Resource Summary Report

Generated by dkNET on May 16, 2025

Clinical Genomic Database

RRID:SCR_006427
Type: Tool

Proper Citation

Clinical Genomic Database (RRID:SCR_006427)

Resource Information

URL: http://research.nhgri.nih.gov/CGD/

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Description: Manually curated database of all conditions with known genetic causes, focusing on medically significant genetic data with available interventions. Includes gene symbol, conditions, allelic conditions, inheritance, age in which interventions are indicated, clinical categorization, and general description of interventions/rationale. Contents are intended to describe types of interventions that might be considered. Includes only single gene alterations and does not include genetic associations or susceptibility factors related to more complex diseases.

Abbreviations: CGD

Synonyms: Clinical Genomics Database

Resource Type: data or information resource, database

Defining Citation: PMID:23696674

Keywords: genomic sequencing, genome, clinical, pediatric, adult human, young human, genomic medicine, whole-genome sequencing, gene, organ system, intervention, gene symbol, condition, allelic condition, clinical categorization, manifestation, inheritance, age group, genetic variant, pathogenic mutation

Funding: NHGRI

Availability: Free, Freely available

Resource Name: Clinical Genomic Database

Resource ID: SCR_006427

Alternate IDs: nlx_152872

Record Creation Time: 20220129T080236+0000

Record Last Update: 20250507T060418+0000

Ratings and Alerts

No rating or validation information has been found for Clinical Genomic Database.

No alerts have been found for Clinical Genomic Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 9 mentions in open access literature.

Listed below are recent publications. The full list is available at dkNET.

Trost B, et al. (2018) A Comprehensive Workflow for Read Depth-Based Identification of Copy-Number Variation from Whole-Genome Sequence Data. American journal of human genetics, 102(1), 142.

Makrythanasis P, et al. (2016) Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American journal of human genetics, 98(4), 615.

Bornstein AT, et al. (2016) Tracking medical genetic literature through machine learning. Molecular genetics and metabolism, 118(4), 255.

Ruderfer DM, et al. (2016) Patterns of genic intolerance of rare copy number variation in 59,898 human exomes. Nature genetics, 48(10), 1107.

Kim JS, et al. (2016) Cardiovascular autonomic dysfunctions in elderly patients with essential tremor: comparison with healthy controls. Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 37(5), 711.

Kim JH, et al. (2016) De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American journal of human genetics, 99(3), 711.

Solomon BD, et al. (2016) A 2.5-year snapshot of Mendelian discovery. Molecular genetics & genomic medicine, 4(4), 392.

Johnston JJ, et al. (2015) Individualized iterative phenotyping for genome-wide analysis of loss-of-function mutations. American journal of human genetics, 96(6), 913.

Bi C, et al. (2013) Vitamin D receptor, an important transcription factor associated with aldosterone-producing adenoma. PloS one, 8(12), e82309.