

Resource Summary Report

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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: data analysis service, service resource, production service resource, analysis 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: 20260801T042603+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 669 mentions in open access literature.

Listed below are recent publications. The full list is available at [kravitz2](#) .

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.

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.

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

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.

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.

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

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.

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

- 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.
- 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.
- Ruiz-Alías G, et al. (2025) Missense variants pathogenicity annotation from homologous proteins. *Bioinformatics (Oxford, England)*, 41(5).
- Yang H, et al. (2025) A multimodal framework for comprehensive driver variant prediction in cancer. *Communications medicine*, 5(1), 499.
- Sarker DK, et al. (2025) Exploring the impact of deleterious missense nonsynonymous single nucleotide polymorphisms in the DRD4 gene using computational approaches. *Scientific reports*, 15(1), 3150.
- Abdelazim AA, et al. (2025) In-silico screening and analysis of missense SNPs in human CYP3A4/5 affecting drug-enzyme interactions of FDA-approved COVID-19 antiviral drugs. *Scientific reports*, 15(1), 2153.
- Li J, et al. (2025) Germline alteration analysis reveals EPHB4R91H mutation as a key player in multiple primary lung tumors. *Carcinogenesis*, 46(1).
- Bayanova M, et al. (2025) Genetic landscape and phenotypic spectrum of osteogenesis imperfecta in the Kazakhstani pediatric population. *Scientific reports*, 15(1), 11223.
- Bonetti E, et al. (2025) RENOVO-NF1 accurately predicts NF1 missense variant pathogenicity. *Human genomics*, 19(1), 106.
- Zin OA, et al. (2025) Genotype-Phenotype Correlations of Nance-Horan Syndrome in Male and Female Carriers of a Novel Variant. *Genes*, 16(1).