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 processing software, simulation 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

Funding:

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: 20250429T055602+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 4019 mentions in open access literature.

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

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.

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.

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

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

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.

Fan X, et al. (2025) Genotype-phenotype correlations for 17 Chinese families with inherited retinal dystrophies due to homozygous variants. Scientific reports, 15(1), 3043.

Anania M, et al. (2025) Identification of Four New Mutations in the GLA Gene Associated with Anderson-Fabry Disease. International journal of molecular sciences, 26(2).

Dong J, et al. (2025) A Case Study Identified a New Mutation in the TTN Gene for Inherited Hypertrophic Cardiomyopathy. International journal of general medicine, 18, 447.

Yamamoto S, et al. (2025) Congenital Hypogonadotropic Hypogonadism With Novel Pathogenic Variants in FGFR1 and GNRHR. JCEM case reports, 3(1), luae254.

Shen X, et al. (2025) The tomato gene Ty-6, encoding DNA polymerase delta subunit 1, confers broad resistance to Geminiviruses. TAG. Theoretical and applied genetics. Theoretische und angewandte Genetik, 138(1), 22.

Kesdiren E, et al. (2025) Heterozygous variants in the teashirt zinc finger homeobox 3 (TSHZ3) gene in human congenital anomalies of the kidney and urinary tract. European journal of human genetics : EJHG, 33(1), 44.

Kim JM, et al. (2025) Uncovering potential causal genes for undiagnosed congenital anomalies using an in-house pipeline for trio-based whole-genome sequencing. Human genomics, 19(1), 1.

El Houdi M, et al. (2025) Association study of the JAK/STAT signaling pathway with susceptibility to COVID-19 in moroccan patient and in-silico analysis of rare variants. Virus research, 351, 199509.

Zhang S, et al. (2025) Clinicopathological features of Lynch syndrome pedigrees with MSH2 c.351G>A gene variant. Molecular genetics & genomic medicine, 13(1), e2506.

Huang X, et al. (2025) Mutation spectra and genotype?phenotype analysis of congenital hypothyroidism in a neonatal population. Biomedical reports, 22(2), 30.

Kamal MM, et al. (2025) Investigating the functional and structural effect of non-synonymous single nucleotide polymorphisms in the cytotoxic T-lymphocyte antigen-4 gene: An in-silico study. PloS one, 20(1), e0316465.

Zhang L, et al. (2025) Preimplantation genetic testing for four families with severe combined immunodeficiency: Three unaffected livebirths. Orphanet journal of rare diseases, 20(1), 14.

Lu YL, et al. (2025) Identification of novel RIPK4 variants in a Chinese patient with Arthrogryposis Multiplex Congenita (AMC). Italian journal of pediatrics, 51(1), 6.

Yuan M, et al. (2025) Prevalence of IMPG1 and IMPG2 Mutations Leading to Retinitis Pigmentosa or Vitelliform Macular Dystrophy in a Cohort of Patients with Inherited Retinal Dystrophies. Genes, 16(1).

Meng L, et al. (2025) Heterozygous pathogenic STT3A variation leads to dominant congenital glycosylation disorders and functional validation in zebrafish. Orphanet journal of rare diseases, 20(1), 46.