Resource Summary Report

Generated by dkNET on Sep 9, 2026

ClinGen

RRID:SCR_014968
Type: Tool

Proper Citation

ClinGen (RRID:SCR_014968)

Resource Information

URL: https://www.clinicalgenome.org

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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: 20260905T133211+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 dkNET .

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).

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.

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.

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

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.

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.

Wang B, et al. (2026) Genetic testing and analysis of 1024 children with global developmental delay or intellectual disability: a single-center cohort study. European journal of pediatrics, 185(7).

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.

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).

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.

Lezirovitz K, et al. (2026) A Structural Variant in the 5' Regulatory Region of DIAPH3 Segregates with Postlingual Hearing Loss and Auditory Neuropathy in a Multigenerational Brazilian Family. International archives of otorhinolaryngology, 30(2), 1.

Brenner D, et al. (2026) A rare missense variant impacting NEK1 kinase function is associated with ALS. Acta neuropathologica communications, 14(1).

Geng J, et al. (2026) Optimising POU3F4 variant interpretation through gene-specific evidence in X-linked hearing loss. EBioMedicine, 128, 106318.

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).

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).

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.