Resource Summary Report

Generated by ASWG on Aug 3, 2026

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

Defining Citation: PMID:30371827, PMID:24487276

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

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Brotman Baty Institute for Precision Medicine ;
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Charite University Medicine Berlin

Availability: Restricted

Resource Name: Combined Annotation Dependent Depletion

Resource ID: SCR_018393

Record Creation Time: 20220129T080340+0000

Record Last Update: 20260803T040746+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 579 mentions in open access literature.

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

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

Ncir WB, et al. (2026) First LDLRAP1 and Recurrent LDLR Mutations in Tunisian Families With Familial Hypercholesterolemia. Journal of cellular and molecular medicine, 30(1), e70997.

Scarpato M, et al. (2025) A Novel Variant in TUBB4B Causes Progressive Cone-Rod Dystrophy and Early Onset Sensorineural Hearing Loss. Molecular genetics & genomic medicine, 13(2), e70068.

Yuan M, et al. (2025) Prevalence of IMPG1 and IMPG2 Mutations Leading to Retinitis Pigmentosa or Vitelliform Macular Dystrophy in a Cohort of Patients with Inherited Retinal Dystrophies. Genes, 16(1).

Gentiluomo M, et al. (2025) A genome-wide association study identifies eight loci associated with intraductal papillary mucinous neoplasm progression toward malignancy. Cancer, 131(1), e35678.

Laugwitz L, et al. (2025) EEFSEC deficiency: A selenopathy with early-onset neurodegeneration. American journal of human genetics, 112(1), 168.

Elasbali AM, et al. (2025) A structural genomics approach to investigate Dystrophin mutations and their impact on the molecular pathways of Duchenne muscular dystrophy. Frontiers in genetics, 16, 1517707.

Jih KY, et al. (2025) Early presentation of spastic paraparesis in individuals carrying PSEN1 mutations: a clinical and genetic analysis. Alzheimer's research & therapy, 17(1), 96.

Bu H, et al. (2025) Clinical and genetic characteristics of diseases caused by CFTR gene mutations in 15 Chinese children: a retrospective analysis. BMC pediatrics, 25(1), 701.

Hatzikotoulas K, et al. (2025) Translational genomics of osteoarthritis in 1,962,069 individuals. Nature, 641(8065), 1217.

Greene D, et al. (2025) Mutations in the small nuclear RNA gene RNU2-2 cause a severe neurodevelopmental disorder with prominent epilepsy. Nature genetics, 57(6), 1367.

Rowe L, et al. (2025) A proposed role for CDO1 in CNS development: Three children with rare missense variants and a neurological phenotype. HGG advances, 6(2), 100417.

Pillai J, et al. (2025) Predicting the impact of missense mutations on an unresolved protein's stability, structure, and function: A case study of Alzheimer's disease-associated TREM2 R47H variant. Computational and structural biotechnology journal, 27, 564.

Asagai Y, et al. (2025) Clinical characteristics of patients with SALL1-related disorder. Pediatric nephrology (Berlin, Germany), 40(11), 3407.

Kinnersley B, et al. (2025) Genomic landscape of diffuse glioma revealed by whole genome sequencing. Nature communications, 16(1), 4233.

Khan MR, et al. (2025) Identifying potential genetic biomarkers for sperm dysfunction through whole-genome sequencing. Scientific reports, 15(1), 36476.

Xu X, et al. (2025) A pedigree with homocystinuria caused by a novel homozygous CBS gene mutation: A case report. The Journal of international medical research, 53(10), 3000605251386166.

Singh S, et al. (2025) Biallelic variants in CCN2 underlie an autosomal recessive kyphomelic dysplasia. European journal of human genetics : EJHG, 33(1), 30.

Kubota T, et al. (2025) Hydrops fetalis due to loss of function of hNav1.4 channel via compound heterozygous variants. Journal of human genetics, 70(1), 3.

Huang Y, et al. (2025) FBN2 pathogenic mutation in congenital contractural arachnodactyly with severe skeletal manifestations. Molecular genetics and metabolism reports, 42, 101193.