

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: web application, software resource

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: 20260805T044532+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 493 mentions in open access literature.

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

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.

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.

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

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.

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.

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.

Rodriguez-Salamanca J, et al. (2025) Integrating next-generation sequencing and artificial intelligence for the identification and validation of pathogenic variants in colorectal cancer. Frontiers in oncology, 15, 1568205.

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.

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.

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

Chen X, et al. (2025) De Novo Missense Variant c.170 C > A of ELANE in a Chinese Infant with Congenital Neutropenia: Case Report and Literature Review. *Journal of clinical immunology*, 45(1), 113.

Adda Neggaz L, et al. (2025) Computational prediction of deleterious nonsynonymous SNPs in the CTNS gene: implications for cystinosis. *BMC genomic data*, 26(1), 35.

Murase A, et al. (2025) Variants of NLRP genes encoding subcortical maternal complex components are linked to biparental placental mesenchymal dysplasia. *Human genomics*, 19(1), 94.

Pettinato F, et al. (2025) Acute neurological regression following fever as presenting sign of pontocerebellar hypoplasia type 2D (SEPSECS mutation). *Biomedical reports*, 22(4), 67.

Huang X, et al. (2025) Mutation spectra and genotype?phenotype analysis of congenital hypothyroidism in a neonatal population. *Biomedical reports*, 22(2), 30.

Försti A, et al. (2025) Search for germline gene variants in colorectal cancer families presenting with multiple primary colorectal cancers. *International journal of cancer*, 156(7), 1393.