

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/>

Proper Citation: PolyPhen: Polymorphism Phenotyping (RRID:SCR_013189)

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: 20260821T193945+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 [NIF](#) .

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.