

# Resource Summary Report

Generated by SPARC SAWG on Sep 17, 2026

## GEMINI

RRID:SCR\_014819

Type: Tool

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### Proper Citation

GEMINI (RRID:SCR\_014819)

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### 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:** 20260912T195822+0000

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## Ratings and Alerts

No rating or validation information has been found for GEMINI.

No alerts have been found for GEMINI.

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## Data and Source Information

**Source:** [SciCrunch Registry](#)

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## Usage and Citation Metrics

We found 532 mentions in open access literature.

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

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.

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.

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.

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

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.

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.

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.

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

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

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

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.

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.

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.

Neubauer JC, et al. (2025) Performance of large language models ChatGPT and Gemini in child and adolescent psychiatry knowledge assessment. *PloS one*, 20(9), e0332917.