

# Resource Summary Report

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## [ClinGen](#)

RRID:SCR\_014968

Type: Tool

### Proper Citation

ClinGen (RRID:SCR\_014968)

### Resource Information

**URL:** <https://www.clinicalgenome.org>

**Proper Citation:** ClinGen (RRID:SCR\_014968)

**Description:** Genomics knowledgebase for clinical relevance of genes and variants for use in research. ClinGen's primary function is to store and share information for the benefit of the scientific community. Laboratory scientists, clinicians, and patients can share and access data.

**Abbreviations:** CGR

**Synonyms:** Clinical Genome Resource (ClinGen), Clinical Genome Resource

**Resource Type:** database, data or information resource

**Defining Citation:** [PMID:26014595](#)

**Keywords:** database, knowledgebase, genomics, healthcare, clinical, FASEB list

**Funding:** Eunice Kennedy Schriver NICHD ;  
NHGRI U41 HG006834-01A1;  
NHGRI U01 HG007437-01;  
NHGRI U01 HG007436-01;  
NCI HHSN261200800001E;  
NCI contract HHSN261200800001E

**Availability:** Free, Available to the scientific community

**Resource Name:** ClinGen

**Resource ID:** SCR\_014968

**Record Creation Time:** 20220129T080323+0000

**Record Last Update:** 20260805T044332+0000

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## Ratings and Alerts

No rating or validation information has been found for ClinGen.

No alerts have been found for ClinGen.

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## Data and Source Information

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

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## Usage and Citation Metrics

We found 898 mentions in open access literature.

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

Park KS, et al. (2026) Applying National Whole-genome Sequencing Findings for Rare Diseases in Clinical Practice: The Imperative of a Multidisciplinary Approach. *Annals of laboratory medicine*, 46(1), 94.

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.

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.

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.

Minami S, et al. (2025) Auditory neuropathy spectrum disorder and related auditory features in patients with hearing loss associated with the MT-TS1 m.7471dup variant. *Mitochondrion*, 84, 102056.

Lim C, et al. (2025) Clinical Implementation of Nephrologist-Led Genomic Testing for Glomerular Diseases in Singapore: Rationale and Protocol. *American journal of nephrology*, 56(2), 158.

Pezeshkpoor B, et al. (2025) Modulation of Haemostatic Balance in Combined von Willebrand Disease and Antithrombin Deficiency: A Comprehensive Family Study. *Haemophilia : the official journal of the World Federation of Hemophilia*, 31(1), 140.

Furukawa S, et al. (2025) Whole-genome sequencing analysis of Japanese autism spectrum disorder trios. *Psychiatry and clinical neurosciences*, 79(3), 87.

Arai Y, et al. (2025) Novel OTOG Variants and Clinical Features of Hearing Loss in a Large Japanese Cohort. *Genes*, 16(1).

- Yang S, et al. (2025) The evolution of cell-free fetal DNA testing: expanded non-invasive prenatal testing and its effect on target populations. *Frontiers in medicine*, 12, 1522680.
- Ma M, et al. (2025) A Novel Compound Heterozygous Variant in the ABHD12 Gene Cause PHARC Syndrome in a Chinese Family: The Proband Presenting New Genotype and Phenotype. *Molecular genetics & genomic medicine*, 13(2), e70055.
- Tsiortou P, et al. (2025) Immunogenetic Studies in Patients With GAD-Positive Stiff-Person Syndrome Reveal Novel Lymphocytic Genes and KLK10-Gene Variants. *Neurology(R) neuroimmunology & neuroinflammation*, 12(2), e200373.
- Lam WKJ, et al. (2025) The implementation of genome sequencing in rare genetic diseases diagnosis: a pilot study from the Hong Kong genome project. *The Lancet regional health. Western Pacific*, 55, 101473.
- Mori GH, et al. (2025) Pathogenicity of germline VHL variants is associated with renal cell carcinoma size in von Hippel-Lindau disease. *Archives of endocrinology and metabolism*, 69(1), e240354.
- Hahn E, et al. (2025) Copy number variant analysis improves diagnostic yield in a diverse pediatric exome sequencing cohort. *NPJ genomic medicine*, 10(1), 16.
- Xu H, et al. (2025) Landscape of human protein-coding somatic mutations across tissues and individuals. *bioRxiv : the preprint server for biology*.
- Henkel J, et al. (2025) Joint analysis of germline genetic data from over 29,000 cases with suspected hereditary breast and ovarian cancer (HBOC) as part of the NASGE initiative. *Breast (Edinburgh, Scotland)*, 80, 103887.
- Schei-Andersen AJ, et al. (2025) Non-serous ovarian cancer in PTEN Hamartoma Tumor Syndrome: additional evidence for increased risk. *Familial cancer*, 24(2), 28.
- Alay MT, et al. (2025) A novel seven-tier framework for the classification of MEFV missense variants using adaptive and rigid classifiers. *Scientific reports*, 15(1), 9054.
- Zhao W, et al. (2025) GoFCards: an integrated database and analytic platform for gain of function variants in humans. *Nucleic acids research*, 53(D1), D976.