

Resource Summary Report

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GEMINI

RRID:SCR_014819

Type: Tool

Proper Citation

GEMINI (RRID:SCR_014819)

Resource Information

URL: <https://gemini.readthedocs.io/en/latest/>

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Description: Framework for exploring genetic variation in the context of the genome annotations available for the human genome. Users can load a VCF file into a database and each variant is automatically annotated by comparing it to several genome annotations from source such as ENCODE tracks, UCSC tracks, OMIM, dbSNP, KEGG, and HPRD.

Synonyms: GEnome MINIng (GEMINI), GEMINI - a flexible framework for exploring genome variation, Genome Mining, GEnome MINIng

Resource Type: software resource

Defining Citation: [DOI:10.1371/journal.pcbi.1003153](https://doi.org/10.1371/journal.pcbi.1003153)

Keywords: framework, genetic variation, annotation, human, genome, vcf, database, , bio.tools, FASEB list

Availability: Freely available

Resource Name: GEMINI

Resource ID: SCR_014819

Alternate IDs: biotools:gemini

Alternate URLs: <https://github.com/arq5x/gemini>, <https://bio.tools/gemini>

License: MIT license

Record Creation Time: 20220129T080322+0000

Record Last Update: 20260905T132751+0000

Ratings and Alerts

No rating or validation information has been found for GEMINI.

No alerts have been found for GEMINI.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 532 mentions in open access literature.

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

Mounier N, et al. (2026) Genetics identifies obesity as a shared risk factor for co-occurring multiple long-term conditions. *Communications medicine*, 6(1), 67.

Maaser-Hecker AK, et al. (2026) RIN3 mutations impairing binding of the Alzheimer's disease-associated protein BIN1 lead to RAB5 hyperactivation and endosomal pathology. *Science advances*, 12(5), eadx2127.

Massimi D, et al. (2026) Large Language Model-Driven Analysis and Report Generation of Endoscopy Videos-A Pilot Study. *Digestive endoscopy : official journal of the Japan Gastroenterological Endoscopy Society*, 38(3), e70134.

Miller SJ, et al. (2026) Intracellular protein GBF1 displays significant associations with amyloid pathology in Alzheimer's disease. *Alzheimer's & dementia : the journal of the Alzheimer's Association*, 22(4), e71271.

Salvo MA, et al. (2026) Alzheimer's disease-linked ACE variants increase ACE1 catalytic activity and production of angiotensin II. *The Journal of biological chemistry*, 302(3), 111171.

Iyengar AK, et al. (2026) Cell modeling and rescue of a novel noncoding genetic cause of glycogen storage disease IX. *Genetics in medicine open*, 4, 103474.

Yang K, et al. (2026) Evaluating the clinical utility of large language models for hepatocellular carcinoma treatment recommendations: A nationwide retrospective registry study. *PLoS medicine*, 23(1), e1004855.

Russ P, et al. (2026) Potential of large language models for rapid clinical information support: evidence from acute kidney injury knowledge testing. *Scientific reports*, 16(1).

Jung K, et al. (2026) Evaluation of the Performance of Advanced Large Language Models in Laboratory Medicine Using Residency Examinations. *Annals of laboratory medicine*, 46(3), 327.

Tomasini D, et al. (2025) Genetic and Phenotypic Features of 2 Northern Italy Families with Dowling-Degos Disease Type 4. JID innovations : skin science from molecules to population health, 5(4), 100364.

Bai AD, et al. (2025) Oseltamivir Treatment vs Supportive Care for Seasonal Influenza Requiring Hospitalization. JAMA network open, 8(6), e2514508.

Fielden J, et al. (2025) Comprehensive interrogation of synthetic lethality in the DNA damage response. Nature, 640(8060), 1093.

Delsoz M, et al. (2025) Large Language Models: Pioneering New Educational Frontiers in Childhood Myopia. Ophthalmology and therapy, 14(6), 1281.

LaFramboise WA, et al. (2025) Cell-Free DNA, Tumor Molecular Concordance, and Clinical Correlates of Patients with Cancer Treated in a Large Community Health Care Network. The Journal of molecular diagnostics : JMD, 27(9), 882.

Medalcho TH, et al. (2025) Effects of spices mixture and cooking on antioxidant activity in Ethiopian spicy hot red pepper powder. Scientific reports, 15(1), 5203.

Stefanovi?? P, et al. (2025) The influence of Gen-AI tools application for text data augmentation: case of Lithuanian educational context data classification. Scientific reports, 15(1), 26010.

Achuthan K, et al. (2025) The role of music in ADHD: A multi-dimensional computational and theoretical analysis. PloS one, 20(8), e0324369.

Sozen Yanik I, et al. (2025) Cross-lingual performance of large language models in maxillofacial prosthodontics: a comparative evaluation. BMC oral health, 25(1), 1630.

Barnet MB, et al. (2025) Common inherited loss-of-function mutations in the innate sensor NOD2 contribute to exceptional immune response to cancer immunotherapy. Proceedings of the National Academy of Sciences of the United States of America, 122(28), e2314258122.

Subasri V, et al. (2025) Detecting and Remediating Harmful Data Shifts for the Responsible Deployment of Clinical AI Models. JAMA network open, 8(6), e2513685.