Resource Summary Report

Generated by PRECISE-TBI on Jul 31, 2026

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: service resource, production service resource, analysis service resource, data analysis service

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: 20260731T042836+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 PRECISE-TBI .

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.