Resource Summary Report

Generated by RRID on Aug 5, 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: storage service resource, data repository, service resource, database, data or information 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: 20260805T044131+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 6595 mentions in open access literature.

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

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.

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.

Quarello P, et al. (2026) DNA Methylation Episignature as a Novel Diagnostic Tool for Diamond-Blackfan Anemia Syndrome. American journal of hematology, 101(2), 228.

Gao Y, et al. (2026) The intricate dance of RNA-binding proteins: unveiling the mechanisms behind male infertility. Human reproduction update, 32(1), 58.

Habano E, et al. (2026) Germline pathogenic variants detected by GenMineTOP: insight from a nationwide tumor/normal paired comprehensive genomic profiling test, in Japan. Journal of human genetics, 71(1), 1.

Simonati A, et al. (2026) Age at onset and gene variants predict lifespan and disease duration in childhood neuronal ceroid lipofuscinoses. Developmental medicine and child neurology, 68(2), 276.

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.

Ncir WB, et al. (2026) First LDLRAP1 and Recurrent LDLR Mutations in Tunisian Families With Familial Hypercholesterolemia. Journal of cellular and molecular medicine, 30(1), e70997.

Kim CY, et al. (2026) Integrative multi-omics of matched primary and liver metastatic colorectal cancer organoids identifies putative molecular targets. Translational oncology, 63, 102592.

Lucaciu SA, et al. (2025) Skin disease-associated GJB4 variants differentially influence connexin stability, cell viability and channel function. The Journal of physiology, 603(15), 4383.

Goshayeshi L, et al. (2025) Germline variants in patients from the Iranian hereditary colorectal cancer registry. Cancer cell international, 25(1), 140.

Kurian AW, et al. (2025) ClinVar Database Evolution and Impact on Potential Pathogenic Germline Variant Reporting from Tumor Comprehensive Genomic Profiling. Cancer research communications, 5(8), 1282.

Usui Y, et al. (2025) Clinical Significance of TP53-Mutant Clonal Hematopoiesis Across Diseases. Blood cancer discovery, 6(4), 298.

Treger TD, et al. (2025) Predisposition Footprints in the Somatic Genome of Wilms Tumors. Cancer discovery, 15(2), 286.

Dev H, et al. (2025) Cambridge Neoadjuvant Cancer of the Prostate (CANCAP03): A Window Study into the Effects of Olaparib ?? Degarelix in Primary Prostate Cancer. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(12), 2347.

Sol Ruiz M, et al. (2025) Genetic characterization of pediatric B-cell acute lymphoblastic leukemia in Argentina uncovers molecular heterogeneity and novel variants. Frontiers in pharmacology, 16, 1701680.

van Ravensteijn SG, et al. (2025) Cemiplimab in Locally Advanced or Metastatic Secondary Angiosarcomas (CEMangio): A Phase II Clinical Trial and Biomarker Analyses. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(17), 3678.

Gu J, et al. (2025) Mapping multimodal phenotypes to perturbations in cells and tissue with CRISPRmap. Nature biotechnology, 43(7), 1101.

Hsiao YJ, et al. (2025) Tp53 determines the spatial dynamics of M1/M2 tumor-associated macrophages and M1-driven tumoricidal effects. Cell death & disease, 16(1), 38.

Hanenberg H, et al. (2025) Reclassification of ATM Missense Variants of Uncertain Significance by Integrating Results from Systematic Functional Assays into an ACMG Points-Based Framework. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(12), 2426.