Resource Summary Report

Generated by NIF on Aug 26, 2026

MutationAssessor

RRID:SCR_005762
Type: Tool

Proper Citation

MutationAssessor (RRID:SCR_005762)

Resource Information

URL: http://mutationassessor.org/

Proper Citation: MutationAssessor (RRID:SCR_005762)

Description: A web server that predicts the functional impact of amino-acid substitutions in proteins, such as mutations discovered in cancer or nonsynonymous polymorphisms. The functional impact is assessed based on evolutionary conservation of the affected amino acid in protein homologs. The method has been validated on a large set (51k) of disease associated (OMIM) and polymorphic variants., THIS RESOURCE IS NO LONGER IN SERVICE. Documented on September 16,2025.

Abbreviations: mutationassessor.org

Synonyms: MutationAssessor - functional impact of protein mutations, MutationAssessor - functional impact of mutations, mutationassessor.org - functional impact of protein mutations

Resource Type: analysis service resource, data analysis service, production service resource, service resource

Defining Citation: PMID:21727090

Keywords: cancer, protein, mutation, function, amino-acid, substitution

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: MutationAssessor

Resource ID: SCR_005762

Alternate IDs: OMICS_00134, nlx_149228

Record Creation Time: 20220129T080232+0000

Record Last Update: 20260821T193747+0000

Ratings and Alerts

No rating or validation information has been found for MutationAssessor.

No alerts have been found for MutationAssessor.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 693 mentions in open access literature.

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

Jiang Q, et al. (2026) Beyond the genome: clinical challenges in diagnosing LONP1-related mitochondrial disorders. Frontiers in cell and developmental biology, 14, 1779332.

Chen ZX, et al. (2026) A case report of congenital sideroblastic anemia caused by a novel ALAS2 mutation in conjunction with thalassemia. Annals of hematology, 105(5).

Kovalskaia VA, et al. (2026) Mutational spectrum of EDA, EDAR, EDARADD, and WNT10A genes in the largest cohort of Russian patients with hypohidrotic ectodermal dysplasia. Orphanet journal of rare diseases, 21(1), 45.

Huang WH, et al. (2026) Identification of gene variants in 30 patients from southeastern China with severe hypospadias by whole-exome sequencing. Asian journal of andrology, 28(3), 276.

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

Johansson I, et al. (2025) Proof of principle concept for the analysis and functional prediction of rare genetic variants in the CYP2C19 and CYP2D6 genes. Human genomics, 19(1), 62.

Imangaliyeva A, et al. (2025) Genetic Insights into Severe Obesity: A Case Study of MC4R Variant Identification and Clinical Implications. Genes, 16(5).

Katsonis P, et al. (2025) Meta-EA: a gene-specific combination of available computational tools for predicting missense variant effects. Nature communications, 16(1), 159.

Huang X, et al. (2025) Identification of novel compound heterozygous variants in the PEX10 gene in a Han-Chinese family with PEX10-related peroxisome biogenesis disorders. PloS one, 20(4), e0322137.

Heo JY, et al. (2025) PRP: pathogenic risk prediction for rare nonsynonymous single nucleotide variants. Human genetics, 144(6), 679.

Hou Z, et al. (2025) An isolated 17,20-lyase deficiency patient achieved a successful live birth after in vitro fertilization: a case report and narrative review. Journal of assisted reproduction and genetics, 42(2), 679.

Xie L, et al. (2025) A new heterozygous TYK2 gene mutation: Case report and review of the literature. International journal of immunopathology and pharmacology, 39, 3946320251351138.

Ling D, et al. (2025) Compound Heterozygous Loss-of-Function Variants in CCM2L in a Fetus With Tetralogy of Fallot. Molecular genetics & genomic medicine, 13(6), e70117.

Li N, et al. (2025) Novel SSR4 gene splice variant leads to congenital disorder of glycosylation, type Iy. Frontiers in pediatrics, 13, 1651524.

Yang H, et al. (2025) A multimodal framework for comprehensive driver variant prediction in cancer. Communications medicine, 5(1), 499.

Antolí A, et al. (2025) From Rare to Common: Genetic Insights into TLR7 Variants in a Multicentric Spanish Study on COVID-19 Severity. Journal of clinical immunology, 45(1), 100.

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

Jadidi M, et al. (2025) Identification of a rare variant in TNNT3 responsible for familial dilated cardiomyopathy through whole-exome sequencing and in silico analysis. European journal of medical research, 30(1), 424.

Ruiz-Alías G, et al. (2025) Missense variants pathogenicity annotation from homologous proteins. Bioinformatics (Oxford, England), 41(5).

Park HS, et al. (2025) Clinical Exome-Based Redefinition and Reclassification of Retinitis Pigmentosa. Journal of Korean medical science, 40(16), e54.