

Resource Summary Report

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Combined Annotation Dependent Depletion

RRID:SCR_018393

Type: Tool

Proper Citation

Combined Annotation Dependent Depletion (RRID:SCR_018393)

Resource Information

URL: <https://cadd.gs.washington.edu/>

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Description: Web tool for predicting deleteriousness of variants throughout human genome. Software tool for scoring deleteriousness of single nucleotide variants as well as insertion and deletions variants in human genome.

Abbreviations: CADD

Synonyms: Combined Annotation Dependent Depletion

Resource Type: data access protocol, data analysis software, data processing software, sequence analysis software, service resource, software application, software resource, web service

Defining Citation: [PMID:30371827](#), [PMID:24487276](#)

Keywords: Human genome, disease, prediction, injurious variant, single nucleotide variant, insertion variant, deletion variant, deleteriousness scoring

Funding: Berlin Institute of Health ;
Brotman Baty Institute for Precision Medicine ;
Charite University Medicine Berlin ;
German Research Foundation ;
Howard Hughes Medical Institute ;
NCI R01 CA197139;
NHGRI U54 HG006493

Availability: Restricted

Resource Name: Combined Annotation Dependent Depletion

Resource ID: SCR_018393

Record Creation Time: 20220129T080340+0000

Record Last Update: 20260903T235513+0000

Ratings and Alerts

No rating or validation information has been found for Combined Annotation Dependent Depletion.

No alerts have been found for Combined Annotation Dependent Depletion.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 784 mentions in open access literature.

Listed below are recent publications. The full list is available at [kravitz2](#) .

Stoiber S, et al. (2026) [18F]FDG PET/CT multiomics identifies Hedgehog-driven HPV-negative head and neck squamous cell carcinoma. *Molecular cancer*, 25(1).

Smolen C, et al. (2026) Protocol for identifying genetic modifiers of phenotypes in individuals with disease-associated variants. *STAR protocols*, 7(2), 104546.

Le DM, et al. (2026) Distinct Associations of GTF2I, TP53, and NOTCH1 Variants with Indolent and Aggressive Thymic Epithelial Tumors in Vietnamese Patients. *Genes*, 17(5).

Poulain F, et al. (2026) Clonal origin and early lineage divergence in adenosquamous carcinoma. *Cell reports*, 45(6), 117470.

Gan XY, et al. (2026) Compound heterozygous variants of the DMRTC2 gene are associated with non-obstructive azoospermia in a patient. *Asian journal of andrology*, 28(2), 219.

Yamazawa K, et al. (2026) Comprehensive evidence for the pathogenicity of the BRCA2 c.7847C>T (p.Ser2616Phe) variant specific to the Japanese population. *Journal of medical genetics*, 63(6), 393.

Watanabe T, et al. (2026) Whole exome sequencing in Japanese spinocerebellar ataxia identifies novel variants. *Journal of human genetics*, 71(1), 35.

Drackley A, et al. (2026) Recognition of a Critical Functional Domain and Improved PHOX2B Missense Variant Interpretation by Utilization of In Silico Prediction Tools. *Human mutation*, 2026, 8001520.

Shokouhian E, et al. (2026) Diagnostic Yield of Genome Sequencing in an Iranian Exome-Negative Autosomal-Recessive Intellectual Disability Cohort. *Human mutation*, 2026,

4623457.

Dai J, et al. (2026) Functional germline variants together with somatic mutations alter the integrity of cancer hallmark regulatory networks. *Genome medicine*, 18(1).

Capecki JA, et al. (2026) Evidence that disruption of Discoidin domain receptor 2 contributes to palate malformations through effects on the extracellular matrix. *Human molecular genetics*, 35(12).

Tan X, et al. (2026) Functional analysis of a novel splice site variant of RAB3GAP2 in a fetus with congenital cataracts. *BMC medical genomics*, 19(1).

Luo J, et al. (2026) Whole-exome sequencing identifies PRSS56 variants in Chinese patients with microphthalmia. *BMC medical genomics*, 19(1).

Sun D, et al. (2026) Analysis of familial exudative vitreoretinopathy (FEVR) cases in the UK 100 000 genomes project increases diagnostic rate and implicates heterozygous CTNND1 mutations in FEVR. *Journal of medical genetics*, 63(3), 187.

Samra S, et al. (2026) Human germline biallelic loss-of-function OSMR variants cause severe allergic disease. *Journal of human immunity*, 2(4), e20260067.

Jiang Z, et al. (2026) Clinical and Genetic Characteristics of a Chinese Occult Maculopathy Cohort. *Translational vision science & technology*, 15(3), 32.

Alimoradi E, et al. (2026) Identification of a Novel Missense Homozygous Variant in LINS1 in Two Distinct Iranian Families With Consanguineous Marriage. *Molecular genetics & genomic medicine*, 14(2), e70184.

Vettiati D, et al. (2026) Ultrarare Variants in DNA Damage Repair and Mitochondrial Genes in Pediatric Acute-Onset Neuropsychiatric Syndrome and Acute Behavioral Regression in Neurodevelopmental Disorders. *Developmental neuroscience*, 1.

Lange LM, et al. (2026) Rare but Relevant? Assessing Variants in Dystonia-Linked Genes in Parkinson's Disease. *Movement disorders : official journal of the Movement Disorder Society*, 41(1), 247.

Seidizadeh O, et al. (2026) Genetic determinants of clinical variability in type 2 von Willebrand disease: bridging genotype and phenotype. *Haematologica*, 111(2), 609.