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: data or information resource, organization portal, 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: 20260912T200056+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 61 mentions in open access literature.

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

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).

Zhao J, et al. (2026) Novel compound heterozygous PKHD1 mutations in a Chinese ARPKD pedigree and analysis of genotype-phenotype correlations. Frontiers in medicine, 13, 1741795.

Dohi S, et al. (2026) Pontocerebellar hypoplasia type 9 with a novel combination of compound heterozygous variants in AMPD2. Human genome variation, 13(1).

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.

Ncube KT, et al. (2025) Genomic Tools for Medicinal Properties of Goat Milk for Cosmetic and Health Benefits: A Narrative Review. International journal of molecular sciences, 26(3).

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.

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.

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.

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.

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.

Abiib S, et al. (2025) Exploring the Genetic Causes of Nonsyndromic Retinal Dystrophies in Qatar. Genes, 16(12).

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.

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.

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.

Alsebeyi M, et al. (2024) Segregation of Trans Mutations in the CDH23 Gene in an Emirati Family with Sensorineural Hearing Loss. Genes, 15(11).

Baz-Redón N, et al. (2024) Patients with Thyroid Dyshormonogenesis and DUOX2 Variants: Molecular and Clinical Description and Genotype-Phenotype Correlation. International journal of molecular sciences, 25(15).

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