequencing in Japanese spinocerebellar ataxia identifies novel variants. Journal of human genetics, 71(1), 35.

Ncir WB, et al. (2026) First LDLRAP1 and Recurrent LDLR Mutations in Tunisian Families With Familial Hypercholesterolemia. Journal of cellular and molecular medicine, 30(1), e70997.

Lucaciu SA, et al. (2025) Skin disease-associated GJB4 variants differentially influence connexin stability, cell viability and channel function. The Journal of physiology, 603(15), 4383.

Roush SM, et al. (2025) Shared genomic features of HIV+ diffuse large B-cell lymphoma in two African cohorts. Scientific reports, 15(1), 24599.

Sol Ruiz M, et al. (2025) Genetic characterization of pediatric B-cell acute lymphoblastic leukemia in Argentina uncovers molecular heterogeneity and novel variants. Frontiers in pharmacology, 16, 1701680.

Fiorica PN, et al. (2025) Germline Pathogenic DROSHA Variants Are Linked to Pineoblastoma and Wilms Tumor Predisposition. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(8), 1491.

Qin Z, et al. (2025) Structural and functional assessment of TBX20 gene variants in pediatric ventricular septal defect. Hereditas, 162(1), 149.

Diakité M, et al. (2025) Unusual catecholaminergic polymorphic ventricular tachycardia and bradycardia caused by a novel triadin variant in 2 siblings from a Malian family. Medicine, 104(31), e43596.

Van Haute L, et al. (2025) Pathogenic PDE12 variants impair mitochondrial RNA processing causing neonatal mitochondrial disease. EMBO molecular medicine, 17(1), 193.

Yang J, et al. (2025) Manic Fringe promotes endothelial-to-mesenchymal transition mediated by the Notch signalling pathway during heart valve development. Journal of molecular medicine (Berlin, Germany), 103(1), 51.

Zhou Q, et al. (2025) Molecular and Clinical Profiles of Pediatric Monogenic Diabetes Subtypes: Comprehensive Genetic Analysis of 138 Patients. The Journal of clinical endocrinology and metabolism, 110(8), 2314.

Bayam E, et al. (2025) Bi-allelic variants in WDR47 cause a complex neurodevelopmental syndrome. EMBO molecular medicine, 17(1), 129.

Smith TB, et al. (2025) Bi-allelic variants in DAP3 result in reduced assembly of the mitoribosomal small subunit with altered apoptosis and a Perrault-syndrome-spectrum phenotype. American journal of human genetics, 112(1), 59.

Iijima H, et al. (2025) A girl with intragenic variants in MARS2 and a chondrodysplasia phenotype. Molecular genetics and metabolism reports, 42, 101198.

Liu Z, et al. (2025) Novel pathogenic mtDNA variants in Chinese children with neurological mitochondrial disorders. Annals of clinical and translational neurology, 12(3), 586.

Naveed M, et al. (2025) Exploration of alcohol dehydrogenase EutG from Bacillus tropicus as an eco-friendly approach for the degradation of polycyclic aromatic compounds. Scientific reports, 15(1), 3466.

Yin D, et al. (2025) Evolution of canonical circadian clock genes underlies unique sleep strategies of marine mammals for secondary aquatic adaptation. PLoS genetics, 21(3), e1011598.

Naveed M, et al. (2025) Insight into molecular and mutational scrutiny of epilepsy associated gene Gabrg2 leading to novel computer-aided drug designing. Scientific reports, 15(1), 6770.

Pereira R, et al. (2025) The Spectrum of IDH- and H3-Wildtype High-Grade Glioma Subgroups Occurring across Teenage and Young Adult Patient Populations. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(15), 3259.

Kim JA, et al. (2025) Systematic genetic assessment of hearing loss using whole-genome sequencing identifies pathogenic variants. Experimental & molecular medicine, 57(4), 775.