

Resource Summary Report

Generated by T1D on Aug 1, 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: data analysis service, service resource, production service resource, analysis 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: 20260801T042710+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 [T1D](#) .

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.

Peng SQ, et al. (2025) A gain-of-function mutation in ATP6V0A4 drives primary distal renal tubular alkalosis with enhanced V-ATPase activity. *The Journal of clinical investigation*, 135(13).

Yao M, et al. (2025) A case report of SPONASTRIME dysplasia with novel TONSL mutation: genetic analysis, clinical manifestations, and the effect of growth hormone treatment. *Human molecular genetics*, 34(19), 1665.

Baris S, et al. (2025) PGAP1-Related Encephalopathy in an Infant With Neurodevelopmental Delay: Novel Variant and Review of Literature. *International journal of developmental neuroscience : the official journal of the International Society for Developmental Neuroscience*, 85(5), e70044.

Qin Z, et al. (2025) Structural and functional assessment of TBX20 gene variants in pediatric ventricular septal defect. *Hereditas*, 162(1), 149.

Diakité M, et al. (2025) Unusual catecholaminergic polymorphic ventricular tachycardia and bradycardia caused by a novel triadin variant in 2 siblings from a Malian family. *Medicine*, 104(31), e43596.

Yang J, et al. (2025) Manic Fringe promotes endothelial-to-mesenchymal transition mediated by the Notch signalling pathway during heart valve development. *Journal of molecular medicine (Berlin, Germany)*, 103(1), 51.

Moresco G, et al. (2025) A novel frameshift TBX4 variant in a family with ischio-coxo-podo-patellar syndrome and variable severity. *Genes & genomics*, 47(3), 341.

Heimer G, et al. (2025) Biallelic PIGM Coding Variant Causes Intractable Epilepsy and Intellectual Disability Without Thrombotic Events. *Clinical genetics*, 107(2), 179.

Zhou Q, et al. (2025) Molecular and Clinical Profiles of Pediatric Monogenic Diabetes Subtypes: Comprehensive Genetic Analysis of 138 Patients. *The Journal of clinical endocrinology and metabolism*, 110(8), 2314.

Bayam E, et al. (2025) Bi-allelic variants in WDR47 cause a complex neurodevelopmental syndrome. *EMBO molecular medicine*, 17(1), 129.

Smith TB, et al. (2025) Bi-allelic variants in DAP3 result in reduced assembly of the mitoribosomal small subunit with altered apoptosis and a Perrault-syndrome-spectrum phenotype. *American journal of human genetics*, 112(1), 59.

Cif L, et al. (2025) KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. *ArXiv*.

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.

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.