Resource Summary Report

Generated by NIF on Sep 2, 2026

ClinVar

RRID:SCR_006169
Type: Tool

Proper Citation

ClinVar (RRID:SCR_006169)

Resource Information

URL: http://www.ncbi.nlm.nih.gov/clinvar/

Proper Citation: ClinVar (RRID:SCR_006169)

Description: Archive of aggregated information about sequence variation and its relationship to human health. Provides reports of relationships among human variations and phenotypes along with supporting evidence. Submissions from clinical testing labs, research labs, locus-specific databases, expert panels and professional societies are welcome. Collects reports of variants found in patient samples, assertions made regarding their clinical significance, information about submitter, and other supporting data. Alleles described in submissions are mapped to reference sequences, and reported according to HGVS standard.

Abbreviations: ClinVar

Resource Type: data or information resource, data repository, database, service resource, storage service resource

Keywords: sequence variation, variation, phenotype, genetics, genetic variation, clinical, allele, aggregator, geneotype, gene, disease, clinical assertion, bio.tools

Availability: Free, Freely available

Resource Name: ClinVar

Resource ID: SCR_006169

Alternate IDs: nlx_151671, r3d100013331, biotools:clinvar, OMICS_00262

Alternate URLs: https://bio.tools/clinvar, https://doi.org/10.17616/R31NJMS3

Record Creation Time: 20220129T080234+0000

Record Last Update: 20260829T182749+0000

Ratings and Alerts

No rating or validation information has been found for ClinVar.

No alerts have been found for ClinVar.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 7407 mentions in open access literature.

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

Kojima M, et al. (2026) Precision Oncology for Pediatric Solid Tumors Using In-Hospital Pediatric/AYA Malignancy-Specific Panel Sequencing. Cancer science, 117(3), 797.

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.

Gatz SA, et al. (2026) Phase I/II Study of the PARP Inhibitor Olaparib and Irinotecan in Children and Young Adults with Recurrent/Refractory Malignancies: Arm D of the AcSé-ESMART Trial. Clinical cancer research : an official journal of the American Association for Cancer Research, 32(7), 1210.

Lee S, et al. (2026) Functional characterization of SDHB variants clarifies hereditary pheochromocytoma and paraganglioma risk and genotype-phenotype relationships. The Journal of clinical investigation, 136(4).

Monti M, et al. (2026) Integrating interferon gamma receptor pathways, antigenicity, and immune contexture as predictors of immunotherapeutic strategies for mucosal melanomas. Journal for immunotherapy of cancer, 14(3).

Pehrsson M, et al. (2026) Opportunistic Screening of High-Risk Breast Cancer Variants in Hospital Biobank Setting. Cancer epidemiology, biomarkers & prevention : a publication of the American Association for Cancer Research, cosponsored by the American Society of Preventive Oncology, 35(6), 883.

Botticelli A, et al. (2026) The Impact of Concordance between Liquid and Tissue Biopsy for Actionable Mutations: Insights from the ROME Trial. Clinical cancer research : an official journal of the American Association for Cancer Research, 32(1), 45.

Fuentes-Bolanos NA, et al. (2026) Integrated Germline and Somatic Molecular Profiling to Detect Cancer Predisposition Has a High Clinical Impact in Poor-Prognosis Pediatric Cancer. Clinical cancer research : an official journal of the American Association for Cancer Research, 32(10), 2047.

Tran U, et al. (2026) BICC1 interacts with PKD1 and PKD2 to drive cystogenesis in ADPKD. eLife, 14.

Howard CJ, et al. (2026) Deep mutational scanning reveals pharmacologically relevant insights into TYK2 signaling and disease. eLife, 15.

Robinson E, et al. (2026) STK11 mutations and deletions define a distinct subtype of cervical adenocarcinoma. medRxiv : the preprint server for health sciences.

Hanff AM, et al. (2026) Progression of GBA1 severe and risk variants: a longitudinal mixed model analysis. Frontiers in aging neuroscience, 18, 1771789.

Yun KH, et al. (2026) Phase IB/II Trial with Correlative Analyses of Doxorubicin plus Durvalumab Combination in Patients with Advanced Soft Tissue Sarcoma. Clinical cancer research : an official journal of the American Association for Cancer Research, 32(9), 1686.

Mishra S, et al. (2026) Decoding Genetic Risk Factors in Recurrent Pregnancy Loss: An Integrative Chromosomal and Bioinformatics Approach. Biochemical genetics.

Luo S, et al. (2026) Recessive variants in WSB2 encoding a substrate receptor of E3 ubiquitin ligase underlie a neurodevelopmental syndrome. European journal of human genetics : EJHG, 34(1), 37.

Song S, et al. (2026) Clostridium butyricum and its metabolites regulate macrophage polarization through miR-146a to antagonize gouty arthritis. Journal of advanced research, 80, 943.

Skálová A, et al. (2026) Basal cell adenoma with S100 protein-positive "stroma": a distinct triphasic salivary gland neoplasm characterized by CTNNB1 mutation. Virchows Archiv : an international journal of pathology, 488(2), 387.

Guo W, et al. (2026) Pathogenicity of novel GLA gene missense mutations in Fabry disease and the therapeutic impact of migalastat. Journal of advanced research, 79, 549.

Wu H, et al. (2026) Treatment with Minicircle DNA Expressing a FGF23 Fragment in a Clinically relevant Mouse Model of X-Linked Hypophosphatemic Rickets. Advanced science (Weinheim, Baden-Wurttemberg, Germany), 13(4), e08870.

Dahir KM, et al. (2026) Hypophosphatasia: low penetrance of pathogenic and likely-pathogenic ALPL variants identified through an unselected biorepository. Journal of bone and mineral research : the official journal of the American Society for Bone and Mineral Research, 41(3), 270.