

Resource Summary Report

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ANNOVAR

RRID:SCR_012821

Type: Tool

Proper Citation

ANNOVAR (RRID:SCR_012821)

Resource Information

URL: <http://www.openbioinformatics.org/annovar/>

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Description: An efficient software tool to utilize update-to-date information to functionally annotate genetic variants detected from diverse genomes (including human genome hg18, hg19, as well as mouse, worm, fly, yeast and many others). Given a list of variants with chromosome, start position, end position, reference nucleotide and observed nucleotides, ANNOVAR can perform: 1. gene-based annotation. 2. region-based annotation. 3. filter-based annotation. 4. other functionalities. (entry from Genetic Analysis Software)

Abbreviations: ANNOVAR

Synonyms: functional ANNOtation of genetic VARiants, ANNOVAR: Functional annotation of genetic variants

Resource Type: software application, software resource

Defining Citation: [PMID:20601685](#)

Keywords: genomic analysis, imaging genomics, next generation sequencing, snp, gene, bio.tools

Availability: Free

Resource Name: ANNOVAR

Resource ID: SCR_012821

Alternate IDs: nlx_154225, biotools:annovar, OMICS_00165

Alternate URLs: <https://bio.tools/annovar>, <https://bio.tools/annovar>

Record Creation Time: 20220129T080312+0000

Ratings and Alerts

No rating or validation information has been found for ANNOVAR.

No alerts have been found for ANNOVAR.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 5946 mentions in open access literature.

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

Chang LJ, et al. (2026) Assessment of Circulating Tumor DNA for Early Detection of Hepatocellular Carcinoma in Alpha-Fetoprotein-Negative Patients With Cirrhotic Nodules. JGH open : an open access journal of gastroenterology and hepatology, 10(1), e70331.

Huang X, et al. (2026) PMEL governs autosomal dominant inheritance of white-tail independent of yellow body plumage in chickens (Gallus gallus domesticus). Poultry science, 105(1), 106127.

Upadhyayula BS, et al. (2026) Mutational Spectrum of T-Cell Large Granular Lymphocytic Leukemia: Insights From the AACR Project GENIE Consortium. Cancer genomics & proteomics, 23(1), 135.

Eising E, et al. (2026) De novo protein-coding gene variants in developmental stuttering. Molecular psychiatry, 31(1), 104.

Yuan J, et al. (2026) Genome-wide association study reveals genetic architecture and evolution of human retinal pigmentation. Science advances, 12(1), eadw7768.

Bonamici L, et al. (2026) Characterization of Copy Number Variants in Hereditary Cancer Patients Through NGS Shows a Distinctive PALB2 Contribution to the Diagnostic Yield. Human mutation, 2026, 6601291.

Yu Z, et al. (2026) Multi-trait genome-wide analysis identified risk loci and candidate drugs for heart failure. HGG advances, 7(1), 100540.

Wong NKY, et al. (2026) Establishment and characterization of preclinical models of human gynecologic tract carcinosarcomas demonstrates targetable FGFR1 alterations. Translational oncology, 63, 102591.

Teng CS, et al. (2026) Actomyosin contractility and a threshold of cadherin cell adhesion are required during tissue fusion. The Journal of cell biology, 225(1).

Zhang X, et al. (2026) Genetic and Functional Evidence Links Germline Biallelic Inactivating Variants in WWOX to Histological Mixed-Type Thyroid Cancer. *Advanced science* (Weinheim, Baden-Wurttemberg, Germany), 13(1), e07602.

Xiangwen W, et al. (2026) A De Novo Splicing Mutation of SRP72 in Bone Marrow Failure Syndrome Type 1: Case Report and Review of the Literature. *Molecular genetics & genomic medicine*, 14(1), e70168.

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

Gu Y, et al. (2026) Multi-omics analyses identify potential epigenetic loci associated with survival in amyotrophic lateral sclerosis across diverse populations. *EBioMedicine*, 123, 106071.

Rychlicka-Buniowska E, et al. (2026) Loss of Y chromosome in Alzheimer's patients co-occurs with somatic mutations beyond CHIP drivers. *Life science alliance*, 9(2).

Lan T, et al. (2025) Enhancing inbreeding estimation and global conservation insights through chromosome-level assemblies of the Chinese and Malayan pangolin. *GigaScience*, 14.

Li Z, et al. (2025) Metabolite-dependent m6A methylation driven by mechanotransduction-metabolism-epitranscriptomics axis promotes bone development and regeneration. *Cell reports*, 44(5), 115611.

Wang Y, et al. (2025) Telomere-to-telomere genome of common bean (*Phaseolus vulgaris* L., YP4). *GigaScience*, 14.

Liu Z, et al. (2025) ctDNA detects residual disease after neoadjuvant chemoradiotherapy and guides adjuvant therapy in esophageal squamous cell carcinoma. *Cell reports. Medicine*, 6(9), 102334.

Andersson N, et al. (2025) Tracing metastatic spread in pediatric solid tumors using copy number and targeted deep sequencing. *The Journal of pathology*.

Smith JD, et al. (2025) Genomic Signatures of Poor Prognosis in Merkel Cell Carcinoma: A Single-Institution Prospective Study. *Molecular cancer research : MCR*.