

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: data or information resource, database

Defining Citation: [PMID:26014595](#)

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

Funding: Eunice Kennedy Schriver NICHD ;
NHGRI U41 HG006834-01A1;
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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: 20260811T043516+0000

Ratings and Alerts

No rating or validation information has been found for ClinGen.

No alerts have been found for ClinGen.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 1153 mentions in open access literature.

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

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).

Llinares-Burguet I, et al. (2026) Generation of functional noncanonical donor splice sites by +2T variants in breast cancer susceptibility genes: impact on clinical interpretation. The Journal of pathology, 268(2), 150.

Mustafa HJ, et al. (2026) Yield of Whole Genome Sequencing for Pathogenic Single Nucleotide Variants in Congenital Heart Disease: A Systematic Review and Meta-Analysis. Prenatal diagnosis, 46(5-6), 780.

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.

Theobald K, et al. (2026) Expanding Access to Genome Sequencing: Higher Diagnostic Yield in Self-Referred Participants From the CincyKidsSeq Study and Implications for Hybrid Models of Genetic Service Delivery. Clinical genetics, 109(4), 717.

Yu K, et al. (2026) Aggregation of gene regulatory information and knowledge on FAIR principles enables discovery of pathogenic gene regulatory variants. Bioinformatics (Oxford, England), 42(3).

Zhuang J, et al. (2026) Molecular diagnosis of rare biallelic CDC45 gene variants causing Meier-Gorlin syndrome-7 using whole exome sequencing. Human genomics, 20(1).

Quinodoz M, et al. (2026) De novo and inherited dominant variants in U4 and U6 snRNA genes cause retinitis pigmentosa. Nature genetics, 58(1), 169.

Br??nger T, et al. (2026) Gene Portals: A Framework for Integrating Clinical, Functional, and Structural Evidence into Rare Disease Variant Classification. medRxiv : the preprint server for health sciences.

Yehia L, et al. (2026) Genomic modifiers of malignant and neurodevelopmental phenotypes in individuals with PTEN hamartoma tumor syndrome. NPJ genomic medicine, 11(1).

Liang Y, et al. (2026) Residual risk of clinically significant copy number variations in fetuses with ultrasonographic soft markers following exclusion of non-invasive prenatal screening-detectable findings. Archives of gynecology and obstetrics, 313(1).

DeLuca M, et al. (2026) Medicines, Diseases, Indications, and Contraindications (MeDIC): a foundational resource to support drug repurposing. Nucleic acids research, 54(D1), D1477.

van Rensburg I, et al. (2026) Expanding the RB1 variant landscape of heritable retinoblastoma: unlocking precision oncology potential in Southern Africa. BMC cancer, 26(1).

Schwartz S, et al. (2026) Prenatal Microarray Analysis of Pregnancies Without Ultrasound Anomalies: Establishment of Copy Number and Homozygosity Frequencies in Low-Risk Population. Genes, 17(2).

La Monica I, et al. (2026) Clinical Insights into the Neurodevelopmental Impact of 16p CNVs in an Italian Clinical Cohort. Genes, 17(2).

Kim J, et al. (2026) Leveraging Quantitative Proteomics and Extracellular Vesicle Data to Uncover Druggable Receptor Kinases Across Cancers. Journal of extracellular vesicles, 15(4), e70275.

Liu J, et al. (2026) Whole-exome sequencing increases variant detection compared to karyotyping and CMA in an unselected FGR cohort. Scientific reports, 16(1).

Ganoza CA, et al. (2026) Pathogenic or Likely Pathogenic GRN Variants Are Found in 0.1% of Parkinson's Disease Patients. Movement disorders : official journal of the Movement Disorder Society, 41(4), 1020.

Bedei I, et al. (2026) Uncovering the Genetic Landscape of Spinal Dysraphism: A Retrospective Analysis of 150 Fetal Cases. Prenatal diagnosis, 46(5-6), 849.

Zhao Q, et al. (2026) Investigating the contribution of rare non-coding variants in BRCA1, BRCA2 and PALB2 to hereditary breast cancer. NPJ breast cancer, 12(1).