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

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: 20260903T235210+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 4723 mentions in open access literature.

Listed below are recent publications. The full list is available at kravitz2 .

Hollis RL, et al. (2026) Molecular Profiling and Tumor Biomarker Analysis of GOG281/LOGS: A Positive Late-Phase Trial of Trametinib for Recurrent/Persistent Low-Grade Serous Ovarian Carcinoma. Clinical cancer research : an official journal of the American Association for Cancer Research, 32(4), 724.

Roush SM, et al. (2026) Mutational profiling of HIV+ diffuse large B-cell lymphoma reveals distinct mutational features with evidence of genomic instability. AIDS (London, England), 40(6), 719.

Jin M, et al. (2026) Genomic insights into the population history of fat-tailed sheep and identification of two mutations that contribute to fat tail adipogenesis. Journal of advanced research, 80, 725.

Huang M, et al. (2026) WFS1-related isolated diabetes induced by a WFS1 missense mutation: focus on the isolated diabetes phenotype. Orphanet journal of rare diseases, 21(1).

Gao Y, et al. (2026) Clinical and Functional Characterization of Novel GALNT3 Mutations in a Chinese Child with Hyperphosphatemic Familial Tumoral Calcinosis. International journal of molecular sciences, 27(6).

Fatima S, et al. (2026) A novel protein truncating mutation of TTC8 causes Bardet-Biedl Syndrome (BBS) in a Pakistani family. Genetics and molecular biology, 49(1), e20250020.

Song H, et al. (2026) Emergence of Polymyxin Resistance Driven by a PhoQ Mutation in KPC-2-Producing Klebsiella pneumoniae. Antibiotics (Basel, Switzerland), 15(2).

Sala E, et al. (2026) Intrafamilial phenotypic variability in hypophosphatasia: evidence from two families and the literature. Frontiers in endocrinology, 17, 1826307.

Wang Z, et al. (2026) Identification of a Novel De Novo Heterozygous SEC61A1 Variant in a Patient With Severe Congenital Neutropenia. Molecular genetics & genomic medicine, 14(5), e70217.

Werren EA, et al. (2026) Diagnostic utility of clinical genome reanalysis in rare pediatric disorders using long-read sequencing. HGG advances, 7(3), 100620.

Jenni R, et al. (2026) Novel genetic variants identification and immune profiling in ataxia telangiectasia patients. Journal of translational medicine, 24(1).

Bartylla MM, et al. (2026) Identification of novel PROS1 variants through systematic analysis of patients with suspected hereditary protein S deficiency. Research and practice in thrombosis and haemostasis, 10(3), 103432.

Vaghefinezhad N, et al. (2026) In vitro and In silico Analysis of SCIN rs376349889 as a Potential Biomarker for Gastric and Colorectal Cancers. Avicenna journal of medical biotechnology, 18(1), 79.

Zhu R, et al. (2026) Natural variation in Phosphatidylinositol 4-Kinase OsPI4K??7 and its interaction with OsLIC balance rice yield and latitudinal adaptation. Nature communications, 17(1).

Santacruz Reyes MD, et al. (2026) Isobutyryl-coenzyme a dehydrogenase deficiency: disease, or non-disease? Orphanet journal of rare diseases, 21(1), 35.

Wang M, et al. (2026) Exploration of the Pathogenic Mechanism of the Factor XIII A Subunit in a Patient With Congenital Factor XIII Deficiency. Molecular genetics & genomic medicine, 14(1), e70193.

Kikuchi S, et al. (2026) Compound heterozygous CHAT gene mutations, a missense and a splice site variant, in two siblings with congenital myasthenic syndrome. Scientific reports, 16(1).

Kashyap DK, et al. (2026) Novel MC1R variants cause red hair and lighter skin color. HGG advances, 7(3), 100603.

Waleed Iqbal M, et al. (2026) Delving Into the Depths of AGTR2: In Silico Identification of Deleterious Nonsynonymous SNPs Associated With Cardiovascular Diseases. Human mutation, 2026, 9394808.

Ren Y, et al. (2026) Identification and Functional Characterization of Novel and Recurrent NTRK1 Variants in Chinese Families With Congenital Insensitivity to Pain With Anhidrosis: A Combined Clinical, Genetic, and Functional Study. European journal of neurology, 33(5), e70610.