

Resource Summary Report

Generated by [ASWG](#) on Aug 26, 2026

PolyPhred

RRID:SCR_002337

Type: Tool

Proper Citation

PolyPhred (RRID:SCR_002337)

Resource Information

URL: <http://droog.gs.washington.edu/polyphred/>

Proper Citation: PolyPhred (RRID:SCR_002337)

Description: Software program that compares fluorescence-based sequences across traces obtained from different individuals to identify heterozygous sites for single nucleotide substitutions. Its functions are integrated with the use of three other programs: Phred (Brent Ewing and Phil Green), Phrap (Phil Green), and Consed (David Gordon and Phil Green). PolyPhred identifies potential heterozygotes using the base calls and peak information provided by Phred and the sequence alignments provided by Phrap. Potential heterozygotes identified by PolyPhred are marked for rapid inspection using the Consed tool.

Abbreviations: PolyPhred

Resource Type: software resource

Defining Citation: [PMID:17115056](#), [PMID:16493422](#), [PMID:9207020](#)

Keywords: windows, sequence, nucleotide substitution, heterozygote, polymorphic, genotype, single nucleotide polymorphism, fluorescence, single nucleotide substitution, polymorphism, insertion, deletion, indel, bio.tools

Availability: Free for academic use, Commercial use requires a license

Resource Name: PolyPhred

Resource ID: SCR_002337

Alternate IDs: biotools:polyphred, OMICS_01815

Alternate URLs: <https://bio.tools/polyphred>

Record Creation Time: 20220129T080212+0000

Ratings and Alerts

No rating or validation information has been found for PolyPhred.

No alerts have been found for PolyPhred.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 127 mentions in open access literature.

Listed below are recent publications. The full list is available at [ASWG](#) .

Zhang S, et al. (2026) Single-cell polygenic risk scores dissect cellular and molecular heterogeneity of complex human diseases. *Nature biotechnology*, 44(5), 845.

Wang Y, et al. (2026) Association of SMAD4 gene polymorphisms with human papillomavirus infection and risk of cervical cancer: a case-control study. *BMC cancer*, 26(1), 213.

Da Fontoura CSG, et al. (2026) Exploring the role of TWIST1 in malocclusion and craniofacial morphology. *Frontiers in physiology*, 17, 1749243.

He Y, et al. (2026) Identification of two novel MVD mutations and one novel FDPS mutation in Chinese patients with porokeratosis. *Frontiers in medicine*, 13, 1760287.

Danbaki AS, et al. (2025) Pathogenic FANCC Variants Are Associated with Accessory Breasts in a Sub-Saharan African Multiplex Family. *Current issues in molecular biology*, 47(11).

Elshaer M, et al. (2025) Aberrant DNA methylation and overexpression of NR0B1 are prognostic biomarkers in KEAP1-mutant lung adenocarcinomas. *Discover oncology*, 16(1), 1641.

Zhu Y, et al. (2024) Two Novel and Two Recurrent Variants of the ADAR1 Gene in Three Chinese Families with Dyschromatosis Symmetrica Hereditaria. *Clinical, cosmetic and investigational dermatology*, 17, 2373.

Wang X, et al. (2024) Two Novel and Three Recurrent Mutations in the Mevalonate Pathway Genes in Chinese Patients with Porokeratosis. *Clinical, cosmetic and investigational dermatology*, 17, 191.

Hop PJ, et al. (2024) Systematic rare variant analyses identify RAB32 as a susceptibility gene for familial Parkinson's disease. *Nature genetics*, 56(7), 1371.

Li S, et al. (2024) A monoallelic variant in CCN2 causes an autosomal dominant spondyloepimetaphyseal dysplasia with low bone mass. *Bone research*, 12(1), 60.

Mei Y, et al. (2024) Echocardiographic abnormalities and joint hypermobility in Chinese patients with Osteogenesis imperfecta. *Orphanet journal of rare diseases*, 19(1), 116.

Ren N, et al. (2024) Clinical features, treatment, and follow-up of OPPG and high-bone-mass disorders: LRP5 is a key regulator of bone mass. *Osteoporosis international : a journal established as result of cooperation between the European Foundation for Osteoporosis and the National Osteoporosis Foundation of the USA*, 35(8), 1395.

Wang PY, et al. (2023) Distilling functional variations for human UGT2B4 upstream region based on selection signals and implications for phenotypes of Neanderthal and Denisovan. *Scientific reports*, 13(1), 3134.

Ye M, et al. (2023) New Endothelial Corneal Dystrophy in a Chinese Family. *Cornea*, 42(5), 529.

He J, et al. (2023) Case report: A novel mutation in the EYA1 gene in a child with branchiootic syndrome with secretory otitis media and bilateral vestibular hypofunction. *Frontiers in genetics*, 14, 1292085.

Tao XH, et al. (2022) Clinical characteristics and identification of a novel TGFB1 variant in three unrelated Chinese families with Camurati-Engelmann disease. *Molecular genetics & genomic medicine*, 10(5), e1922.

Zhang W, et al. (2022) Identification of a novel CNV at the EYA4 gene in a Chinese family with autosomal dominant nonsyndromic hearing loss. *BMC medical genomics*, 15(1), 113.

Wu J, et al. (2022) Mutations in exon region of BRCA1-related RING domain 1 gene and risk of breast cancer. *Molecular genetics & genomic medicine*, 10(3), e1847.

Wang L, et al. (2021) Analysis of genetic variation in human papillomavirus type 16 E1 and E2 in women with cervical infection in Xinjiang, China. *BMC medical genomics*, 14(1), 268.

Wu J, et al. (2021) Correlation between ZBRK1/ZNF350 gene polymorphism and breast cancer. *BMC medical genomics*, 14(1), 7.