

Resource Summary Report

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MutationTaster

RRID:SCR_010777

Type: Tool

Proper Citation

MutationTaster (RRID:SCR_010777)

Resource Information

URL: <http://www.mutationtaster.org/>

Proper Citation: MutationTaster (RRID:SCR_010777)

Description: Evaluates disease-causing potential of sequence alterations.

Abbreviations: MutationTaster

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

Defining Citation: [PMID:20676075](#)

Keywords: bio.tools

Availability: Acknowledgement requested

Resource Name: MutationTaster

Resource ID: SCR_010777

Alternate IDs: biotools:mutation_taster, OMICS_00153

Alternate URLs: https://bio.tools/mutation_taster

Record Creation Time: 20220129T080300+0000

Record Last Update: 20260728T043349+0000

Ratings and Alerts

No rating or validation information has been found for MutationTaster.

No alerts have been found for MutationTaster.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 4180 mentions in open access literature.

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

Xiangwen W, et al. (2026) A De Novo Splicing Mutation of SRP72 in Bone Marrow Failure Syndrome Type 1: Case Report and Review of the Literature. *Molecular genetics & genomic medicine*, 14(1), e70168.

Guo W, et al. (2026) Pathogenicity of novel GLA gene missense mutations in Fabry disease and the therapeutic impact of migalastat. *Journal of advanced research*, 79, 549.

Watanabe T, et al. (2026) Whole exome sequencing in Japanese spinocerebellar ataxia identifies novel variants. *Journal of human genetics*, 71(1), 35.

Ncir WB, et al. (2026) First LDLRAP1 and Recurrent LDLR Mutations in Tunisian Families With Familial Hypercholesterolemia. *Journal of cellular and molecular medicine*, 30(1), e70997.

Fiorica PN, et al. (2025) Germline Pathogenic DROSHA Variants Are Linked to Pineoblastoma and Wilms Tumor Predisposition. *Clinical cancer research : an official journal of the American Association for Cancer Research*, 31(8), 1491.

Gaikwad AS, et al. (2025) Functional and clinical insights into nuclear receptor variants for advancing precision diagnostics in male infertility. *EBioMedicine*, 119, 105899.

Tanaka Y, et al. (2025) Phenotype and genotype of autosomal dominant tubulointerstitial kidney disease in a Japanese cohort. *Clinical and experimental nephrology*, 29(6), 788.

Hong Z, et al. (2025) Unveiling the Pathogenic Role of Novel CPLANE1 Compound Heterozygous Variants in Joubert Syndrome: Insights Into mRNA Stability and NMD Pathway. *Journal of cellular and molecular medicine*, 29(5), e70484.

Wu Y, et al. (2025) Myocardial dysfunction caused by MyBPC3 P459fs mutation in hypertrophic cardiomyopathy: evidence from multi-omics approaches and super-resolution imaging. *Frontiers in cardiovascular medicine*, 12, 1529921.

Wen X, et al. (2025) Clinical phenotype and genetic analysis of patients with severe oligoasthenospermia carrying heterozygous SOHLH1 c.346-1G>A mutation. *Frontiers in genetics*, 16, 1531697.

Garc??a-Tenorio EM, et al. (2025) Novel CRISPR-Cas9 iPSC knockouts for PCCA and PCCB genes: advancing propionic acidemia research. *Human cell*, 38(3), 64.

Chang SH, et al. (2025) Rare SRRM2 mutation in neurodevelopmental disorders involving hyperphagia triggering severe obesity and other complication. *Frontiers in medicine*, 12,

1492851.

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.

Albitar L, et al. (2025) Detection of exon2-MED12 mutations in uterine leiomyomas from Syrian patients. *Scientific reports*, 15(1), 452.

Fang W, et al. (2025) Genetic analysis of a pedigree with hereditary coagulation factor XII deficiency. *Annals of hematology*, 104(2), 1281.

Liu Z, et al. (2025) Novel pathogenic mtDNA variants in Chinese children with neurological mitochondrial disorders. *Annals of clinical and translational neurology*, 12(3), 586.

Zhang S, et al. (2025) Exploratory analysis of a Novel RACK1 mutation and its potential role in epileptic seizures via Microglia activation. *Journal of neuroinflammation*, 22(1), 27.

El Fissi H, et al. (2025) Mucopolysaccharidoses types I and IIIA: Diagnosis and identification of novel polymorphisms associated with common mutations in Moroccan patients. *Molecular genetics and metabolism reports*, 42, 101186.

Wang S, et al. (2025) Effects of asfotase alfa on fracture healing of adult patient with hypophosphatasia and literature review. *Orphanet journal of rare diseases*, 20(1), 162.

Wu J, et al. (2025) Expanding the Clinical and Genetic Spectrum of Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency: Insights From Two Unrelated Chinese Families. *Molecular genetics & genomic medicine*, 13(4), e70097.