Resource Summary Report

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American College of Medical Genetics and Genomics

RRID:SCR_005769
Type: Tool

Proper Citation

American College of Medical Genetics and Genomics (RRID:SCR_005769)

Resource Information

URL: http://www.acmg.net

Proper Citation: American College of Medical Genetics and Genomics (RRID:SCR_005769)

Description: An organization composed of biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals committed to the practice of medical genetics to Improve Health Through Medical Genetics. The American College of Medical Genetics and Genomics will: * Define and promote excellence in the practice of medical genetics and genomics in the integration of translational research into practice; * Promote and provide medical genetics and genomics education; * Increase access to medical genetics and genomics services and integrate them into patient care; * Advocate for and represent providers of medical genetics and genomics services and their patients; and * Maintain structure and integrity of ACMG and its value to members and the public.

Abbreviations: ACMG

Synonyms: American College of Medical Genetics, ACMG - Translating Genes Into Health, American College of Medical Genetics Genomics

Resource Type: organization portal, data or information resource, portal

Defining Citation: PMID:21311339

Keywords: genetics, genomics, medical, biochemical, clinical, cytogenetic, molecular, geneticist, genetic counselor, health care professional, medical genetics

Resource Name: American College of Medical Genetics and Genomics

Resource ID: SCR_005769

Alternate IDs: OMICS_01775, nlx_149234

Old URLs: http://www.acmg.net

Record Creation Time: 20220129T080232+0000

Record Last Update: 20260803T040421+0000

Ratings and Alerts

No rating or validation information has been found for American College of Medical Genetics and Genomics.

No alerts have been found for American College of Medical Genetics and Genomics.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 50 mentions in open access literature.

Listed below are recent publications. The full list is available at PRECISE-TBI .

Azuma N, et al. (2025) Sonic Hedgehog Determines Early Retinal Development and Adjusts Eyeball Architecture. International journal of molecular sciences, 26(2).

Rodriguez-Hernandez A, et al. (2025) Low Allele Frequency Variants Identified on Germline Multi-Gene Panel Testing for Cancer Predisposition Can Suggest the Presence of Constitutional Mosaicism. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(13), 2814.

Gatticchi L, et al. (2025) Functional analysis of amino acid substitutions within human AGT1 in a cell-based platform to support the diagnosis of primary hyperoxaluria type 1. The Journal of biological chemistry, 301(8), 110494.

Aref-Eshghi E, et al. (2025) De novo missense variants in CHTF18: The potential to expand the clinical spectrum of cohesinopathies. HGG advances, 6(4), 100485.

Mellone S, et al. (2025) Integrating genome and transcriptome analysis to decipher balanced structural variants in unsolved cases of neurodevelopmental disorders. Frontiers in genetics, 16, 1603513.

Görükmez O, et al. (2025) Clinical Exome Sequencing in Pediatric Patients. Cureus, 17(3), e80330.

Hwang J, et al. (2025) Structurally Oriented Classification of FOXA1 Alterations Identifies Prostate Cancers with Opposing Clinical Outcomes and Distinct Molecular and Immunologic Subtypes. Clinical cancer research : an official journal of the American

Association for Cancer Research, 31(5), 936.

Ullah N, et al. (2025) Identification of missense DMC1 variants in males with non-obstructive azoospermia. Journal of assisted reproduction and genetics, 42(11), 3723.

Han D, et al. (2024) Targeted next-generation sequencing reveals the genetic mechanism of Chinese Marfan syndrome cohort with ocular manifestation. Molecular genetics & genomic medicine, 12(7), e2482.

Mo L, et al. (2024) Metabolic cardiomyopathy associated with a compound heterozygous variant in NAD(P)HX dehydratase: a case report and literature review. Translational pediatrics, 13(12), 2292.

Özen S, et al. (2024) Molecular Genetic Diagnosis with Targeted Next Generation Sequencing in a Cohort of Turkish Osteogenesis Imperfecta Patients and their Genotype-phenotype Correlation. Journal of clinical research in pediatric endocrinology, 16(4), 431.

Fernández-Cancio M, et al. (2024) Clinical and molecular study of patients with thyroid dyshormogenesis and variants in the thyroglobulin gene. Frontiers in endocrinology, 15, 1367808.

Zhang X, et al. (2024) Clinical phenotype and genetic characteristics of SZT2 related diseases: A case report and literature review. Seizure, 114, 111.

Zhang X, et al. (2023) A novel heterozygous ATP1A2 pathogenic variant in a Chinese child with MELAS-like alternating hemiplegia. Molecular genetics & genomic medicine, 11(5), e2146.

Sun X, et al. (2023) Clinical features and underlying mechanisms of KAT6B disease in a Chinese boy. Molecular genetics & genomic medicine, 11(9), e2202.

Fu Y, et al. (2023) A novel homozygous missense variant in LRP4 causing Cenani-Lenz syndactyly syndrome and literature review. Molecular genetics & genomic medicine, 12(1), e2319.

Diaz-Lombana N, et al. (2023) Case report: Novel frameshift mutation in LAMA2 gene causing congenital muscular dystrophy type 1A. Frontiers in genetics, 14, 1158350.

Souche E, et al. (2022) Recommendations for whole genome sequencing in diagnostics for rare diseases. European journal of human genetics : EJHG, 30(9), 1017.

Shen Y, et al. (2022) Rare variant of TBL1XR1 in West syndrome: A case report. Molecular genetics & genomic medicine, 10(7), e1991.

Sodsai P, et al. (2022) TIGIT Monoallelic Nonsense Variant in Patient with Severe COVID-19 Infection, Thailand. Emerging infectious diseases, 28(11), 2350.