

Resource Summary Report

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Mutation Assessor

RRID:SCR_024502

Type: Tool

Proper Citation

Mutation Assessor (RRID:SCR_024502)

Resource Information

URL: <http://mutationassessor.org/r3/>

Proper Citation: Mutation Assessor (RRID:SCR_024502)

Description: Web server predicts functional impact of amino-acid substitutions in proteins, such as mutations discovered in cancer or missense polymorphisms. Functional impact is assessed based on evolutionary conservation of affected amino acid in protein homologs., THIS RESOURCE IS NO LONGER IN SERVICE. Documented on September 16,2025.

Resource Type: software resource, web application

Keywords: functional impact of amino-acid substitutions in proteins prediction,

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: Mutation Assessor

Resource ID: SCR_024502

Record Creation Time: 20231002T161336+0000

Record Last Update: 20260829T182828+0000

Ratings and Alerts

No rating or validation information has been found for Mutation Assessor.

No alerts have been found for Mutation Assessor.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 535 mentions in open access literature.

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

Vitanova K, et al. (2026) Gene Panel Analysis Reveals Overlapping Genetic Causes of Inherited Cataracts and Other Ocular Phenotypes in Bulgarian Patients. *Molecular genetics & genomic medicine*, 14(7), e70255.

Waleed Iqbal M, et al. (2026) Delving Into the Depths of AGTR2: In Silico Identification of Deleterious Nonsynonymous SNPs Associated With Cardiovascular Diseases. *Human mutation*, 2026, 9394808.

Luo L, et al. (2026) Comparative Diagnostic Assessment of Karyotyping, Microarray, and Whole Exome Sequencing in Genetically Associated Fetal Growth Restriction. *Diagnostics (Basel, Switzerland)*, 16(2).

Iqbal MW, et al. (2026) Exploring Deleterious Nonsynonymous SNPs in the ACADM Gene: Insights Into Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCADD) via In Silico Analysis. *Genetics research*, 2026, 6682668.

Viakhireva I, et al. (2026) A Rare Clinical Presentation of Variegate Porphyrria. *Molecular genetics & genomic medicine*, 14(6), e70243.

Song SN, et al. (2026) Endothelial NOX4-driven oxidative stress inhibition reverses HtrA1 deficiency-induced blood-brain barrier disruption and cognitive impairment. *International journal of molecular medicine*, 58(1).

Marohl T, et al. (2025) PCSK5M452I is a recessive hypomorph exclusive to MCF10DCIS.com cells. *bioRxiv : the preprint server for biology*.

Marohl T, et al. (2025) PCSK5M452I is a recessive hypomorph exclusive to MCF10DCIS.com cells. *Molecular cancer research : MCR*.

Arcos-Burgos M, et al. (2025) Neurodevelopment Genes Encoding Olduvai Domains Link Myalgic Encephalomyelitis to Neuropsychiatric Disorders. *Diagnostics (Basel, Switzerland)*, 15(12).

Wu T, et al. (2025) DNA damage and repair (DDR) gene mutation profiles in driver gene wild-type advanced non-small cell lung cancer and the predictive role of response to platinum-based chemotherapy. *Translational lung cancer research*, 14(4), 1089.

Florio CA, et al. (2025) POLR3A rare variants in a patient with intellectual disability, ataxic gait and cortical malformations: a case-report. *Italian journal of pediatrics*, 51(1), 191.

Heimer G, et al. (2025) Biallelic PIGM Coding Variant Causes Intractable Epilepsy and Intellectual Disability Without Thrombotic Events. *Clinical genetics*, 107(2), 179.

Yang H, et al. (2025) Genetic analysis of congenital unilateral renal agenesis in children based on next-generation sequencing. *Pediatric research*, 97(1), 273.

Huang Y, et al. (2025) RMVar 2.0: an updated database of functional variants in RNA modifications. *Nucleic acids research*, 53(D1), D275.

Ettaki I, et al. (2025) Missense variants weakening a SOX9 phosphodegron linked to odontogenesis defects, scoliosis, and other skeletal features. *HGG advances*, 6(2), 100404.

Pauzi FA, et al. (2025) Histopathological spectrum of common aldosterone-driver gene mutations in aldosterone-producing adenomas. *Frontiers in medicine*, 12, 1569619.

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.

Iqbal MW, et al. (2025) Exploring deleterious non-synonymous SNPs in FUT2 gene, and implications for norovirus susceptibility and gut microbiota composition. *Scientific reports*, 15(1), 10395.

Cheng Y, et al. (2025) Clinical and genetic characteristics of PLA2G6-related parkinsonism in Southwest China and a comprehensive literature review. *Journal of medical genetics*, 62(8), 508.

Li X, et al. (2025) Clinical and Molecular Landscape of GLRA2 in X-Linked Early-Onset High Myopia. *Investigative ophthalmology & visual science*, 66(4), 30.